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Granular tumors of the central nervous system: A case series

Janese Trimaldi, Nicole D Riddle, Jeremy W Bowers, Harry R van Loveren, Kondi Wong

ABSTRACT

Introduction: Granular cell tumors of the central nervous system are rare tumors. To date, eight cases arising from cranial nerves have been reported. Granular cell tumors have also been found arising from the neurohypophysis and its stalk. Due to their rarity and histological similarity to other central nervous system (CNS) tumors with a granular appearance, they often pose a diagnostic conundrum. The differential diagnosis is surprisingly diverse and includes granular cell astrocytoma, infundibular granular cell tumor, spindle cell oncocytoma of the adenohypophysis, granular and oncocytic variants of pituitary adenoma, meningioma, pituicytoma and intrasellar schwannoma. Distinguishing between the CNS tumors with granular features is important because some tumors have an increased recurrence risk or a poor prognosis. Case Report: To highlight the histological features of granular lesions of the central nervous system, including the immunohistochemical profile and electron microscopic depiction, we review two cases each with a similar granular histology and a different final diagnosis. Conclusion: Thorough online literature search revealed several cases of granular cell lesions of the CNS, however, oftentimes the diagnosis is difficult to come by and the differential is long. Conclusion: Granular cell tumor and its variants, though uncommon, must be included in the differential diagnosis of CNS lesions.

Keywords: Granular, Tumors, Central nervous system (CNS)


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INTRODUCTION

Granular cell lesions of the central nervous system (CNS) are rather uncommon and may pose a diagnostic challenge. Granular histology may be present as the main characteristic of a neoplasm or as a variation in a number of benign and malignant lesions. The differential diagnosis must always include a true granular cell tumor when granular morphology is identified, especially when it encompasses a majority of the lesion. The differential diagnosis of a CNS granular cell tumor is widely varied and includes infundibular granular cell tumor, granular/oneocytic variants of pituitary adenoma, meningioma, pituicytoma and intrasellar schwannoma, as well as spindle cell
oncocytoma of the adenohypophysis, which may be prone to recur and grow despite adjuvant therapy, and granular cell astrocytoma, which has a poor prognosis.

CASE REPORT

Case 1: A 37-year-old male presented with a history of depression, fatigue, and decreased libido for two years. Magnetic resonance imaging (MRI) was performed revealing a left-sided pituitary mass that was enlarging the sella with suprasellar extension. Post-contrast images demonstrated an intensely enhancing mass in the right middle cranial fossa that was hypointense on T2-weighted images and isointense to adjacent gray matter on T1-weighted images (Figure 1). It was centered in Meckel’s cave with involvement of the cisternal segment of the trigeminal nerve. There was distal extension of the mass along the inferior alveolar branch of V3 into the mandibular foramen. The right side of the pituitary gland appeared to be uninvolved. Surgical removal was performed through a transnasal endoscopic approach and yielded a 1.4x0.4x0.1 cm tumor. Frozen section analysis revealed a granular epithelial cell neoplasm. The differential diagnosis at that time included: pituitary adenoma, xanthoma, infundibular granular cell tumor, langerhans cell histiocytosis, hemangioblastoma and granular cell astrocytoma.

Routine histological examination with hematoxylin and eosin (H&E) stained sections revealed diffuse, densely granulated, eosinophilic cells (Figure 2). Special staining for PAS was also positive (Figure 3). Immunohistochemical studies showed the tumor cells to be weakly positive for prolactin and strongly synaptophysin positive. The tumor cells were non-reactive for CD68, inhibin, S100, chromogranin; as well as pituitary markers such as ACTH, FSH, LH, GH, and TSH. Pancytokeratin and GFAP were also negative. Complete disruption of the native lobular reticular architecture was evident on reticulin stain, characteristic of a pituitary adenoma. A diagnosis of pituitary adenoma with granular cell morphology, and evidence of prolactin expression was given.

Case 2: A 54-year-old woman with a history of Bell’s palsy one year prior with recurrence nine months later, presented with numbness primarily in the third division of the right trigeminal nerve.

MRI scan showed a tumor homogeneously enhancing in the gasserian ganglion with an extension into the foramen ovale (Figure 4). A right temporal craniotomy and zygomatic osteotomy was performed, and the foramen ovale was opened for resection of a 6.1x4.3x1.2 cm tan-brown mass with multi-nodular areas. Frozen section analysis suggested a possible ganglieneuroma. Following histological evaluation a
diagnosis of granular cell tumor of the right trigeminal ganglion was rendered. Permanent section analysis revealed peripheral nerve and entrapped ganglion cells (trigeminal ganglion) infiltrated by round, spindled and epithelioid cells with abundant, granular, PAS-positive cytoplasm consistent with granular cell tumor (Figure 5). The tumor cells were strongly positive for CD 68 with abundant PAS positive cytoplasmic granules and strong reactivity to S100 (Figures 6 and 7). They were non-reactive for: synaptophysin, smooth muscle actin, muscle specific actin, melan-A, myogenin, GFAP, smooth muscle myosin heavy, and desmin. Ki67 demonstrated moderate proliferative activity as well as moderate amounts of reticulin collagen and neuron specific enolase (not shown). Electron microscopy demonstrated tumor cell cytoplasm filled and expanded with dark, granular lysosomes, characteristic of granular cell tumor (Figure 8).

DISCUSSION

A number of granular or similarly oncocytic tumors have remarkably similar histomorphology on initial staining with hematoxylin and eosin. Differing intracranial locations may be helpful at times for diagnosis, but at other times they may be misleading. Tumors containing granular cells can present in two forms: (i) as a “pure” form composed entirely of granular cells (“granular cell tumor”), (ii) or as a focal change that occurs in a neoplasm of a recognizable cell type [1]. In the latter form, one theory considers granular cell change to be a degenerative phenomenon
[2]. The hypothesis is that the granular cell phenotype seems to be not specific of a certain tumor type, but rather a peculiar change characterized by an increase in intracellular lysosomes.

Reported CNS tumors with granular cell or oncocytic change include: astrocytoma, medulloblastoma, ganglioglioma, glioblastoma, meningioma, schwannoma, ependymoma, oligodendroglioma, neurofibroma, anterior and posterior pituitary [2–7]. Spindle cell oncocytoma, granular cell astrocytoma, and pituitary are distinct entities and possibly the infundibular granular cell tumor as well. Morphology on hematoxylin and eosin stain and special stains alongside tumor immunophenotype are a valuable aide in pathologic evaluation and distinguishing the different tumor types.

There are distinguishing characteristics of different tumor types. Granular cell astrocytomas are GFAP reactive in more than 95% cases [19] and have a poor prognosis. Spindle cell oncocytomas of the adenohypophysis have S100 reactivity, but are negative for neuroglial markers and CD68 because they are filled with mitochondria, not phagolysosomes. Spindle cell oncocytomas are reactive for TTF-1 (thyroid transcription factor), but negative for thyroglobulin. Meningiomas are generally EMA (epithelial membrane antigen) reactive, at least focally, with a lobular reticulin pattern and meningeal pattern on ultrastructure. Granular/oncocytic pituitary adenomas lose the native lobular reticulin architecture of the pituitary gland and are reactive for neuroendocrine markers. Intrasellar schwannoma may have granular cytoplasm, but can be distinguished by the dense pericellular basal lamina on reticulin staining, diffuse S100 staining and non reactivity for other neuroglial markers. Electron microscopy shows a profuse basal laminar or basement membrane pattern with long spaced collagen or “Luse bodies”[20].

Conventional granular cell tumors are similar and differ by primary location and possibly cell of origin.

Eight cases of cranial nerve granular cell tumors have been reported in literature arising from the following nerves: vagus, oculomotor, abducens, optic, facial; and an additional three arising from the trigeminal nerve [8–15]. Of the four arising from the trigeminal ganglion, two were reported as confined to Meckel’s cave, while one reported the tumor originating from the brain stem and extending into Meckel’s cave [13–15]. All patients experienced symptoms of trigeminal neuralgia. All masses were composed of clusters or nests of cells, most polyhedral, with round nuclei, separated by connective tissue ranging from dense strands of collagen to delicate reticulin fibers. Tumor in our case was non-reactive for synaptophysin and GFAP, helping to rule out this entity in our differential diagnoses.

Granular cell change occurs as a cell accumulates lysosomes and phagosomes. There are numerous types of cytoplasmic accumulations that result in distinctive cell morphologies which correlate with the type of material that is being increased. Often a certain type of accumulated material favor a certain cell type. For example, clear cell change is usually due to glycogen accumulation, which is easily seen with a PAS stain with diastase digestion. Epithelial cells are the primary type of cell involved. Cytoplasmic vacuolization that leads to indentation of the nuclear membrane is due to lipid containing vacuoles, and is usually seen in sebaceous gland cells, adrenal cortex fasciculata cells, lipoblasts or adipose tissue neoplasms. In addition to phagosomes and lysosomes, other types of material may be secretory granules or mast cell granules can lead to a granular appearance of the cytoplasm. In granular cell tumors, toluidine blue semithin sections of epon embedded tissue show cytoplasm filled with densely staining, but variegated vacuolar material that prove to be degenerated phagolysosomes on electron microscopy [16].

Granular cell tumors may arise in the neurohypophysis or infundibulum of the pituitary gland. They have also been referred to as choristomas (microscopic nodules in the infundibulum) or myoblastomas. They show a preference for the stalk, are usually suprasellar and composed of nests of large, eosinophilic cells with abundant granular cytoplasm due to high lysosomal content. Most commonly, they are an incidental finding, usually at autopsy, composed of small, round, eosinophilic, infundibular nodules. Their origin is presumed to be the pitucyte—a specialized type of glial cell or modified astrocyte of the neurohypophysis [1]. Electron microscopy shows pituicytes to possess variable amounts of electron dense lysosomes, identical to those found in granular cell tumors. Hence, a newly described tumor, the pitucityoma, may possibly be related. Isolated or clusters of granular cells are a normal part of the histology of the neurohypophysis. It is rare for these microscopic nodules to enlarge enough to become symptomatic. When they do, they are usually suprasellar, discrete, and show intense heterogenous or homogenous enhancement with contrast. Common symptoms, including visual acuity deficits, or rarely, endocrine deficiency are usually the result of stalk compression. They show a predilection for adult females. Grossly, the tumor is lobulated, soft but rubbery (firmer than an adenoma) and very vascular, with a gray-yellow cut surface that usually lacks necrosis, cystic degeneration or invasion of surrounding structures. The tumor cells are polygonal, with small nuclei containing evenly dispersed chromatin and inconspicuous nucleoli, low mitosis/proliferation, and can grow in sheets, nodules, or in a spindled/fascicular pattern. The abundant cytoplasm contains granules that are diastase resistant with PAS staining. “Atypical” tumors show increased mitosis (up to 5/10 HPF, and Ki67 index of 7%), nuclear pleomorphism, prominent nucleoli and multi-nucleation. The significance of this difference is unknown. Giant cell tumors (GCTs) are positive for: CD68, S100, alpha-1-antitrypsin, alpha-1-antichymotrypsin, and cathepsin B; and negative for neurofilament proteins, cytokeratins, chromogranin A, synaptophysin, desmin, smooth muscle actin and pituitary hormones. Though most are negative for glial
neural filament protein (GFAP), some variable reactivity has been reported. A presumed lineage from pituicytes may explain the conflicting results of IHC since there are different types of pituicytes. Electron microscopy demonstrates phagolysosomes with unevenly distributed electron dense material and membranous debris. Neurosecretory granules are absent. Most are slow growing, non-invasive and clinically benign [17].

Granular cell astrocytomas (GCA) are infiltrative neoplasms and ill-defined. They are considered an aggressive variant (WHO grade 3) despite the fact that they are cytologically bland with few mitosis. The majority are found in cerebral hemispheres. Since they are associated with high grade astrocytomas, they display ring enhancement on imaging. Their cytoplasm, like other granular cell neoplasms, contains PAS positive, diastase resistant granules that often create an eccentric nucleus. Some tumor cells display a clear central cytoplasmic area and the granules accumulate at the periphery beneath the cell membrane. Granular cell astrocytomas are positive for CD68, EMA, and S100 in the majority of cases. GFAP is positive in over 95% of tumors (18, 19, 21). The malignant nuclei are larger and courser than normal astrocytes, but are not as hyperchromatic or pleomorphic as in glioblastoma (WHO grade 4). The granular cell component can either be mixed with an otherwise typical diffuse astrocytoma or be the exclusive cell type in a lesion. Diagnosis may be particularly difficult in the diffuse exclusive cases and they are often mistaken for non-neoplastic processes such as infarcts or demyelinating diseases or GCTs of the infundibulum. Making this distinction, however, is paramount, as GCA’s have a poor prognosis, whereas, granular cell tumor of the neurohypophysis is a benign, slowly progressive tumor without an invasive growth pattern.

CONCLUSION

Granular cell lesions of the central nervous system are very rare. However, granular histology may be seen in a vast array of benign and malignant lesions. Histomorphology on hematoxylin and eosin stain and special stains with tumor immunophenotype can be a helpful aid in distinguishing the various tumor types. The differential of a granular cell tumor must always be included when granular cell morphology is present. Also, a granular cell astrocytoma must be ruled out due to prognostic implications.

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Janese Trimaldi – Conception and design, Acquisition of data, Drafting the article, Final approval of the version to be published
Nicole D Riddle – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Final approval of the version to be published

Jeremy Bowers – Conception and design, Acquisition of data, Critical revision of the article, Final approval of the version to be published
Harry Van Loveren – Acquisition of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Kondi Wong – Conception and design, Analysis and interpretation of data, Critical revision of the article, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

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REFERENCES

Magnetic resonance imaging confirmed clinical diagnosis of amyoplasia in two infants with arthrogryposis multiplex congenita

Ariam Diaz, Dominic Sia, Valerie May G Sia, Evelyn Erickson, Sergey Prokhorov, Menachem Gold

ABSTRACT

Introduction: We present two cases of arthrogryposis multiplex congenita (AMC) with involvement of the lower extremities. In both cases amyoplasia was confirmed by a magnetic resonance imaging (MRI). The degree of amyoplasia correlated with the severity of arthrogryposis and determined the child's prognosis. Case Series: Case 1 was a 16-month-old male child with prenatally diagnosed Klinefelter syndrome was born at 36 weeks gestation. Brain MRI was reported as normal. Joint rigidity was detected in upper and lower extremities. Amyoplasia was suspected at nine months of age since the lower limb muscles were hardly palpable. Case 2 was a 5 1/2-month-old female child and the first child of non-consanguinous parents was noticed to have rigid right calcaneovalgus and left equinovarus feet deformities as well as knee rigidity with limitation of knee extension. Bilateral hip displacement was also diagnosed. Absence of muscles on thigh palpation prompted MRI study. Conclusion: Although amyoplasia is the most common type of arthrogryposis multiplex congenita, muscle underdevelopment in these patients remains puzzling for pediatric practitioners. Amyoplasia congenita is usually symmetrical and involves either all extremities or selectively only the lower or upper extremities. Absence of muscle groups on MRI confirms diagnosis of amyoplasia. Early recognition of amyoplasia in children with arthrogryposis multiplex congenita can help in tailoring their treatment and prognosis.

Keywords: Amyoplasia, Arthrogryposis multiplex congenita (AMC)

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INTRODUCTION

Arthrogriposis multiplex congenita (AMC) affects about 1/3000 birth in North America [1]. Amyoplasia (A=no, myo=muscle and plasia=growth) is the most common type of arthrogryposis seen clinically [2]. We present two cases of arthrogryposis multiplex congenita with prominent involvement of the lower extremities. In both cases amyoplasia was confirmed by magnetic resonance imaging (MRI) of the thighs. The degree of amyoplasia correlated with the severity of arthrogryposis and determined the child’s prognosis. MRI may be helpful in the differentiation between amyoplasia and other congenital myopathies and muscular dystrophies [3].
CASE SERIES

Case 1: The patient was a 16-month-old male baby, prenatally diagnosed with Klinefelter syndrome born at 36 weeks gestation from a non-consanguineous marriage. The delivery was by emergency cesarean section due to fetal heart rate deceleration. Apgar score was 1, 2 and 5 at 1, 5 and 10 minutes. During the first two days of life the baby developed seizures. Brain MRI was reported as normal. Joint contractures were recognized on the fourth day of the baby’s life. Amyoplasia was suspected at nine months of age since the lower limb muscles were barely palpable. MRI of the thigh revealed paucity of muscles (Figures 1 and 2). On examination, the baby presented with microcephaly (head circumference 44 cm, <2%), bilateral epicanthus and clinodactyly of the fifth fingers. The child was able to hold sitting position, however, he was not able to sit up by himself. There was stiffness in the wrists with limitation of wrist extension. Hand grasp was weak. The child was unable to hold a spoon. Hip abduction was limited to 30°. Knee flexion was limited to 90°. There was also equinovarus feet deformity. The child moved the upper extremities well, but only slightly raised the extended lower extremities and minimally flexed them at the knees. He supported his weight on his legs when in standing position with support. In the lower extremities only hip adductors were slightly palpable. Gluteus muscle contraction was evident. Biceps, brachioradial and knee reflexes were normal. Plantar response was down-going. There was no deficiencies in sensation to the touch and pinprick.

Case 2: A 5½-month-old female was the first child of non-consanguineous parents was born after full term uneventful pregnancy by normal vaginal delivery. Apgar score was 9 and 9 at 1 and 5 minutes. Birth weight was 3.8 kg. At birth the baby was noticed to have rigid right calcaneovalgus and left equinovarus feet deformities, as well as knee rigidity with limitation of knee extension. Bilateral hip displacement was also diagnosed. Current neurological examination revealed peripheral right facial palsy and torticollis due to shortened right sternocleidomastoid muscle. The upper extremities were without any neurological deficiencies. The lower extremities were in fixed frog-leg position with rigid feet deformity. Active movements manifested with very slight hip adduction and minimal toe movements. The muscles were not detectable on leg palpation. Knee and ankle jerks were absent. Plantar response was mute. Sensation to pinprick and touch was preserved throughout. The anus was closed and anal blink reflex was brisk. The baby demonstrates good head control, raising the head and chest while being in prone position; grasping and transferring an object from one hand to another with good visual attention to the grasped object. The baby had positive stranger anxiety. MRI of the hips showed absence of musculature with preserved fascial planes, vessels, and adipose tissue (Figure 3).

As part of management, both patients underwent surgical correction of their tulip equinovarus. On follow-up at three years of age, their neurological evaluation showed that they both were able to sit up, but not stand up. Only the first patient was able to stand with support.

Figure 1: Magnetic resonance imaging of the thigh revealed paucity of muscles in patient from Case 1.

Figure 2: Another magnetic resonance imaging of the thigh revealed paucity of muscles in patient from Case 1.

Figure 3: Magnetic resonance imaging of the hips showed absence of musculature in patient from Case 2.

DISCUSSION

Despite the fact that amyoplasia is the most common type of arthrogryposis multiplex congenital [2, 3], recognition of considerable muscle underdevelopment in these patients remains surprising and puzzling for pediatric practitioners. Amyoplasia congenita is usually
CONCLUSION

Early recognition of amyoplasia with an assessment of muscle underdevelopment via limb magnetic resonance imaging in children with arthrogryposis multiplex congenita elucidates their clinical presentation and can help in tailoring their treatment and in the prognostication of the degree of their future disability.

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Ariam Diaz – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Dominic Sia – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Valerie May G Sia – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Evelyn Erickson – Acquisition of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Sergey Prokhorov – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Menachem Gold – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published

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Conflict of Interest
Authors declare no conflict of interest.

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REFERENCES


Multiple nocardial brain abscesses in an immunocompromised patient with myasthenia gravis

Ayesha Shaikh, Maria Lola Cevallos, Fang Lan, Jean Pratt Daniel

ABSTRACT

Introduction: Nocardia are gram positive, variably acid-fast positive diphtheroid-like to branched, filamentous, aerobic actinomycetes. Nocardiosis is an opportunistic infection that has been noted in patients with malignancies, systemic lupus erythematosus, HIV infection, hematopoietic stem cell transplant recipients and long-term steroid users. Case Report: A 32-year-old female presented with history of myasthenia gravis on long-term glucocorticoid therapy. During her last admission, wound culture of her left shoulder abscess showed diphtheroid organisms. Patient presented with severe headache, nausea, vomiting and altered mental status. She was initially diagnosed with metastatic cerebral abscess and treated with empiric antimicrobial therapy. Imaging study of the brain showed bilateral occipital ring enhancing lesions. Biopsy results came back as culture positive for nocardia. Patient was subsequently treated with intravenous antibiotics for a total of six months. Conclusion: Cases of nocardiosis may go undiagnosed, either because they respond to empiric antimicrobial treatment or because Nocardia spp. may be difficult to identify in cultures of clinical specimens. They may be mistaken for nonpathogenic microorganisms (diphtheroids) and discarded. High suspicion and early long term institution of therapy are key to a favorable outcome of this disease which has high mortality rates.

Keywords: Nocardiosis, Brain, Abscess, Steroid use

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INTRODUCTION

Nocardia can be found almost universally in soil and plants. Nocardia was first identified by Edmund Nocard in 1888 in bovine farcy. The first human disease was described by Eppinger in 1890. Nocardiosis is an opportunistic infection caused by gram positive, weakly acid-fast, filamentous, aerobic organisms [1, 2]. Nocardia asteroides is the most common species to cause infection in humans [3–5]. There are at least thirteen, Nocardia spp. but N. asteroides, N. farcinia and N. nova (N. asteroides complex) constitute about 80–90% of the total cases [3, 4]. Though nocardiosis is a relatively rare bacterial infection, it is frequently associated with immunosuppression. The majority of infections occur in patients with weakened cell-mediated immunity. Infected population generally comprises those who have received bone marrow or
solid organ transplantation, patients on immunosuppressive therapy, those with human immunodeficiency virus/acquired immunodeficiency syndrome (HIV/AIDS), patients on long-term steroid therapy and those with malignancies [3, 6].

CASE REPORT

A 32-year-old female was brought to the emergency room (ER) with complaints of severe headache, nausea, vomiting and altered mental status. A diagnosis of myasthenia gravis had been made three years earlier, associated mainly with diplopia. Her medications included 60 mg of pyridostigmine and 60 mg of prednisone daily. No history of seizures, fever or photophobia was obtained. On neurological examination, she was confused and incoherent. There was generalized weakness with no focal neurological deficits and preserved deep tendon reflexes. The patient’s mental status deteriorated to the point that she had to be intubated and placed on mechanical ventilation. Non-contrast computed tomography (CT) scan of brain showed multiple ring lesions in both the hemispheres. On review of the chart, it was found that the patient had been recently discharged from surgical service after drainage of left shoulder abscess. Wound culture at that time had grown diphtheroid organisms, and the patient was discharged home on oral augmentin after a brief course of intravenous vancomycin in the hospital.

In view of the immunocompromised status and recent history of shoulder abscess, metastatic cerebral abscess was ascertained to be the most likely cause, and the patient was started on empiric antimicrobial therapy including coverage for possible toxoplasmosis (ampicillin, vancomycin, ceftriaxone, metronidazole and trimethoprim-sulfamethoxazole (TMP/SMX)).

Magnetic resonance imaging (MRI) of the brain with contrast was obtained for better evaluation of the cerebral lesions and showed bilateral occipital ring enhancing lesions (Figure 1, 2). Initial set of blood cultures remained negative after 48 hr. Toxoplasma titers were negative. A stereotactic biopsy of one of the cerebral lesion was done and frank pus was aspirated, however, gram stain failed to reveal any organisms. While awaiting biopsy culture results, empiric antibiotic coverage was continued with the exception of ampicillin.

Serial interval neurological examinations continued to be nonfocal and unchanged from presentation (Figure 3). Computed tomography scan of the chest and abdomen showed no abnormalities. The patient’s mental status improved gradually and the patient was successfully weaned off the ventilator on the seventh day of hospitalization. Aerobic and anaerobic bacterial cultures of the brain biopsy aspirate continued to show no growth. However, on the 10th day of hospitalization, fungal cultures were reported to be positive for Nocardia. At this time, staphylococcal and anaerobic coverage was discontinued and patient was continued on IV ceftriaxone and IV TMP/SMX.

Figure 1: (A, B) Magnetic resonance imaging of the brain with contrast (gadolinium) at presentation, showing bilateral occipital ring enhancing lesions.

Speciation and further identification confirmed the organism to be Nocardia astroides sensitive to the antibiotic regimen.

A permanent intravenous access was established as prolonged antibiotic therapy was necessary and the patient was discharged home with plan to continue intravenous antibiotics for total of six months according to current guidelines.

The patient was subsequently followed-up in the out patient clinic and is doing well with some complaints of occasional sharp pain in left parietal area and no focal neurological deficits. The patient is presently in the fifth week of antibiotic therapy.

DISCUSSION

Nocardiaceae are gram positive, aerobic actinomycetes found naturally in the soil, air and sewage. Nocardia astroides is the predominant species and the one most commonly associated with disseminated disease. Although there have been many reports of disseminated nocardiosis in immunocompromised patients, primary cerebral nocardiosis is a very rare presentation.
Figure 2: (A, B) Magnetic resonance imaging of the brain with contrast (gadolinium) shows punctate pontine lesions slightly larger than on the prior study. Bilateral occipital abscesses have slightly decreased in size and edema as compared to the prior MRI.

Figure 3: The magnetic resonance imaging of the patient post right occipital craniotomy with biopsy of one of the lesions and aspiration of pus.

Nocardial abscess associated mortality is reported to be three times higher than in patients with other bacterial brain abscesses. Management of nocardial brain abscess remains a clinical challenge and is associated with very high morbidity and mortality rates (about 90%) [7–9]. A definitive diagnosis can only be made with the isolation and identification of the organism by invasive procedures and therefore, a high index of suspicion is required as early institution of therapy can be lifesaving. Cultures can take up to 13 weeks to grow and speciation is difficult.

All treatment modalities for nocardiosis generally involve TMP/SMX. However, there have been isolated reports of benefit from amikacin and ceftriaxone. Aspiration has been recommended as the preferred modality initially for nocardial brain abscess, with aggressive surgical management being reserved for the small proportion of patients who do not respond to minimally invasive surgery.

Our patient was a housewife with no history of recent travel, trauma or engagement in water sports. Infection with Nocardia was therefore most likely via the respiratory tract, which is the generally accepted mode of inoculation. Her recent shoulder abscess was in all probability a Nocardial infection as Nocardia can be misidentified for diphtheroid organisms owing to analogous gram staining and morphological features. This fact, coupled with the immunocompromised status raised the suspicion for nocardial abscess initially. Nocardial cerebral abscess can give rise to focal neurological deficits depending on the intracranial location. In our patient, no motor or sensory focal deficits were noted on serial neurological examinations as all of the abscesses were localized to the occipital and posterior cortex. A good clinical response to the combination therapy with intravenous ceftriaxone and TMP/SMX was noted.

CONCLUSION

We report a single case of multiple nocardial brain abscesses in an immunocompromised patient on long term corticosteroid therapy for myasthenia gravis. Nocardiosis frequently goes undiagnosed initially as the patient would have either responded to empiric antimicrobial treatment given for some other reason or because Nocardia can very easily be mistaken for nonpathogenic microorganisms (diphtheroids) and discarded on account of its morphological similarities. The diagnosis requires a high clinical suspicion with early tissue and microbiological diagnosis. Prolonged antimicrobial therapy of 6–12 months and serial imaging is key in treatment and prevention of relapse.

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Author Contributions

Ayesha Shaikh – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the
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REFERENCES

Sporadic pseudohypoaldosteronism: A challenging diagnosis
Suman Preet Kaur Bhullar, Raouf Seifeldin, Nikhil Hemady

ABSTRACT
Introduction: Pseudohypoaldosteronism (PHA) is a rare form of salt-wasting syndrome, caused by peripheral resistance to aldosterone. PHA is of three types: PHA type 1, 2, 3. Pseudohypoaldosteronism type 1 (PHA1) is further differentiated into, (i) hereditary forms, autosomal recessive and dominant, which are caused by epithelial sodium channel and mineralocorticoid receptor mutations respectively and (ii) secondary form which is associated with urological problems. Case Report: We present a case of a male infant who presented with failure to thrive, vomiting, mild dehydration and reflux. Evaluation revealed hyperkalemia with normal glucose and carbon dioxide levels. A preliminary diagnosis of CAH (congenital adrenal hyperplasia) was made. Further workup showed high serum aldosterone and renin levels with normal renal and adrenocortical functions. In line with the investigations the diagnosis of pseudohypoaldosteronism was made. The patient was treated with sodium supplementation, which normalized his clinical state and serum electrolytes. Follow-up revealed weight gain and improved status. Conclusion: Diagnosis of PHA1 is based on plasma electrolyte assessment, elevated renin activity and aldosterone levels with normal renal function. PHA1 results from a renal or systemic resistance to aldosterone. In our reported patient we suspected a renal form of PHA1, which is a milder form and responded well to treatment with salt supplements. Infants who present with electrolyte imbalance like hyperkalemia, hyponatremia and weight loss should be evaluated for adrenocortical function and need careful management. Though PHA is a group of rare syndromes, a high degree of suspicion along with extensive laboratory workup should be pursued in cases with electrolyte imbalances.

Keywords: Pseudohypoaldosteronism, Congenital Adrenal Hyperplasia, Aldosterone

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INTRODUCTION
Pseudohypoaldosteronism (PHA) is a rare form of salt-wasting syndrome, caused by peripheral resistance to aldosterone. PHA has three types: PHA type 1, 2, and 3. Pseudohypoaldosteronism type 1 (PHA1) is further differentiated into, (i) hereditary forms - autosomal recessive and dominant, which are caused by epithelial
sodium channel and mineralocorticoid receptor mutations respectively and (ii) secondary form which is associated with urological problems. PHA being a rare disorder, only about seventy cases have been reported in the literature. We report a case of PHA - which inspite of being a rare disorder, can be potentially life-threatening and also briefly discuss the types of PHA, diagnosis and treatment.

CASE REPORT

A 5-week-old Caucasian male infant was admitted to the hospital for evaluation of failure to thrive. He was born at term via cesarean section and weighed 3.07 kg at birth. He was noted to have mild dehydration in spite of feeding well along with on and off vomiting and some reflux. His weight was 2.96 kg and physical examination was normal at presentation to the hospital. External genitalia were normal.

Initial laboratory investigations showed hyperkalemia with potassium levels of 5.1 mmol/L, with sodium of 122 mmol/L. Repeat testing later on showed potassium level of 8.1 mmol/L. He was resuscitated with D5 0.45 NS. Further biochemistry revealed low urine and serum osmolality, normal urine sodium and urine potassium levels with trace of reducing substance in the urine. Upper gastrointestinal studies were unremarkable. Results of urine analysis and renal functions were normal. Abdominal ultrasound was negative for renal and adrenal abnormalities. A preliminary diagnosis of CAH (congenital adrenal hyperplasia) was made and patient was started on hydrocortisone 50 mg/m², i.e. 1.5 mg thrice a day.

Further workup showed serum aldosterone level of 2174 ng/dL (10–160 mg/dL), PRA (pre renal activity) of 103.9 ng/mL/hr (0.5–1.19 mg/mL), serum renin level of 193998 ng/dL/hr (0.29–3.7 ng/dL/hr), carbon dioxide of 19 mmol/L (normal levels in brackets). Normal levels were found for serum cortisol, androstenedione, 17-OH progesterone, ACTH, FSH, LH and pregnenolone.

In view of the laboratory results hydrocortisone was discontinued and he was started on oral fludrocortisone 0.1 mg twice daily along with NaCl supplements. Potassium lowering therapies were also used along with sodium supplementation. Patient responded to the treatment and started gaining weight. He was discharged home with medications and weighed 3520 gms. On follow-up he was found to be gaining appropriate weight and was kept on oral salt supplements alone. Genetic analysis for PHA in the family was negative.

DISCUSSION

Pseudohypoaldosteronism is a salt-wasting syndrome due to peripheral resistance to aldosterone. This may be either a primary (mutation of MR or ENaC) or a secondary (infection, uropathy, medication) phenomenon. In all cases, sodium reabsorption and potassium excretion are impaired in the principal cell of the collecting duct. The biological characteristics are hyponaatraemia, hyperkalaemia and acidosis [1]. It is thus characterized by three essential features: hyperkalaemia, metabolic acidosis and elevated aldosterone concentration with normal glomerular filtration rate (GFR). Volume depletion or hypervolemia, renal salt wasting or retention, hypotension or hypertension and elevated, normal-high or low levels of renin and aldosterone may be observed in the various conditions that result in differentiating this syndrome in three types of PHA.

Pseudohypoaldosteronism type 1: This was first described in 1958 by Cheek and Perry [2]. This rare syndrome starts during the neonatal period with a wide spectrum [1], PHA1 occurs in two genetic forms, (i) a renal form of autosomal dominant inheritance due to a heterozygous mutation of the mineralocorticoid receptor (MR) gene coding for the mineralocorticoid receptor and, (ii) a severe systemic form of autosomal recessive inheritance due to a mutation of the epithelial sodium channel (ENaC) gene, which is a secondary form usually in association with urinary tract malformation and acute pyelonephritis. Autosomal dominant variant is Renal PHA1; while systemic one due to autosomal recessive inheritance is also known as multiple target organ disorder (MTOD).

Renal PHA1 or early childhood hyperkalemia is probably due to a maturation disorder in the number or function of aldosterone receptors and also in sporadic cases. This form manifests with renal salt loss in infancy and failure to thrive and a gradual improvement with advancing age.

In systemic variant, other organs are involved, such as the sweat glands, salivary glands and colon. The fundamental abnormality in multiple target organ defect (MTOD) PHA1 is a loss-of-function mutation in the alpha or beta subunits of the epithelial sodium channel (ENaC) gene, resulting in defective sodium transport in many organs containing the ENaC gene, (e.g., kidney, lung, colon, sweat and salivary glands). This amiloride-sensitive member of the degenerin/epithelial sodium channel (Deg/ENaC) super family of ion channels is comprised of three homologous units (alpha, beta and gamma) and is expressed in the apical membrane of epithelial cells lining the airway, colon, and distal nephron. ENaC plays an essential role in transepithelial Na+ and fluid balance. PHA1 presents with potential life-threatening salt wasting and failure to thrive in early infancy.

Pseudohypoaldosteronism type 2: This is also known as familial hyperkalemia and hypertension or Gordon syndrome [3, 4]. The classification of this heterogeneous syndrome as PHA is, however, controversial because plasma aldosterone concentrations are highly variable, usually almost normal, and patients respond adequately to mineralocorticoid hormone [5]. The organ involvement and genetic abnormality in pseudohypoaldosteronism type 2 (PHA2) is similar to PHA1. The hallmarks of PHA2 are hypertension, hyperkalemia and correction of
these abnormalities by low doses of thiazide diuretics [6, 7].

**Pseudohypoaldosteronism type 3:** This is transient and secondary to various pathologies related to kidneys or other organs [7]. Rare cases of major intestinal resection [8] or sweat gland dysfunction associated with excessive loss of sodium [9] have been described as leading to PHA III. However, renal causes are encountered more frequently. Nephropathies such as obstructive uropathy [10] or urinary tract infection [11] are reported as causes of transient aldosterone resistance [7]. The main characteristic of this type of PHA is a decreased GFR.

In all cases of PHA, sodium reabsorption and potassium excretion are impaired in the principle cells of collecting ducts. The biological characteristics are hypokalemia, hyperkalemia and metabolic acidosis. After having excluded pseudohyperkalemia due to hemolysis, the diagnosis may be challenging. If serum chloride is normal while serum sodium has decreased and GFR is not impaired, type-4 renal tubular acidosis can be ruled out. The normal hormone levels of ACTH, 17-OH progesterone and cortisol allows exclusion of adrenal insufficiency. Finally, high aldosterone and plasma renin levels lead to the diagnosis of PHA [1].

We diagnosed our patient as a case of PHA1 which is characterized by neonatal salt wasting, vomiting, dehydration and failure to thrive. We would like to summarize our observations in the management of this patient. In our case, diagnosis of PHA1 was based on plasma electrolyte assessment, high renin activity, high aldosterone levels, low level of carbon dioxide with normal renal function resulting from a renal or systemic resistance to aldosterone. Normal levels of cortisol, 17-OH progesterone, ACTH and androstenedione excluded the diagnosis of congenital adrenal hyperplasia and other corticoid dysfunctions. By doing upper gastrointestinal studies, upper gastrointestinal abnormalities were excluded. Normal ultrasound of kidneys and adrenals excluded anatomical abnormalities. We suspected the sporadic form of PHA1 in our patient and treated him with sodium supplementation which normalized his clinical state and serum electrolytes. There was good response to sodium chloride supplementation and he was thriving well on follow up.

**CONCLUSION**

While evaluating any infant with suspected CAH, one should consider pseudohypoaldosteronism as one of the differential and infants who present with electrolyte imbalance like hyperkalemia, hypokalemia and weight loss should be evaluated for adrenocortical function and need careful management. Genetic analysis should be done as the disease can have a genetic predisposition or may be sporadic. Though PHA is a group of rare syndromes, but a high degree of suspicion along with extensive laboratory workup should be pursued in cases with electrolyte imbalances.

A multidisciplinary team approach including a neonatologist, an endocrinologist, genetic expert and a diettian is essential for evaluation of longitudinal growth and neurological development in PHA patients.

**********

**Author Contributions**

Suman Preet Kaur Bhullar – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published

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Nikhil Hemady – Drafting the article, Critical revision of the article, Final approval of the version to be published

**Guarantor**

The corresponding author is the guarantor of submission.

**Conflict of Interest**

Authors declare no conflict of interest.

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**REFERENCES**


Community acquired methicillin sensitive *Staphylococcus aureus* bacteremia, meningitis and brain abscess: A unique presentation

Carlos Gonzalez, Juan Roa, Nehad Shabarek

ABSTRACT

Introduction: *Staphylococcus aureus* causing isolate central nervous system (CNS) infection is rare. It is mostly related to either neurosurgical intervention or previous local or systemic infection rather than spontaneous isolated infection in adults. Case Report: We report a case of a previously healthy 19-year-old male with no risk factors of *Staphylococcus aureus* infection, who was found to have methicillin sensitive *Staphylococcus aureus* (MSSA) bacteremia, meningitis and cerebral abscess, without an apparent source of infection. Conclusion: Our case is a unique presentation of a young male presenting with isolated CNS infection by MSSA. This case highlights the challenge of early diagnosis of brain abscess, an entity that presents very often with non-specific signs, the diagnosis of which can be easily missed, with repercussions on long-term disability and mortality.

Keywords: *Staphylococcus aureus*, Bacteremia, Meningitis, Brain abscess

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INTRODUCTION

*Staphylococcus aureus* is the most virulent of the staphylococcal species with unique versatility to cause various types of infections, from community acquired mild skin infections to highly lethal infections such as necrotizing pneumonia, endocarditis or central nervous system (CNS) infections. CNS infections caused by *Staphylococcus aureus* are in general uncommon. Various series report *Staphylococcus aureus* as the fourth or fifth most common etiology for neuro-infection; more commonly as cause of brain abscess and less commonly as an isolated cause of meningitis (less than 5%) [1–5, 6]. In either case, CNS infection caused by *Staphylococcus aureus* is related to recent neurosurgical intervention or associated with systemic or local infection classically described as a complication of endocarditis or soft tissue infection [1–2, 6]. Although there are a few case reports of CNS infection in patients without an apparent source, most of them are related to community acquire methicillin resistant *Staphylococcus aureus* (CA-MRSA) infections [7]. To date, we have not found any reported case of confirmed meningitis with brain abscess, caused by MSSA infection in a previous healthy patient with no
predisposing infection or risk factors. We report a case of MSSA bacteremia with meningitis and brain abscess in a previous healthy young male without any risk factors and no obvious source of infection, the detection of which was delayed due to a normal brain non-contrast computed tomography (CT) scan findings.

CASE REPORT

A 19-year-old Hispanic male student, presented to the emergency department (ED) complaining of dull frontal and occipital headache associated with fever, few episodes of vomiting, poor oral intake and myalgia for two days. His past medical history was only remarkable for a recent dental procedure. He denied any sick contacts and drug abuse. Initial physical examination was remarkable for tachycardia of 123 bpm and temperature of 38°C. No meningeal signs or focal deficits were found on the initial presentation. The remaining physical examination was unremarkable. Initial laboratory work up was positive for mild hyperglycemia with blood sugar 143 mg/dL, Na 132 mEq/dL and neutrophilia 92% with WBC count 11x10^3/mm^3. Brain CT scan and lumbar puncture (LP) were performed to exclude meningitis or any intracranial pathology. Both the investigations were negative. The patient received symptomatic management with IV fluids and non-steroidal anti-inflammatory drugs (NSAIDs) for fever, with notable improvement. He was discharged home from the ED with symptomatic management. One day after the initial presentation, blood cultures were reported positive for MSSA. Multiple attempts to contact the patient and his family were unfortunately unsuccessfully.

Four days after the initial presentation, the patient was brought to the ED again, after being found unresponsive. Patient’s mother stated he was doing well until early morning when he complained of malaise, fever and headache. Physical examination was positive for temperature of 40.5°C. Other vital signs were normal. The patient was lethargic, found to have nuchal rigidity and was only responsive to painful stimuli. Glasgow coma scale (GCS) was 9/15. Mild patchy folliculitis was noted in the suprapubic area with crusted lesions. He was intubated and transferred to the medical intensive care unit (MICU) with a diagnosis of possible meningitis and MSSA bacteremia based on his previous blood culture. Repeat LP and blood culture were performed. Lumbar puncture showed xanthochromia with RBC 11/mm^3, WBC count 817/mm^3 (neutrophils 90%, lymphocytes 2%, monocytes 8%), glucose 28 mg/dL and protein 126 mg/dL. Blood cultures grew MSSA with the same pattern of sensitivity as the first culture. Non-contrast brain CT scan was again performed, with normal findings (Figure 1). The patient was initiated on bacterial meningitis treatment with ceftriaxone 2 g q12hr, nafcillin 2 g q4hr, and dexamethazone 4 mg q6hr. Ceftriaxone was switched to gentamicin 60 mg q8hr on day-2.

During the hospital course in MICU, transthoracic echocardiogram (TTE) was negative for endocarditis, urine toxicology was negative and HIV status was negative. The patient was extubated four days after admission, at which time a left palpebral ptosis was noted. His pupils were symmetric, 3 mm in diameter, and reactive. He remained lethargic and a contrast enhanced magnetic resonance imaging (MRI) was ordered due to his lack of neurological improvement. The MRI scan of brain showed a focal lesion centered at the left anterior thalamus with contiguous extension into the subthalamic region and left cerebral peduncle of the left midbrain, with a small focus of contralateral extension into the right thalamus (left lesion measure: 3.5 cm height, 2 cm antero-posterior and 1.8 transverse, right lesion 0.4 cm diameter) (Figure 2). These findings were compatible with brain abscesses.

After eight days of medical treatment and observation with the patient was sent for stereotactic biopsy and aspiration of the abscess due to no neurological improvement. The procedure was done without complication and patient started showing signs of improvement. He became more alert and followed commands. Pathology reported gliotic brain tissue with organizing necrosis and hemorrhage without purulent inflammation. Fluid culture was negative.

Ten days after treatment with nafcillin, the patient presented with diffuse erythematous rash involving the palm of the hands. Biopsy of the lesions was remarkable for interface dermatitis, compatible with a drug eruption. Nafcillin was then switched to vancomycin 1 g q12hr. Patient remained stable and was transferred to medical floor and subsequently to rehabilitation
determined that the prior dental work was an unlikely cause of the brain abscess and neurological infection that ensued.

The initial presentation of a patient with cerebral abscess could be very non-specific with only generalized symptoms. This makes the initial recognition very challenging. Different series have reported headache as the most common symptom in a patient with a brain abscess (41–63% cases). Fever is an inconsistent finding, only reported in 25% cases [3, 5]. Altered mental status (AMS) and focal neurologic deficit are inconsistent findings reported in different series with a range for AMS being 18–48% and from 25–65% for focal neurologic deficit [1, 3–5]. Other symptoms, especially gastrointestinal symptoms are present in about 27% cases [5]. In our patient, non-contrast CT scan of the head was performed and yielded a negative result on both presentations to the ED. A contrast MRI scan done several days after admission showed the thalamic abscess. The initial negative finding on CT scan and the absence of focal neurological deficit contributed to the delay in the diagnosis of brain abscess. Despite this, non-contrast CT scan is shown to be a good initial test for a patient with suspicion of cerebral abscess, even though in the early phase, non-contrast CT scan could be negative [7, 9, 10]. When it is used in combination with contrast CT scan, the sensitivity is very high, as was demonstrated in a series of 50 cases where positive CT scan finding was reported in 100% of the cases [1]. When CT scan is not diagnostic, MRI scan with contrast is in general the next step. Contrast MRI scan is highly accurate for detection of brain abscess [11].

Treatment of brain abscess is based on pertinent antibiotic therapy and neurosurgical mechanical drainage in most cases. Recommended initial empiric antibiotic treatment is a third generation cephalosporin in combination with metronidazole. If there is suspicion or risk factors for Staphylococcus aureus infection, vancomycin should be added until culture identification and sensitivities are available. Vancomycin remains the drug of choice when there is a suspicion for Staphylococcus aureus infection despite poor penetration in the CNS [1–5, 10]. As a result, some authors have proposed linezolid as a good alternative for vancomycin, as it has better penetration in the CNS [12, 7, 13]. In general, if the abscess is more than 2–2.5 cm, neurosurgical intervention is indicated. Stereotactic aspiration is the treatment of choice for abscesses located in the brain stem [1, 5, 14].

**DISCUSSION**

*Staphylococcus aureus* infections remains a major cause of morbidity and mortality, with presentations ranging from simple cutaneous infection to serious life-threatening infections. Isolated meningitis caused by *Staphylococcus aureus* is rare accounting for less than 5% of all meningitis presentations [2]. On the other hand, *Staphylococcus aureus* is a major cause of cerebral abscess being the second most commonly found bacteria in patients with cerebral abscess, with *Streptococcus milleri* being the most common, and anaerobes the third most common [1, 3–5]. CNS infection caused by *Staphylococcus aureus* is usually associated with prior neurosurgical intervention or with typical risk factors such as endocarditis, soft tissue infection, skin abscesses, AIDS or intravenous drug use [8, 6]. In this case, the only infection that could be associated with the MSSA bacteremia was the mild suprapubic folliculitis, which we determined to be a very unlikely source as it was a mild cutaneous infection in an immunocompetent host. The patient was evaluated by oral maxillofacial surgery (OMS) department who

Figure 2: Brain MRI, Axial view showed a focal lesion centered at the left anterior thalamus with a small focus of contralateral extension into the right thalamus.

facility to continue antibiotic treatment for 6–8 weeks or until resolution of the brain abscess. Patient was seen four weeks after with complete neurological recuperation and resolution of the brain abscess on repeat MRI scan.

**CONCLUSION**

Our case highlights the challenge of early diagnosis of brain abscess, an entity that presents very often with non-specific signs, the diagnosis of which could be easily missed, with repercussions on long-term disability and mortality. The mortality rate in patients with brain abscess remains high despite early detection and improvement in treatment. Long-term disability is also very high. Hence an early diagnosis and neurosurgical
intervention could prove significant in the final outcome for these patients.

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Author Contributions
Carlos Gonzalez — Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Juan Roa — Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Nehad Shabarek — Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

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The corresponding author is the guarantor of submission.

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Authors declare no conflict of interest.

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REFERENCES

Reversible myeloneuropathy and pancytopenia related to copper deficiency from gastric bypass surgery: A case report

Laide Bello, Joseph Fiore

ABSTRACT

Introduction: Weight loss surgery has become an increasingly popular means of combating the obesity epidemic in modern society but like any procedure, it does not shy away from immediate and long-term complications. Copper deficiency has occasionally been reported to occur many years afterwards but with an increased incidence of bariatric procedures and reduced awareness, the effects of this deficiency could now appear to favor an earlier onset. Case Report: We report a case of a 56-year-old Caucasian female with a history of gastric bypass surgery five year ago; with an unsteady gait, weakness, decreased visual acuity, tingling with numbness in her hands and pancytopenia for the last month. She was treated for copper deficiency. Conclusion: Effects of copper deficiency have been shown to cause a wide array of abnormalities related to inactivation of enzymes such as cytochrome c oxidase, superoxide dismutase, dopamine beta hydroxylase and metallothionein. This can lead to reduced nerve transmission within the central nervous system causing motor and sensory polymyeloneuropathy and an overall reduction of energy required for blood cell formation. With early surveillance, such anomalies can be detected and potentially reverse the effects of this micronutrient deficiency.

Keywords: Myeloneuropathy, Hemoglobinopathy, Micronutrient deficiency

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INTRODUCTION

Morbid obesity is one of the major risk factors associated with chronic diseases and conditions such as heart disease, cancer, stroke, diabetes and hypertension [1]. Given this impact on morbidity and mortality in the 21st century, weight loss surgery options are bound to be more prevalent than ever before and the Roux-en-Y gastric bypass remains the most common type done in the United States [2]. This procedure involves dividing the stomach into a small upper pouch and anastomosing it to a distal segment of the jejunum thus creating a gastro-jejunostomy for drainage of gastric remnant contents, bile and pancreatic enzymes. This bypass potentially eliminates common micronutrients such as iron, vitamin B12, calcium and vitamin D from being absorbed through the latter stomach and initial part of the small intestine [3–5]. There have been few reports of other complications such as copper deficiency which caused detrimental long-term outcomes such as
myeloneuropathies, anemia, leucopenia and sometimes thrombocytopenia, but with early surveillance these conditions can be reversed [6, 7].

**CASE REPORT**

A 56-year-old Caucasian female presented to our emergency room with complaints of unsteady gait, visual disturbance, dizziness, fatigue and recurrent tingling with numbness in her hands for the past month. She lost 68.03 kg ever since her gastric bypass surgery five years ago and more recently developed poor appetite, recurrent diarrhea and nausea despite being compliant with once daily iron, thiamine and folic acid supplements. Her gait had worsened to the point of requiring a cane for ambulation due to frequent falls. A recent upper endoscopy showed a gastric-jugal anastomotic ulcer (*H. Pylori* negative) and colonoscopy revealed mild diverticulosis with several previous examinations in the past failing to show any clear cut etiology.

The past medical history of the patient was significant for depression and chronic hepatitis C (Genotype 2). She was a retired home care administrator, with no family history of gastrointestinal malignancy and routinely took pantoprazole, citalopram, calcium and vitamin D supplements.

On physical examination, patient appeared frail with a body mass index 17.5 kg/m² (84% of ideal body weight), blood pressure 124/84 mmHg, heart rate 90/min and respiratory rate 14/min. Neurological review revealed reduced strength and sensation over her lower extremities, decreased ankle jerk, ataxic gait and moderate loss of vibratory and joint position sense in the toes. An ophthalmologic examination showed reduced visual acuity and protracted optic disc swelling. The rest of the physical examination was normal. Her laboratory examination was unique for a white blood cell count 2.2x10⁹/mm³ (nadir 1.4x10⁹/mm³), hemoglobin 8.9 g/dL, mean corpuscular volume (MCV) 72 f and platelet count 8.8x10⁹/mm³. Her iron, percent saturation and vitamin B12 levels were preserved in a high normal range at 76 µg/dL, 40% and 652 µg/mL, respectively. A hepatitis C viral load was undetectable while creatinine phosphokinase remained within normal limits. Serologies for Lyme titer and syphilis were undetectable. A brain and entire spine magnetic resonance imaging was otherwise normal except for minimal T2 hyper-intensities within the periventricular white matter suggesting demyelination [8–10]. Her cerebrospinal fluid analysis was essentially unremarkable with no evidence of oligoclonal banding. An electroencephalogram recording revealed subtle slowing of the background suggestive of mild encephalopathy with no epileptiform activity and nerve conduction studies showed some motor and sensory polyneuropathy affecting different parts of her upper and lower extremities. At that juncture given her history of gastric bypass surgery, ongoing pancreatepnea and complains of dizziness with unsteady gait, it was decided to assess for copper deficiency. This was seen low at 0.44 µg/mL (normal range 0.75–1.45 µg/mL) along with zinc at 0.37 µg/mL (normal range 0.66–1.1 µg/mL) but had a normal ceruloplasmin level. The 24 hour urine collection for copper was also low at 9 µg/L (normal range 15–60 µg/L) and over the next three days she received a once daily intravenous infusion (containing 1 mg of copper) in dextrose water along with a high potency Women’s Ultra Mega vitamin supplement four times a day. This contained 2 mg of copper, several fat and water soluble vitamins as well as trace elements like manganese, chromium, selenium, magnesium and zinc. Over the next four to five days, her gait and vision improved remarkably with increased acuity and resolution of the optic disc swelling on examination. She no longer required any assistance with ambulation after a week, and was subsequently discharged on oral copper supplements. Table 1 gives her follow-up laboratory data after the first and second months. On subsequent follow ups in the out-patient clinic, the patient had shown great improvement with her overall strength and ambulation but still had some lingering but subtle tingling with numbness in the hands.

**DISCUSSION**

Copper remains an essential nutrient serving as a ceruloplasmin co-factor in the formation of transferring [11]. It thus facilitates iron uptake and ensures adequate red and white blood cell formation. Copper also plays a critical role in activating enzymes such as cytochrome c oxidase, superoxide dismutase, dopamine beta hydroxylase, metallothionein and its deficiency can lead to reduced nerve transmission within the central nervous system and less adenosine triphosphate production for synthesis of hemoglobin [5]. The actual

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Table 1: Laboratory data compared from admission and one month later after copper supplementation
mechanism on how neutropenia occurs is still unknown, but Lazarhick et al. suggested an inhibition of differentiation and self-renewal of CD34 positive hematopoietic progenitor cells as a likely cause [12]. Most cases of copper deficiency myeloneuropathy typically occur after a few decades of gastric bypass surgery but in our patient the symptoms were seen only after a few years [8, 13]. Shorter gastrointestinal tracts may cause reduced sites for reabsorption and a lack of micronutrient replacement can compound this deficiency. Zinc can interfere with copper metabolism since they compete for absorption via the same site and O’Donnell et al. advised against simultaneous supplementation in situations where both are found to be deficient [14]. Previous studies have linked hyperzincemia from toxic exposures as a potential cause of copper deficiency but this was not the case in our patient. With varying degrees of copper deficiency, patients may not necessarily have all the signs and symptoms listed and in order to make the diagnosis a clinician would need to have a high index of suspicion, along with demonstrable low copper levels. The occurrence of long-term irreversible neurological damage is not known and as such it is paramount to consider early surveillance. Kumar et al. have also studied the value of urinary copper as a measure of its deficiency but concluded that a serum copper level remains the best and most reliable assay [8]. An initial intravenous dose of 1 mg of copper is advised for the first three days after which patients can continue on oral supplementation of 8 mg of copper gluconate daily [15]. Our patient received these and blood levels for copper gradually normalized over the next two months along with other respective hematologic parameters. She did not require any blood transfusions during her stay and the abatement of her symptomatology was quite impressive over the immediate days to weeks of commencing therapy. In the absence of other causes for pancytopenia, blood levels usually improve or normalize anywhere within three days to six months after supplementation [11, 15–17]. The actual threshold between copper concentrations, tissue stores and neurological sequelae remains to be established and more studies shall be required in the future to establish this. The myeloneuropathy described here can also mimic subacute combined degeneration typically seen with vitamin B12 deficiency, as such this should also be assessed and treated promptly. Even though serial levels of serum copper measured over time was seen to rise, potential confounding effects could exist with various vitamins and trace elements contained in the branded high potency Women’s Ultra Mega vitamin supplement.

CONCLUSION

Early surveillance for copper deficiency has its benefits and ought to be routinely evaluated after a patient undergoes gastric bypass surgery as this gives the clinician an avenue to identify preventable and reversible causes of blood cell disorders, leukemic transformation and polynuropathies that would otherwise have been termed idiopathic.

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Author Contributions

Laide Bello – Substantial contributions to conception and design, Analysis and interpretation of data, Drafting the article, revising it critically for important intellectual content, Final approval of the version to be published

Joseph Fiore – Substantial contributions to conception and design, Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

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REFERENCES


Spinal tuberculomas mimicking spinal dural arteriovenous fistula: A case report

Jyoti Sureka, Varsha Mary Khalkho, Binita Riya Chacko

ABSTRACT

Introduction: Tuberculosis is a very common disease in developing countries and has been found to affect almost all the parts of the body. We report the case of a patient who had spinal cord tuberculomas without evidence of symptoms of systemic tuberculosis. The lesions were located at the surface of lower thoracic cord and mimicked a spinal-dural arteriovenous fistula (SDAVF) on magnetic resonance imaging (MRI). Case Report: The patient was 45-year-old man who presented with a history of progressive paraparesis with a clinical suspicion of intramedullary tumor. First diagnosis was made as SDAVF on MRI. Then he underwent diagnostic and therapeutic digital subtraction angiogram which was negative for the same. Again MRI was reviewed by a senior radiologist and a final diagnosis of spinal cord pial surface tuberculomas was made, confirmed by cerebrospinal fluid analysis and treated by appropriate anti-tubercular therapy. Conclusion: Tuberculosis can mimic a number of disease entities. It is important to be familiar with various a typical radiological presentations of tuberculosis.

Keywords: Tuberculoma, Spinal dural, Arteriovenous malformation

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INTRODUCTION

Tuberculosis (TB) is a very common disease in developing countries and has been found to affect almost all the parts of the body. It can affect any organ or organ system of the body. Tuberculosis primarily affects the chest and can involve multiple extrapulmonary sites like heart, bones, joints, gastrointestinal system, genitourinary system, central nervous system, eyes, etc. Spinal TB accounts for more than half of the musculoskeletal TB. The extradural form of spinal TB is most common [1]. Uncommonly, it can present as arachnoidal, intradural extramedullary and intramedullary form. Spinal intradural-intramedullary tuberculoma is extremely rare entity can give rise to a variety of clinical and radiologic features which can mimic a number of other spinal cord lesions particularly intramedullary tumors [2, 3]. We report the case of a patient who had spinal cord tuberculomas without evidence of symptoms of systemic tuberculosis. The lesions were located at the surface of lower thoracic cord and mimicked a spinal dural arteriovenous fistula (SDAVF) on magnetic resonance imaging (MRI).
CASE REPORT

A 45-year-old male presented to the neurology out patient department (OPD) with complaints of back pain and progressive lower limb weakness since six months. There were no bowel or bladder difficulties. On physical examination, there was spine tenderness over the mid and lower thoracic spine, muscle weakness and abnormal reflexes in lower limbs. Laboratory investigations revealed positive results for human immunodeficiency virus (HIV) and hepatitis B virus (HBV). However, complete blood picture, ESR, C-reactive protein and other routine blood examinations were within normal limits. Chest X-ray was also normal. Based on clinical findings possibility of intramedullary tumor was considered. Patient underwent gadolinium-enhanced MRI of spine. Sagittal and coronal T2-weighted MRI of thoracolumbar spine showed multiple tortuous flow voids along the surface of mid and lower thoracic spinal cord. The lower thoracic cord was also slightly enlarged and showed increased intramedullary signal intensity (Figure 1). Diagnosis of SDAVF with edema or ischemia of cord secondary to venous hypertension was made. For further management, patient underwent diagnostic and therapeutic selective spinal DSA. The angiogram did not reveal any feeding vessels or nidus to suggest arteriovenous fistula (AVF) (Figure 2A–B). MRI images were further reviewed by a senior radiologist with a referring doctor. The coronal gadolinium-enhanced T1-weighted images showed multiple, small, almost similar sized ring and nodular enhancing lesions along the pial surface of the cord with central T2 hypointensity which appeared as a flow void on T2-weighted sagittal and coronal images (Figure 3). There was no focal enhancing lesion or abnormal enhancement in the region of high signal intensity of the lower thoracic spinal cord. Considering the immunocompromised status, a final diagnosis of spinal cord pial surface tuberculomas with associated cord edema or myelitis was made. MRI of the brain did not reveal any lesion. Patient underwent CSF analysis which grew acid-fast bacilli and isolated species was *Mycobacterium tuberculosis* and hence the diagnosis was confirmed and first line anti-tubercular therapy (ATT) was started. The patient responded well to ATT.

DISCUSSION

Central nervous system TB is commonly seen in tropical countries [4]. In immunocompromised individuals, the presentation of tubercular lesions may be atypical and can result in delayed diagnosis [5, 6]. Imaging plays an important role in recognition of these atypical cases that mimic other neurologic conditions thus helps in early diagnosis and treatment, which otherwise may result in irreversible neurological sequel [4–6]. A few case reports talk about the isolated meningeal or spinal tuberculoma mimicking spinal tumor. However, spinal TB mimicking a SDAVF not yet reported in literature. Extradural form is the most common while arachnoiditis, intradural and intramedullary tuberculomas are uncommon presentation of spinal TB [4, 7]. This atypical form of TB can occur as a direct extension from the vertebrae, as a downward extension of intracranial tubercular meningitis, and less commonly as tuberculous lesions primarily developing in spinal meninges [5, 8]. MRI is the imaging modality of choice for these lesions. Spinal cord TB, generally, present as intramedullary tuberculomas with or without myelitis and syrinx. Clinically as well as radiologically, intramedullary tuberculomas may be difficult to differentiate from space-occupying lesions such as primary and metastatic intramedullary spinal tumors and other chronic granulomatous diseases (sarcoidosis, histiocytosis and brucellosis) [9]. Commonly the intramedullary tuberculomas have specific MR features and can be diagnosed on imaging. It has a typical “target sign” on T2-weighted imaging, demonstrating low signal center (caseous material) surrounded by high signal rim (peripheral infective granulation tissue), which helps to differentiate tuberculoma from other intramedullary lesions [10, 11]. On intravenous contrast study, these lesions show rounded nodular and ring like peripheral enhancement with non-enhancing center of the lesion. If the lesion has a typical appearance on MRI, and if the patient has systemic tuberculosis, diagnosis of tuberculoma can be made easily [9–11]. If the patient does not have systemic tuberculosis and has immunocompromised status, MRI features can be atypical as seen in our case and diagnosis can be difficult. In our case, all the lesions were of almost similar size and appeared as hypointense dots on the surface of cord on T2-weighted images and thus

![Figure 1: A 45-year-old male presented with progressive worsening of lower limb weakness since several months. T2-weighted (A) mid, (B) para-sagittal, and (C) coronal MRI of mid and lower thoracic spine demonstrate diffuse, lower thoracic spinal cord, high signal intensity and mild swelling (stars) suggesting cord edema with multiple tortuous hypointense dots (open arrows in sagittal and black arrows in coronal images) giving appearance of flow voids along the surface of mid and lower thoracic cord.](image-url)
mimicked SDAVF. Further cord swelling, hyperintensity and involvement of lower thoracic cord again supported the diagnosis of SDAVF. During the initial MRI evaluation, gadolinium enhanced imaging was overlooked and thus the lesion was mistakenly diagnosed as SDAVF and patient had to undergo DSA. Gadolinium enhanced images clearly demonstrated ring and nodular enhancement of all the T2 hypointense lesions thus helped in making the final diagnosis of tuberculosis.

CONCLUSION

Tuberculosis has a variety of clinical and radiologic features and can mimic a number of other disease entities. Therefore it is important to be familiar with the various atypical radiological presentations of TB particularly in immunocompromised patients to ensure early, accurate diagnosis and treatment and to avoid unnecessary invasive and costly investigation like DSA. Spinal cord tuberculosis can mimic SDAVF if the lesions are multiple, very small and similar in size and are located at the pial surface of the cord. The associated cord swelling and hyperintensity can simulate venous hypertension associated with SDAVF. Gadolinium enhanced imaging is extremely important. The coronal T1-weighted post contrast imaging can show enhancing periphery and non-enhancing center of the lesion which is not seen in hypertrophied vessels of SDAVF.

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Author Contributions

Jyoti Sureka – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Varsha Mary Khalkho – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Binita Riya Chucko – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published

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The corresponding author is the guarantor of submission.

Conflict of Interest

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REFERENCES

Unusual case of ascites

Guido Poggi, Benedetta Montagna, Pamela Di Cesare, Erica Quaquarini, Emma Pozzi, Carlo Aprile

ABSTRACT

Introduction: *Clostridium difficile* infection is very commonly related to antibiotic therapy. The spectrum of clinical manifestation of *C. difficile* infection may include, in an increasing order of severity, absence of symptoms, colitis without formation of pseudomembranes and pseudomembranous colitis (PMC). PMC is a severe but rare complication of the infection. It is related to the bacterial production of enterotoxin A and B. Its clinical features include diarrhea, abdominal tenderness and fever. In the worst case, it may progress to toxic megacolon and colonic perforation. Ascites is an infrequent direct complication of most severe cases of PMC. Case Report: We report a case of ascites arising two weeks after the resolution of *C. difficile* infection, in which hypoalbuminemia, caused by protein losing enteropathy, was the most likely pathogenetic mechanism. The patient recovered completely after human albumin intravenous support and diuretics. Conclusion: Protein losing syndrome represents a group of disorders in which hypoproteinemia and edema occur in the absence of either proteinuria or defects in protein synthesis. It is a consequence of the intestinal epithelial cell damage and increased mucosal permeability induced by *C. difficile* enterotoxin. The clinical manifestations are ascites and low serum albumin levels. The treatment requires human albumin intravenous integrations and diuretics. Ascites could be a direct complication of antibiotic-associated pseudomembranous colitis, but it could also be the manifestation of a protein-losing enteropathy, caused by inflammatory damage of gastrointestinal mucosa.

Keywords: *Clostridium difficile*, Infection, Complications, Protein losing enteropathy, Ascites, MSSA infection

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INTRODUCTION

*Clostridium difficile* is a Spore forming, gram positive bacillus. It is a well-recognized cause of antibiotic associated diarrhea. The spectrum of clinical manifestation of *C. difficile* infection may include, in an increasing order of severity, absence of symptoms, antibiotic–associated colitis without the formation of pseudomembranes and pseudomembranous colitis (PMC). PMC is a severe but rare complication of the infection, related to the bacteria production of enterotoxin A and B. Its clinical features include diarrhea, abdominal tenderness, fever, dehydration and leukocytosis. In the worst case, it may progress to toxic megacolon and colonic perforation resulting from full-thickness colonic wall necrosis. The diagnosis of PMC
depends on the demonstration of *C. difficile* toxins in the stool or on endoscopic evidence of adherent yellow plaques along colonic wall. Computed tomography (CT) scan is also useful for PMC diagnosis, showing colonic wall thickening, low-attenuation mural thickening corresponding to mucosal and sub-mucosal edema and pericolonic stranding [1–3]. Early treatment with oral metronidazole or oral vancomycin usually leads to a clinical improvement within 48–72 hours.

Surgical intervention is mandatory in perforation and may be required in severe cases where medical treatment is not sufficient. Ascites is an infrequent direct complication of most severe cases of PMC [4].

We describe a case of ascites arising two weeks after the resolution of *C. difficile* infection, in which hypalbuminemia, secondary to protein losing enteropathy was the most likely pathogenic mechanism.

**CASE REPORT**

A 54-year-old man was admitted to our hospital with complaints of abdominal distension and bilateral peripheral edema. He had no previous history of such illness. Ten weeks before his admission, he developed a soft tissue infection at the back of his left hand which rapidly evolving to a necrotising fascitis of the left forearm and required surgical decompression followed by necrotic tissue debridement. Microbiological culture of the tissue samples taken from the wound identified a growth of a methicillin sensitive staphylococcus aureus (MSSA) producing panton-valentine leukocidin toxin. Blood cultures were negative. The patient was treated with intravenous ciprofloxacin (200 mg/bid) and clindamycin (500 mg/bid) for ten days. After the treatment, he was discharged home and continued therapy with oral ciprofloxacin (500 mg/bid) and oral rifampicin (600 mg/day). After one month of treatment, he developed non-bloody, watery diarrhea consisting of 8-12 stools daily, fever and abdominal pain. The patient was re-hospitalized. Stool cultures were negative for shigella, salmonella, campylobacter and yersinia bacteria while they were positive for *C. difficile* toxins. The EIA test for *C. difficile* was also negative. The patient was treated with oral vancomycin (500 mg every 6 hours for 5 days), with progressive clinical benefit and resolution of diarrhea, and he was discharged home.

Due to the subsequent appearance of abdominal swelling and peripheral edema, occurring two weeks after the resolution of abdominal symptoms, the patient was re-admitted to our hospital. On physical examination, the patient was alert and had little discomfort. Vital signs were normal and the patient had no fever. Abdominal examination showed diffuse abdominal distension with active bowel sounds, a dull percussion note and mild, diffuse tenderness without jaundice. The patient had pitting edema of lower extremity. Lymphadenopathy was not present. Neurological and cardiac examinations were normal. Laboratory tests revealed normal hemoglobin values, normal renal function tests, no electrolytes abnormalities, normal hepatic and coagulation function. Erythrocyte sedimentation rate and C-reactive protein were negative. Total serum protein was 5.0 g/dL with marked hypoalbuminemia (2.5 g/dL). Mild lymphopenia and hypogammaglobulinemia were also detected. Hepatitis B surface antigen and hepatitis C antibody were negative.

Computed tomography scan showed thickened colonic wall and massive ascites without evidence of hepatosplenomegaly, portal or hepatic vein thrombosis or peritoneal thickening (Figures 1, 2). Abdominal paracentesis was performed. Ascitic fluid examination showed WBC count of 400 cells/mm³ with 65% polymorphonuclear leukocytes, total protein 3.0 g/dL, albumin 1.4 g/dL. Serum-ascites albumin gradient (SAAG) was 1.1 and ascitic fluid/serum LDH ratio was 0.78. Cultures of ascitic fluid for bacteria and mycobacteria were negative and cytological examination did not find malignant cells.

The *C. difficile* toxin was not detected in ascitic fluid. Urine collection over 24 hours failed to show proteinuria. A stool collection for fecal fat was normal. Serologic testing for celiac disease was negative. Fecal α-1 antitrypsin (A1AT) clearance was found to be elevated (288 mg per 100 mL; normal value <30 mg/dL) suggesting that the severe hypoalbuminemia might be caused by protein losing enteropathy. Tc-99m labeled human serum albumin scintigraphy demonstrated an abnormal excretion of Tc-99m albumin into the large bowel confirming the diagnosis of protein loosing enteropathy (Figure 3).

During the hospitalization, patient was treated with salt restriction, human albumin intravenous infusion and diuretics. The patient gradually recovered and he was discharged. Three weeks later, ascites had completely disappeared as shown by ultrasound examination. At a follow-up visit one month later, he reported having no diarrhea and serum albumin levels were normal. After three months, an abdominal CT scan

![Image](https://www.icasereportsandimages.com)

Figure 1: Abdominal computed tomography scan showing ascites (marked by 'a').
illness, for example cirrhosis, congestive heart failure, nephrosis or disseminated carcinomatosis. On occasion, ascites may develop as an isolated finding in the absence of a clinically evident disease. In such cases, a careful analysis of ascitic fluid may suggest the etiology: the serum ascites–albumin gradient (SAAG) > or < 1 g/dL can differentiate between a transudate or an exudate fluid. Serum ascite–albumin gradient >1 is usually related to uncomplicated cirrhosis, alcoholic hepatitis, congestive heart failure and Budd–Chiari syndrome; while SAAG <1 is related to other causes, such as peritoneal carcinomatosis, tuberculous peritonitis, pancreatitis, serositis, pyogenic peritonitis, and nephritic syndrome [5].

In our case, ascites was the late complication of a C. difficile associated diarrhea following a prolonged antibiotic therapy for a soft tissue infection. The patient had no other systemic disease that could have justified the occurrence of ascites, such as hepatic or cardiac failure. SAAG was 1.1 g/dL with mild leukocytosis in the fluid and proteinuria was absent.

A literature review revealed that ascites occurs more frequently as an indirect complication of severe cases of PMC. Systemic capillary leak is considered the most likely mechanism of ascites, occurring in the worst cases of PMC [6–8]. However, our patient did not develop a severe form of colitis and the onset of ascites occurred after a few days from the complete resolution of gastrointestinal symptoms. On investigating we found that, in this case, the prevalent mechanism of formation of ascites was a protein-losing enteropathy.

Protein-losing enteropathy is not a specific disease but rather a group of gastrointestinal and non-gastrointestinal disorders occurring with hypoproteinemia and edema in the absence of either proteinuria or defects in protein synthesis. Diseases characterized by excess protein loss into the gastrointestinal tract are caused by mucosal ulceration (e.g., ulcerative colitis, gastrointestinal carcinomas, and peptic ulcer), by damage to mucosa without ulceration (e.g., celiac sprue and Ménétrier’s disease) or by lymphatic dysfunction; either primary lymphatic disease or secondary to partial lymphatic obstruction (e.g. intestinal lymphangiectasia, mesenteric nodes or lymphoma, cardiac disease) [9].

C. Difficile enterotoxins A and B induce intestinal epithelial cell damage, increase mucosal permeability, stimulate interleukin (IL)-8 synthesis, and cause an acute inflammatory response characterized by neutrophil recruitment and tissue damage [10]. This condition can lead to a protein losing syndrome with severe hypoalbuminemia that can result in peripheral edema and ascites, as we described in this case report.

The management of ascites involves, apart from the specific antibiotic therapy for C. difficile infection, human albumin intravenous integrations and diuretics.

**CONCLUSION**

In summary, ascites could be a direct complication of antibiotic-associated pseudomembranous colitis but it
could also be the manifestation of a protein losing enteropathy, caused by the inflammatory damage of the gastrointestinal mucosa.

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Author Contributions
Guido Poggi – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Final approval of the version to be published
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REFERENCES
Feeding dystonia: A classical presentation of neuroacanthocytosis

Suman Kushwaha, Akhila Panda, Vachan Mehta, Seema Malik, Ishita Pant

ABSTRACT

Introduction: Neuroacanthocytosis (NA) is a heterogeneous neurodegenerative genetic disorder caused by disease specific genetic mutation. Being an extremely rare disorder, only a few thousand cases have been reported till date. This clinical entity was described by Citchley et al. and was initially named Levine–Citchley syndrome. It is characterized by movement disorder due to degeneration of the basal ganglia along with cognitive and behavior changes. The classical clinical presentation includes the troublesome abnormal involuntary movements in form of chorea, dystonia and dyskinesia. Self mutilation of the lips and tongue is characteristic of choreoacanthocytosis. The NA syndromes have been broadly divided into two subtypes, (i) core NA syndromes, and (ii) conditions with alterations in the lipoprotein metabolism. The genes of the different NA syndromes have been identified but the mechanism of these genetic mutations is not known. The management of NA is primarily symptomatic and rehabilitative.

Case Report: We are describing a case of neuroacanthocytosis with typical phenotype of choreoacanthocytosis. The phenomenon of feeding dystonia is classically being discussed in this report. Neuroimaging demonstrated the atrophy of the caudate nucleus resembling Huntington’s chorea. Acanthocytic red blood cells were seen in peripheral smear in our patients. Conclusion: The recognition of neuroacanthocytosis is improved due to better characterization of the clinical symptoms and investigations of this heterogeneous entity. The presented case describes the typical clinical characteristics and investigations supporting the diagnosis of this under diagnosed clinical syndrome.

Keywords: Feeding dystonia, Chorea, Acanthocytes, Neuroacanthocytosis

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INTRODUCTION

Neuroacanthocytosis (NA) is an uncommon, heterogeneous, genetic neurodegenerative disorder. This clinical entity was described by Citchley et al. and was initially named Levine–Citchley syndrome. The clinical manifestations include variety of movement disorders and cognitive and behavioral changes closely resembling Huntington's disease. The orofaciolingual
Dyskinesia is prominent and characteristic of neuroacanthocytosis. There is presence of thorny deformation in the lipid membrane of the circulating red blood cells called acanthocytosis. These acanthocytes are distinct and unique for this syndrome. The recognition of neuroacanthocytosis is improved due to better characterization of the clinical symptoms and investigations of this heterogeneous entity. The presented case describes the typical clinical characteristics and investigations supporting the diagnosis of this under diagnosed clinical syndrome.

CASE REPORT

A 30-year-old female presented with three years history of progressive difficulty in eating. She attributed it to abnormal involuntary movement of tongue and sustained spasmodic movement of the neck. She used different maneuvers during eating like pushing the bolus with the fingers inside the mouth and drinking water and food in supine position. She sometimes developed choking sensation and cough while eating food. Occasionally, she had nasal regurgitation while drinking water. She had abnormal, repetitive, non-patterned, involuntary, arrhythmic, non-purposeful movements of the whole body including head and neck. These movements disappeared in sleep and were not suppressed with voluntary action. Since last nine months she had developed abnormal behavior in the form of excessive and non-purposeful talking, repeating the sentences and motor acts. The caregivers reported the habit of self-mutilation. She repeatedly chewed her lips which lead to severe injury of lower lip.

There was no history of visual problems, limb weakness, sensory, cerebellar or autonomic disturbance. She had significant weight loss. Her personal history revealed poor maintenance of hygiene and history of secondary amenorrhea. Family history and past medical history were non-contributory.

General physical examination showed poor self care and cachexia. The vital parameters were normal. A significant lip biting mark was present in inner aspect of lower lip (Figure 1) suggestive of self-mutilation and repeated friction of the lower lip due to orofacial dyskinesia. Other systemic examination was normal. On neurological examination, she was conscious and oriented. Marked behavior changes in form of repetition of words and perseveration of motor acts were present. Neuropsychological assessment shows high frontality on frontal lobe assessment by frontal assessment battery (FAB) battery. Memory functions were normal. She had features of obsessive compulsive behavior and anxiety. Cranial nerve examination was normal. The limbs were hypotonic with power of 5/5 on Medical Research Council (MRC) scale in all the limbs. Generalized areflexia with flexor plantar response was present. All the primitive reflexes were absent. Sensory, cerebellar and autonomic system examination revealed no abnormality.

Extra pyramidal system examination showed combination of abnormal involuntary movements. Chorea was prominent involving orolinguinal, neck, trunk and limbs. Tongue had classical jack in box phenomenon. In addition to chorea, head and neck shows intermittent dystonic movements. Due to combination of these abnormal movements of tongue and neck the patient had classical feeding difficulties described as feeding dystonia. She walked with bizarre gait due to abnormal movements of the trunk. The routine investigations, complete blood count, kidney and liver function tests, serum electrolytes, lipid profile, copper studies, HIV ELISA, blood VDRL were within normal limits.

Creatinine phosphokinase was raised to 614 U/L. Peripheral blood acanthocytes were visible on screening of three consecutive samples (Figure 2). Nerve conduction studies showed sensorimotor axonal neuropathy. Neuroimaging of brain using 3 Tesla MRI showed diffuse cerebral atrophy with significant caudate atrophy (Figure 3).

Figure 1: Self-mutilation of lower lip. Severe injury due to lip biting and dyskinesia causing repeated irritation by teeth.

Figure 2: Peripheral smear showing acanthocytes.
DISCUSSION

Neuroacanthocytosis is a rare movement disorder syndrome. The prevalence is estimated to be less than 1–5 million. Neuroacanthocytosis has varied clinical presentations ranging from involuntary hyperkinetic movements, neuromuscular involvement to cognitive and behavioral changes. Onset of symptoms is usually in adulthood. The core NA syndrome includes autosomal recessive choreoacanthocytosis and X-linked McLeod syndrome. The other subgroup includes neuroacanthocytosis with lipoprotein disorders e.g. abetalipoproteinemia (Bassen, Kornzweig disease). This symptom variation in presentation is responsible for the under diagnosis of this clinical entity. Chorea acanthocytosis is caused by mutation of 73 exon gene on chromosome 9, VPS13A which codes for Chorein [1].

Our patient had characteristic phenotype of neuroacanthocytosis. She had presented in adulthood with typical orofacial and lingual dyskinesia troubling her in eating. Due to abnormal posturing of the neck and repeated tongue protrusion she had characteristic feeding dystonia which has been described in neuroacanthocytosis [2]. Weight loss due to poor nutritional status is a major concern in these patients. Lingual and orofacial dyskinesia causes irritation of lower lip in combination with lip biting resulting in severe lower lip injury. The patient had bizarre movements involving the whole body in form of violent trunk spasms and head thrusting which were evident on walking. This typical gait is described as “rubber man” gait [3]. As the disease progresses the hyperkinetic movement disorder evolves into the hypokinetic or bradykinetic state. These patients have major disability due to combination of movement disorders like dystonia and chorea. Neuropsychiatric symptoms are prominent in neuroacanthocytosis and may appear before the movement disorder.

Seizures were absent in our patient while they are usually seen in approximately 40% patients [4]. The patients have elevated creatinine kinase levels and axonal neuropathy. The neuromuscular involvement in choreoacanthocytosis includes myopathy and axonal sensory-motor neuropathy [5]. McLeod syndrome should be considered as a differential diagnosis of choreoacanthocytosis as neuromuscular involvement is more common in this subtype of neuroacanthocytosis [6]. Hepatosplenomegaly and cardiac involvement is seen in McLeod syndrome. Panthothenate kinase–associated neurodegeneration (PKAN) an autosomal recessive disorder is another differential diagnosis. It is a rapidly progressive childhood disorder. Dystonia along with cognitive and behavior changes are the prominent clinical manifestation. Huntington's disease is the closest differential diagnosis as the phenotype includes chorea, behavior and cognitive deficits. The pattern of caudate atrophy on neuroimaging is similar in both these disorders. The genetic testing for Huntington’s disease is diagnostic.

The classical phenotype of uncommon movement disorder, neuroacanthocytosis presents with combination of abnormal movements and feeding dystonia along with self mutilating behavior and cognitive deficits in our patient. The clinical diagnosis of sporadic variant of choreoacanthocytosis is supported by classical neuroimaging and presence of acanthocytosis in the peripheral smear. Due to lack of facility for analysis of VPS13a or Chorein gene the confirmation of diagnosis could not be reached in our case. The management remains mainly symptomatic. The incapacitating movements can be reduced with typical antipsychotics and dopamine depletors like tetrabenezine. Choreoacanthocytosis progresses over 15–30 years. Long term outcome of patient is poor. Neurehabilitation and behavior therapy plays important role in functional improvement as well as maintainance of better quality of life. Our patient is maintaining well on treatment and regularly followed up in outpatient department.

CONCLUSION

Our case demonstrates the classical phenotype of neuroacanthocytosis. The variation in symptomatology causing the different phenotypes with significant overlap makes the diagnosis difficult. The important clinical differences exist among the phenotypes and should be emphasized while making the clinical diagnosis. The better recognition and understanding of the phenomenology in neuroacanthocytosis will facilitate us in diagnosis of this entity.

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Suman Kushwaha – Conception and design, Analysis and interpretation of data, Critical revision of the
article, Final approval of the version to be published
Akhila Panda – Acquisition of data, Drafting the article,
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A novel method of treating isolated unicondylar fracture of the head of the proximal phalanx: A case report

Aysha Rajeev, John Harrison

ABSTRACT

Introduction: The phalangeal fractures are common hand injuries. The unicondylar fractures of proximal phalanx are unique. They need prompt and accurate treatment to have a good functional outcome. We present a technique for managing a volar displaced (type 4) unicondylar fracture of the proximal phalanx. This provides accurate reduction and stabilization until fracture union, with minimal soft tissue tethering to allow early movement. Case Report: A 26-years-old male sustained an injury to the right index finger while playing cricket. He attended fracture clinic complaining of pain and difficulty in moving the proximal interphalangeal (PIP) joint of the index finger. The X-ray showed a unicondylar fracture of the proximal phalanx in the coronal plane, with a displaced volar fragment. The patient was treated with open reduction and internal fixation with a single Kirschner wire (K-wire) passed from volar to dorsal aspect through the fragment. The patient commenced hand physiotherapy straight away and regained full range of movements. Conclusion: We report the technique of fixing the displaced unicondylar fractures of the proximal phalanx using single K-wire passed from the volar to dorsal aspect after open reduction of the fragment through a volar approach. This technique will allow early mobilization with very minimal soft tissue dissection thus preventing stiffness.

Keywords: Unicondylar, Proximal phalanx, Type 4 fracture

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INTRODUCTION

Unicondylar fractures of the phalangeal head are relatively common injuries. They are more common in young, active patients and define a specific fracture pattern [1]. These are intra-articular fractures and four types are described [2]. Class 1 and 2 occur between the condyles (intercondylar) leading to angular deformity of the digit if displaced. Class 3 is a dorsal fragment in the coronal plane. Class 4 fractures are less common and is a volar fragment. If displaced these fractures require operative management. Intercondylar fractures may be treated successfully with closed reduction and percutaneous transverse fixation. Coronal plane fractures may require open reduction and any metalwork must not be left in the joint [3]. Accurate reduction and stabilization, allowing early motion are the key aims for achieving good results with all types of unicondylar fractures of the proximal phalanx. When stabilization is necessary, soft tissue trauma should be minimized to lessen the risk of scar formation and consequent stiffness at the proximal interphalangeal
joint. Stiffness is the most frequent and serious reported complication of unicondylar fractures [4].

**CASE REPORT**

A 26-years-old male with right dominant hand was seen in the fracture clinic following an injury to his right index finger. He worked as a computer sales representative. He sustained a hyperextension injury to his finger while playing cricket when the cricket ball hit his index finger tip. He complained about pain and difficulty in moving the proximal interphalangeal (PIP) joint. He was otherwise fit and well.

On examination, there was tenderness in the PIP joint. With the finger extended there was no angular deformity. Movement at the PIP joint was limited to 0–40 degrees of flexion. Metacarpophalangeal (MCP) and distal interphalangeal (DIP) joint movements were full. The X-ray showed a unicondylar fracture of the proximal phalanx in the coronal plane (Figure 1), with a displaced volar fragment. The patient was offered surgery after explaining the risk of stiffness and malunion.

Under general anaesthetic and tourniquet control the PIP joint was exposed through a Brunner’s incision (Figure 2). The A3 pulley was released on the ulnar side. The condylar fragment was reduced and held with a Kirschner wire (K-wire) passed from volar to dorsal through the fragment. The K-wire driver was then moved to the leading end of the wire where it protruded through the skin and was then pulled through so the trailing end of the K-wire was just under the articular surface (Figure 3). The reduction was checked with image intensifier, and full flexion of the PIP joint was achieved.

The patient was allowed to commence finger flexion from the first post-operative day. The patient was followed up in the clinic after two weeks for wound inspection and check X-rays were satisfactory (Figure 4). The patient was able to flex the PIP joint at this stage to 80°. The K-wire was removed four weeks after surgery and the patient was referred for further intensive physiotherapy. He regained full range of movements of the PIP joint after eight weeks. The X-ray showed that the fracture had healed in an anatomical position (Figure 5). He was discharged from the outpatient clinic.

**DISCUSSION**

Various methods of open reduction and internal fixation of unicondylar fractures of the proximal phalanx have been described in literature. London et al. noted that some of these fractures were stable so that they can be treated with splints and early mobilization, whereas others needed open reduction and fixation [4]. He also proposed a classification system for these fractures. McCue et al. in a review of twenty cases reported that there was no predominance of either radial or ulnar condyle involvement in these fractures [5]. They described that unicondylar fractures of PIP joint were most commonly associated with sports injuries. Their treatment protocol was to fix all twenty cases with two Kirschner wires which regained an average of 93° of PIP joint movements. The mechanism of distal unicondylar fractures of the proximal and middle phalanx were caused by a trochlear shear rather than compression as described by Soeur et al. [6].

Ramos et al. developed a protocol for isolated unicondylar fracture of the head of the proximal phalanx which included surgical fixation using lag screw
and immediate mobilization by use of a continuous passive motion and controlled active motion [7]. They used specially designed splints and Coban wrap to control the position of the digit during the first six months.

Henry et al. described a wide array of treatment options for these type of fractures of proximal phalanx, including Kirschner wires screws and plate fixation [8]. He stated that early closed reduction is successful for unicondylar fractures of the head of the proximal phalanx.

Blazar et al. in their study on fractures of proximal interphalangeal joint stated that an understanding of the anatomy, the potential for joint instability and the treatment options are essential to manage these fractures [9]. They also described various treatment options including extension-block splinting, percutaneous pinning, traction, external fixation, open reduction and internal fixation and volar-plate arthroplasty. A prompt recognition of the complexity of the injury and appropriate management are also essential for an optimal functional outcome.

The objectives of techniques of internal fixation of proximal phalangeal fractures are pain control and early functional restoration. When the fragment size permits, unstable and displaced proximal phalangeal joint fractures can be secured in an anatomic position by either a direct method through the fragments or by indirect methods (buttress) like pinning or screw fixation techniques. Percutaneous or limited open reduction and internal fixation techniques are preferred in an effort to minimize additional soft tissue trauma and scarring [10].

The classification of distal unicondylar fractures of the proximal phalanx was based on the mechanism of injury. There are four class of fractures [3]. The patient in our case had a class 4 fracture which involved a volar coronal fragment. These injuries are caused by a shearing force across an extended proximal interphalangeal joint causing a transient volar subluxation of the PIP joint with the joint extended. Class 4 fractures have the poorest final range of motion and this may be
Figure 5: Radiographs showing a well healed fracture of head of proximal phalanx (unicondylar).

due to imperfect reduction.

CONCLUSION

Our technique of fixing unicondylar fractures of the proximal phalanx used a single Kirschner wire introduced from the volar aspect to engage the fracture fragment. We felt a screw placed from dorsal surface may not gain adequate fixation in the thin volar fragment and the prominent screw head may affect joint flexion, and if removal was necessary this would require more dissection than a subcutaneous Kirschner wire.

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Author Contributions
Aysha Rajeev – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
John Harrison – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published

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The corresponding author is the guarantor of submission.

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Authors declare no conflict of interest.

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REFERENCES

Pleuropulmonary blastoma in an adult: A case report

Nidhi Paliwal, Kumud Gupta, Shalini Mullick, Ravindra K Dewan, Sandeep Katiyar

ABSTRACT

Introduction: Pleuropulmonary blastoma (PPB) is a dysontogenetic neoplasm of childhood that appears as a pulmonary and/or pleural-based mass. PPB is of three types: cystic (type I), mixed (type II), or solid (type III). It is rarely observed in adults with only a few cases reported in literature. Case Report: We present a case of a 30-year-old male with complaints of chest pain, dyspnea and right lower rib. His computed tomography scan of chest showed three multicystic lesions in right hemithorax. Cytomorphological examination of imprint smears of biopsies taken from the masses suggested malignant lesion and on histological examination, diagnosis of pleuropulmonary blastoma type II was made. Conclusion: Pleuropulmonary blastoma is a rare and aggressive malignant tumor of childhood. Familial disposition and association of PPB tumor with other childhood malignancies has been suggested in literature. Pathologically, the tumor tissue is composed of primitive blastemal cells with focal areas of malignant mesenchyme. Therapy should include surgical tumor resection and subsequent chemotherapy and radiotherapy. PPB is very rare tumor in adults and presentation as multiple lesions in one sided hemithorax has not been reported. PPB should be considered in the clinical and radiographic differential diagnosis of multicystic lesions, even when the patient is a young adult.

Keywords: Blastoma, Malignant, Pleuropulmonary

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INTRODUCTION

Pleuropulmonary blastoma (PPB) is a unique dysontogenetic neoplasm of childhood. Its primitive, sarcomatous features are analogous to those of other dysontogenetic tumors such as Wilms’ tumor, hepatoblastoma, neuroblastoma, and embryonal rhabdomyosarcoma [1]. PPB was classified into three groups by Dehner et al. in 1995 as cystic (type I), mixed (type II), and solid (type III). Type I has a more favorable prognosis than type II and III [2]. This rare and aggressive neoplasm arises from the lung, pleura, or both and is a disease of the first decade of life [3]. PPB is observed rarely in adults and clinical presentation as in our case has not been reported in literature. There is no optimal defined treatment regimen for adult cases [4].
We are reporting a case of PPB in a 30-years-old male presenting with three multicystic lesions in right hemithorax and give a brief review of literature.

CASE REPORT

A 30-year-old male patient presented with chest pain, dyspnea, and low grade fever for last six months. Two years back he had taken antitubercular drugs therapy. He was a non-smoker with no past history of any malignancy. There was no family history of any malignant lesion in his siblings or parents. Physical examination was unremarkable. His chest X-ray revealed homogenous opacity in right mid and lower zones (Figure 1). Computed tomography (CT) scan of chest showed large, multiloculated collection in right thoracic cage with a few foci of calcification in the septations. Underlying lung was collapsed. Two small loculated collections were also seen in right posterior thoracic cavity (Figure 2). The possibilities of hydatid cyst or benign teratoma were considered based on clinical and radiological presentation.

The patient underwent right thoracotomy. Intraoperatively, a large multicystic mass measuring more than 10 cm in diameter, adherent to lower lobe and diaphragm was seen. Two masses measuring 5 cm and 4 cm in diameter were also present in middle and upper lobe of right lung, respectively. Biopsies from masses were taken and sent to the pathology department.

Complete mobilization of largest mass was not possible, so it could not be excised. As facility of frozen section was not available in our hospital, only imprint smears from biopsies were made. All smears showed similar cytological picture and revealed small groups and singly present round to oval to spindle cells with scanty to moderate cytoplasm. Some of the cells were bipolar (Figure 3A). A few cells showed high N:C ratio, moderate cytoplasm with indistinct outline, vesicular chromatin and prominent nucleoli (Figure 3B). Cytomorphological features were suggestive of malignancy, however, exact typing of the lesion was not possible.

Grossly, resected tissue pieces were 2.8x2.8x0.7 cm and 3.5x3.0x2.0 cm in dimensions, multicystic, greyish white to greyish red with multiple, non-communicating cysts, filled with clear fluid and thin intervening septae along with few glistening, solid white nodules (Figure 4). The tissue was fixed in formalin and processed routinely. Histopathological examination revealed cuboidal to low columnar epithelial lined clefts, cysts and fibroepithelial polyps (Figure 5A). Mesenchymal connective tissue revealed disarray of spindle to elongated cells with mild to moderate atypia (Figure 5B). Cartilaginous islands were also present and some of them revealed hypercellularity of lacunae and focal overlapping. Occasional focus of small darkly stained angulated nuclei (blastemal-like cells) was also seen (Figure 5C). On immunohistochemistry, the lining epithelial cells of cystic spaces were positive for cytokeratin (Dako-AE1/AE3). The stromal cells revealed diffuse vimentin (Dako-V9) positivity (Figure 5D) and focal desmin (Dako-D33) positivity. A diagnosis of pleuropulmonary blastoma type II was made. One small piece of lung tissue with necrotic foci was also received which revealed a necrotizing granulomatous lesion. This focus was negative for acid-fast bacilli. Patient was

Figure 1: X-ray of chest showing homogenous opacity in right mid and lower zones, silhouetting right heart border and blunted right costophrenic angle (PA view).

Figure 2: Contrast enhanced computed tomography axial image showing multiple, well-defined, heterogeneously enhancing pleural based focal lesions in right hemithorax with enhancing internal septations within the lesions.
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**DISCUSSION**

Pleuropulmonary blastoma is a rare and aggressive malignant tumor of childhood. Because PPB has been reported in association with "pre-existing" anomalies of the lung, such as cystic adenomatoid malformation, extralobar sequestration, and bronchogenic cyst, it has been suggested that the foundation for neoplastic transformation is laid during development [1]. Some authors suggest familial disposition and association of PPB tumors with other childhood malignancies [3]. Despite its aggressive growth, PPB causes only a few mild complaints. Often presenting symptoms are fever, cough, hemoptysis, dyspnea and chest pain. The clinical picture of PPB in an adult resembles that in children [5]. In our case also, the patient presented with cough, dyspnea and fever.

About 50% of all PPB patients develop local recurrence and metastases after initial surgical treatment [6]. Common localizations are brain and spinal cord (44%), bone (24%), liver (4%), and locoregional (76%). Bilateral manifestations have been described as well [7], but presentation as multiple lesions within one sided hemithorax in an adult has not been reported till date. Atypical presentations such as ruptured cystic teratoma, pleurorrhea and pancreatic metastasis have been reported in literature [6, 8, 9]. The clinical and radiographic differential diagnosis include fungal lesions, congenital cystic adenomatoid malformation, bronchogenic cyst and postinfectious pneumatocele.

Pathologically, the tumor tissue is composed of primitive blastemal cells with focal areas of rhabdomyosarcomatous, chondrosarcomatous and liposarcomatous differentiation. Unlike pulmonary blastoma in adults, the epithelium is never malignant [3]. In our case also, blastemal cell collection was seen with normal appearing epithelium but no well defined sarcomatous component was noted. Sheets and nodules of blastemal-type cells, cystic areas and myxoid change in stroma seen in our case are also described by Hill et al. [1]. Three pathologic types of PPB, based on gross and microscopic features have been defined: type I is a purely cystic lesion that may be mistaken clinically and pathologically for a congenital lung cyst, type II is a cystic and solid lesion with areas of thickening and/or nodularity with or without a relationship to the cysts, and type III is a purely solid tumor consisting of friable, gelatinous to mucoid, lobulated tissue often accompanied by hemorrhage and necrosis [1]. In non-cystic cases, without an overt rhabdomyoblastic component, entities such as monophasic synovial sarcoma, malignant teratoma, and undifferentiated sarcoma should be considered. Immunohistochemistry plays an important role in differentiating between PPB such entities. In immunohistochemical studies; the most common findings are vimentin positivity and S-100 protein.
positivity in cartilaginous foci and desmin positivity in areas of rhabdomyoblastic differentiation. The only typical characteristic of PPB is vimentin positivity [4]. In cytogenetic studies of childhood cases, chromosomal anomalies of trisomies 8 and 2 have been detected. But as studied by Gonullu et al., in adult patients, interestingly, karyotypic abnormalities such as trisomies 8 and 2 were not demonstrated [4]. Germline mutations in DICER1 has also been described in cases of familial PPB and ovarian sex cord-stromal tumors [10]. We were not provided with the opportunity to perform cytogenetic studies in the current case.

While tumor size >5 cm and mediastinal and/or pleural invasion are poor prognostic factors, the most important prognostic factor is the total excision of the mass with clear margins. Postoperative radiotherapy is recommended in cases with incomplete resection [11]. Cyclophosphamide, doxorubicin, ifosfamide, etoposide, vincristine are commonly used agents in the treatment of PPB. The most common combination is vincristine, dactinomycin, cyclophosphamide (VAC) regimen [4]. Therapy should include surgical tumor resection and subsequent chemotherapy and radiotherapy. In a review of 50 cases of PPB in children by Priest et al., overall survival for all patients was 63% after two years and 45% after five years. Median survival was calculated and it was 15.5 months from diagnosis and 5.5 months from recurrence [6].

CONCLUSION

Pleuropulmonary blastoma is very rare tumor in adults and its presentation as multiple lesions in one sided hemithorax has not been reported. Pleuropulmonary blastoma should be considered in the clinical and radiographic differential diagnosis when hydatid cyst or benign teratoma is suspected in view of its multicystic appearance, even when the patient is a young adult.

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Author Contributions

Nidhi Paliwal – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Kumud Gupta – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

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Sandeep Katiyar – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor

The corresponding author is the guarantor of submission.

Conflict of Interest

Authors declare no conflict of interest.

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REFERENCES


Combined aortic and inferior vena cava injury

Basem Marcos, Yomayra Perez, Jennifer Matarlo, Jay A Yelon, Valerie Katz, Robert V Madlinger

ABSTRACT

Introduction: Combined injuries to the aorta and inferior vena cava are among the most severe traumatic injuries where mortality rates can approach 100%. Case Report: A 26-year-old male presented with multiple small caliber gunshot wounds to the right upper and lower back, right posterior arm and right gluteal area. He was diagnosed with large retroperitoneal hematoma on computed tomography (CT) scan. He underwent an exploratory laparotomy with exploration of a zone 1 retroperitoneal hematoma. An injury to the inferior vena cava (IVC) at the level of the left renal vein was identified and repaired by lateral venorrhaphy. Massive transfusion protocol (MTP) was activated. The patient was returned back to the operating room few hours later for recurrent bleeding. Aortic injury was identified one centimeter distal to the renal arteries secondary to ruptured pseudoaneurysm. The left renal vein was ligated for exposure. The aortic injury was repaired. The patient was resuscitated postoperatively and was discharged home without major morbidity. Conclusion: Combined inferior vena cava and infrarenal aortic injury carries a mortality of around 73%. All zone 1 retroperitoneal hematomas should be explored, with the exception of venous hematomas of the juxtahepatic vena cava. Mattox maneuver is the ideal exposure for a supramesocolic retroperitoneal hematoma and Cattell–Brash maneuver is used for inframesocolic retroperitoneal hematomas. Zone 1 retroperitoneal hematoma resulting from combined injuries to the inferior vena cava and aorta is highly lethal. Successful management requires early MTP, rapid and aggressive surgical management and application of elective vascular surgery techniques.

Keywords: Inferior vena cava injury, Aorta injury

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INTRODUCTION

Combined injuries to the aorta and inferior vena cava (IVC) are among the most severe traumatic injuries and are associated with high mortality rates. Mortality rates for such injuries can approach 100%. Adherence to
advanced trauma life support protocol, principles of
damage control surgery, and implementation of an
evidence based massive transfusion protocol (MTP) may
lead to improved survival with a decrease in major
morbidity.

CASE REPORT

A 26-year-old male was brought to the emergency
department (ED) by emergency medical services after
he was found with multiple small caliber gunshot
wounds to the right upper and lower back, right
posterior arm and right gluteal area (Figure 1). On
arrival his vital signs were: heart rate 72/min,
respiratory rate 22/min, blood pressure 100/48 mmHg
and spO2 100%. He complained of abdominal pain. On
physical examination the abdomen was soft, non-
distended and moderately tender over the mid-
abdomen without rebound, rigidity, or guarding. Bowel
sounds were present. Focused abdominal sonography
for trauma (FAST) was performed in the ED which did
not reveal any free fluid. Further diagnostic studies
included a computed tomography (CT) scan of the
abdomen and pelvis was suspicious for injuries to the
abdominal aorta and portal vein.

The patient was immediately taken for operation.
The MTP was activated in preparation for the possibility
of a major abdominal vascular injury. Massive
transfusion protocol in our institution is constituted of
pRBCs:FFPs in 1:1 ratio followed by giving single donor
platelets and 10 units of cryoprecipitate after 10 units
each of pRBCs and FFPs. The patient underwent an
exploratory laparotomy with exploration of a zone 1
retroperitoneal hematoma (Figure 2). An injury to the
IVC at the level of the left renal vein was identified and
repaired by lateral venorrhaphy. No bleeding was noted
from the portal vein or the aorta. Hemorrhage was
controlled, the abdomen was packed and left open
because of mid-gut edema and the patient was
transferred to the surgical intensive care unit (SICU) for
continued resuscitation. A few hours later, the patient
developed signs of hypovolemic shock and was
immediately returned to the operating room. The MTP
was again activated. Supraceliac aortic control was
obtained and an aortic injury was identified one
centimeter distal to the renal arteries (this was the
pseudoaneurysm that was present on the initial imaging
and missed during the first laparotomy and progressed
to subsequent rupture). The left renal vein was ligated
and divided to provide adequate exposure of the injury.
The aortic injury was repaired by primary suture
closure. The patient received a total of 53 units of
packed red blood cells, 49 units of fresh frozen plasma, 7
units of single donor platelets, and 40 units of
cryoprecipitate perioperatively. At the end of surgery,
the patient’s lactate was 9.5 mmol/L and his base deficit
was 18. Resuscitation was continued and these findings
of hypoperfusion were corrected in less than 24 hours.

The patient underwent a planned re-exploration and
abdominal washout with partial fascial closure on
postoperative day (POD)-3. The abdomen was closed on
POD-7. The patient developed acute cholecystitis and
underwent an open cholecystectomy on POD-11. A
tracheostomy was performed on POD-14. The patient
spent 23 days in the SICU, four days in the surgical ward
and he was then transferred to an acute inpatient
rehabilitation center. The patient was seen in the
surgery clinic following his discharge from inpatient
rehabilitation. He was ambulating and tolerating a diet.
His tracheostomy had been removed and the site of his
tracheostomy.

DISCUSSION

The retroperitoneum is divided to zone 1 (around the
abdominal aorta and IVC), zone 2 (around both the
kidneys), and zone 3 (in the pelvis). Zone 1
retroperitoneal hematoma is a sign of a major
abdominal vascular injury. Penetrating trauma accounts
for approximately 90% cases. The overall mortality rate
is 57%. Mortality rates range from 30% for an infrarenal
IVC injury, 50% for an infrarenal aortic injury, 60% for
an injury to the suprarenal abdominal aorta and 100%
for combined injuries to the suprarenal abdominal aorta
and IVC. Our patient had an injury to the IVC as well as
an infrarenal aortic injury, which carries approximately
73% mortality rate. Factors associated with increased

Figure 1: Plain chest and abdominal X-ray showing radio-
opaque bullet fragments.

Figure 2: Computed tomography scan of the abdomen
showing large zone 1 retroperitoneal hematoma.
mortality are presence of shock at the time of admission, bleeding without retroperitoneal tamponade, acidosis, and suprarenal injury [1].

This injury is more common in urban trauma centers than it is in military conflicts. DeBakey et al. reviewed 2,471 patients with arterial injuries during World War II. Two percent patients had abdominal arterial injuries. Rich et al. reviewed 1,000 patients with arterial injuries sustained in the Vietnam War. Only 2.9% injuries involved abdominal vessels. A thirty-year review of 5,760 cardiovascular injures seen at Ben Taub Hospital (Houston, TX, USA) found that 33.8% patients had abdominal vascular injuries. This difference is thought to be due to the increased wounded power of military weapons, delayed transport and torso body armor [2].

Patients can present clinically with one of two pictures: (i) contained hematoma with transient hypotension responding to crystalloid boluses, or (ii) free intraperitoneal rupture with marked hypotension which does not respond to fluid boluses. Patients with contained hematoma, like our patient, can remain hemodynamically normal until the hematoma is opened in the operating room. In one review, patients with a contained hematoma had a mean base deficit of 7.2, mean transfusion of 8.6 units of blood, and a survival rate of 96%. Those with free intraperitoneal rupture had a mean base deficit of 14.7, mean transfusion of 15.1 units and 43% survival [2]. In patients with penetrating abdominal trauma to the back and flank who are hemodynamically normal, CT scan of the abdomen can be obtained for further evaluation of injuries prior to surgery [3].

In cases with penetrating abdominal trauma, standard trauma principles apply. These include exploration via a midline incision, four quadrant packing and evacuation of blood and blood clots. Proximal and distal vascular control is essential and can be achieved by a variety of vascular techniques, including direct digital pressure, pressure with laparotomy pads (packing) or pressure with sponge sticks. Enteric injuries are addressed with clamps or staples. An associated ureteral injury may be treated initially by ligating the ureter during damage control surgery.

All zone 1 retroperitoneal hematomas should be explored, with the exception of venous hematomas of the juxtahepatic vena cava [4]. A left medial visceral rotation (Mattox maneuver) is the ideal exposure for a supramesocolic retroperitoneal hematoma and the right medial visceral rotation or Cattell–Brash maneuver with kocherization of the duodenum is used for inframesocolic retroperitoneal hematomas [5].

After hemorrhage control of the injured aorta or IVC is obtained, exploration of the hematoma is begun at the highest point. Lateral repair (simple primary suture repair) of the aorta and IVC should be attempted. Complex vascular repairs can be performed if there is loss of part of the arterial wall, but this is time consuming. The left renal vein can be ligated and divided. This aids in exposure of injuries at this level. If IVC repair is not possible, the IVC can be ligated. Lower extremity fasciotomies may be needed along with aggressive resuscitation. The infrarenal aorta can be ligated if it cannot be repaired with axillary bifemoral extra-anatomic bypass performed later, if the patient survives [4].

CONCLUSION

Zone 1 retroperitoneal hematoma resulting from combined injuries to the inferior vena cava and aorta is highly lethal. Management should be individualized based on the patient’s presentation. Key maneuvers are hemorrhage control and exposure. Successful management of acute traumatic injury to both the abdominal aorta and inferior vena cava requires early activation of massive transfusion protocol, rapid and aggressive surgical management, including damage control techniques and application of techniques and principles developed for elective vascular surgery.

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Author Contributions
Basem Marcos – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
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Jennifer Matarlo – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
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Guarantor
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Acute urinary retention in a female adolescent

Alberto Mendoza-Paredes, Antonio Pierre

ABSTRACT

Introduction: Acute urinary retention requires timely evaluation and management in order to prevent damage to the kidneys and urinary tract. Case Report: An 11-year-old female child came to the emergency department complaining of abdominal pain for three days and oliguria with dysuria for the last 24 hours. Physical examination showed a palpable mass in lower abdomen up to umbilical level and a bulging mass in the introitus. A Foley catheter was inserted, draining 500 mL of urine with relief of the abdominal pain. After emptying the bladder, a residual mass was palpated. Renal ultrasound showed no abnormalities and pelvic ultrasound demonstrated a large homogeneous echogenic mass in the lower uterine region, diagnosed as hematocolpos. Further surgical hymenectomy resolved patient’s symptoms. Conclusion: Acute urinary retention is relatively infrequent in children. Hematocolpos is a rare gynaecological abnormality that results from imperforate hymen. Retained blood in the vagina can cause compression of the urethra and consequent urinary retention. Hematometrocolpos is another rare cause of acute urinary retention. In the evaluation of a premenarchal adolescent with acute urinary retention and with tanner stage of development 3–4, a high index of suspicion should be placed towards finding an anomaly in the genital tract.

Keywords: Urinary obstruction, Development, Tanner

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INTRODUCTION

Acute urinary retention (AUR) is a condition that requires timely evaluation and management in order to prevent damage to the kidneys and urinary tract. Although AUR in men due to benign prostatic hyperplasia is well known and recognized, but in women and especially in children, it is rare and has mostly been described as case reports [1–3].

CASE REPORT

An 11-year-old female child, came to the emergency department (ED) complaining of abdominal pain for three days localized to suprapubic and periumbilical area, concomitant dysuria, dribbling and oligouria for the last 24 hours. The patient complained of tactile fever at home one day prior to ED visit, denied sexual activity and stated that she is premenarchal. On physical examination she was tanner stage 3 of development. A palpable mass was localized in the lower abdomen up to the umbilical level. In the costovertebral angle (CVA), a
questionable tenderness was elicited. A bulging mass was observed in the introitus. A Foley catheter was inserted which drained 500 mL of urine with relief of abdominal pain. Beta human chorionic gonadotrophin (hHCG) was negative. Complete blood count and basic metabolic profile were within normal limits. Urinalysis showed no abnormalities and urine was sent for culture and sensitivity testing. After emptying the bladder, a residual mass was palpated two fingers above the pubic symphysis. A renal ultrasound showed no abnormalities. An abdominal pelvic ultrasound demonstrated a large homogeneous echogenic mass in the lower uterine segment and the cervix (Figures 1A and 1B), most consistent with hematometocolpos. The patient was admitted to the pediatric ward and a specialist in obstetrics and gynecology was consulted. The patient was taken to the operating room where a cruciate incision was performed with evacuation of 450 mL of old blood from vagina with subsequent hysterecomy. After the surgical procedure the abdominal pain resolved, the Foley catheter was removed and the patient was able to void freely. Urine culture follow-up was negative.

**DISCUSSION**

Acute urinary retention is relatively infrequent in children. There are a variety of causes that are poorly defined in literature and they differ greatly from those seen most frequently in adults. Among the main etiologies; neurological processes, severe voiding dysfunction, urinary tract infection, constipation, adverse drug effect, local inflammatory causes, locally invading neoplasms, benign obstructing lesions and idiopathic cases are included [4].

Menarche is associated more with developmental stages rather than chronological age. It usually occurs by Tanner stage 4 of development, but it can be achieved in Tanner stage 3 [5], as in our patient.

Imperforate hymen is reported at an approximate rate of 0.1% and occurs due to the incomplete canalization of the Mullerian and the urogenital system [6]. Collection of blood in the vagina (hematocolpos) is a rare gynecological abnormality that results from imperforate hymen. However, vaginal atresia or iatrogenic injury can also result in hematocolpos [7].

Retained blood in the vagina can cause compression of the urethra and urinary retention [8–9]. It can also present as low back pain [10] or constipation [11]. In such cases thorough physical examination (sometimes difficult in this particular age group due to personal and cultural reasons) will suggest the diagnosis and an imaging test such as pelvic ultrasound or magnetic resonance imaging will be confirmatory [12].

**CONCLUSION**

Hematometocolpos is a rare cause of acute urinary retention. In the evaluation of a premenarchal adolescent with acute urinary retention and tanner stage of development 3–4, a high index of suspicion should be placed towards finding an anomaly in the urogenital tract.

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**Author Contributions**

Alberto Mendoza-Paredes – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published

Antonio Pierre – Final approval of the version to be published
Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

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REFERENCES
Rib fractures: Accidental or non-accidental

Muhammad Waseem, Evelyn Erickson

ABSTRACT

Introduction: We report an incidental discovery of multiple rib fractures in a wheezing child without a history of an injury or the presence of metabolic bone disease. As a result, the child was evaluated for the presence of non-accidental trauma. Case Report: An 11-month-old African-American child was brought to the emergency department by his father with a 2-day history of fever, cough and breathing difficulty. After receiving nebulizer treatments, the child was still wheezing. A chest X-ray was obtained which showed bilateral fractures of the ribs. No history of trauma was provided. Given the radiographic findings, Child Protective Services was contacted and a report of child abuse was made. Conclusion: Child abuse is a complex phenomenon. Any skeletal injury in young children can be due to abuse. Rib fractures are uncommon in the pliable chest of a child. When discovered, however, they raise the suspicion of a non-accidental trauma. They are often uncovered during the assessment of children who present to the emergency department for unrelated reasons. The physician’s ability to differentiate accidental from non-accidental trauma may depend on gathered information. This report emphasized the importance to evaluate for non-accidental trauma after the finding of bilateral rib fractures on a chest X-ray. Non-accidental trauma should be considered when there is evidence of injury without a history of trauma.

Keywords: Non-accidental trauma, Rib fractures, Child abuse

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INTRODUCTION

Rib fractures are uncommon in infants and children. Non-accidental trauma is a common cause of morbidity and mortality in young children, but the diagnosis is not always apparent. Most abused children present without a plausible explanation for their injuries. In the absence of a documented history of significant injury or the presence of metabolic bone disease, non-accidental trauma is the most likely presumed diagnosis. We report an incidental discovery of multiple rib fractures in a wheezing child.

CASE REPORT

An 11-month-old African-American, asthmatic child was brought by his father to the emergency department during the winter with a two-day history of fever, cough and breathing difficulty. On arrival, he was noted to be wheezing and was directly brought to the asthma room.
He had a temperature of 101.2°F, heart rate of 112/min, respiratory rate of 30/min, and an oxygen saturation of 95%. The rest of the physical examination was normal.

The medical history revealed prior episodes of wheezing in a developmentally appropriate child. No history of trauma was given. The patient lived with his father and had not attended school or daycare. There were no other siblings. Physical examination revealed no bruising, swelling, abnormal marks or other signs of trauma. His complete blood count and basic metabolic profile were normal. Subsequent laboratory investigations were as follows: serum phosphate level 5.3 mg/dL (2.7–4.5 mg/dL), serum 25-OH Vitamin D 31 ng/ml (3–67 ng/mL) and serum alkaline phosphatase 139 U/L (30–90 U/L). Normal range is given in parenthesis. A chest X-ray was obtained due to persistent wheezing despite three nebulizer treatments. No infiltrate was noted but bilateral posterior fractures of the 9th and 10th ribs were identified (Figures 1, 2). A subsequent skeletal survey confirmed the presence of bilateral healed fractures of the 9th and 10th ribs. Given the radiographic findings, Child Protective Services was consulted in the emergency department and a report of child abuse was made.

**DISCUSSION**

Child abuse is a complex phenomenon. Any skeletal injury can be due to abuse. Rib fractures are uncommon in the pliable chest of a child. The presence of bilateral rib fractures in an infant should prompt a thorough medical and social evaluation for child abuse [1]. When discovered, however, they raise the suspicion of non-accidental trauma. These findings may be uncovered during the assessment of children who present to the emergency department for unrelated reasons. Non-accidental trauma is a relatively common occurrence and fractures are the second most common presentation of child abuse [2]. Bilateral rib fractures, particularly in infants, should always raise the suspicion. Many children with non-accidental trauma have healing fractures. Multiple rib fractures are considered a marker of serious injury in children.

Often, a chest X-ray is obtained in a wheezing child with fever to rule out pneumonia. This child had an asthma exacerbation due to an upper respiratory infection and received care for his asthma. If the chest radiograph is not carefully reviewed, rib fractures may be overlooked; this is especially true in a busy emergency department. The diagnosis of rib fractures is often made by obtaining plain X-ray of the chest, as in our patient. A dedicated rib series may better define the fracture including the age and location. As one might expect, location of rib fractures may provide information regarding the mechanism of injury. The posterior fractures occur due to the mechanical stress at the costovertebral junction as the child is grabbed and shaken. A detailed history of how the injury occurred is, therefore, essential. When non-accidental trauma is being considered, it is imperative to evaluate the child.

![Figure 1: Chest X-ray of an 11-month-old infant showing bilateral rib fractures.](image1)

![Figure 2: X-ray showing bilateral posterior fractures of the 9th and 10th ribs in an 11-month-old infant.](image2)
for other fractures; a complete skeletal survey may uncover additional injuries. It is uncommon to detect these fractures in the acute phase as they are better seen when callus formation is advanced. A follow-up chest X-ray may, therefore, provide useful information in children with suspected non-accidental injury and may improve detection [3, 4].

The physician’s ability to differentiate accidental from non-accidental trauma may depend on gathered information. It is difficult to ascertain the cause of rib fractures when no plausible history to explain the injury is offered. Often, the trauma is only recalled after the fracture is identified. Generally, most rib fractures in non-accidental trauma are the consequence of thoracic compression [3]. Posterior rib fractures are considered to have a strong association with non-accidental trauma. Overall, a rib fracture had a positive predictive value of 95% for the diagnosis of non-accidental trauma [3]. The compliance of the rib cage may allow significant injury to occur with little apparent external signs of trauma [3]. Because of the delay in clinical presentation in such cases, healing fractures with callus are more prevalent than acute fractures [3]. Posterior rib fractures, in particular, have a well-known association with non-accidental injury [3].

Table 1 gives other causes of rib fractures in children [3, 5]. Rib fractures may be associated with birth trauma. If birth related injuries are not identified initially, they may later be attributed to non-accidental trauma [3]. Rib fractures may occur with cardiopulmonary resuscitation but the possibility of non-accidental injury should be considered [3]. In the presence of unexplained fractures, causes of bone fragility such as osteogenesis imperfecta and rickets must also be considered [3, 5–7]. The patients with rickets, laboratory evaluation usually reveals low to normal serum calcium, low serum phosphorus, and an elevated alkaline phosphatase level. The physical signs of rickets include growth retardation, metaphyseal flaring, prominence of the costochondral junctions (rachitic rosary) and frontal bossing [8]. Our patient did not have any physical findings consistent with the diagnosis of rickets. There was also no history of trauma or cardio pulmonary resuscitation.

Table 1: Important Causes of Rib Fractures in Children

<table>
<thead>
<tr>
<th>Trauma</th>
<th>Metabolic Bone Diseases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Accidental (rare)</td>
<td>Osteogenesis Imperfecta</td>
</tr>
<tr>
<td>Non-Accidental</td>
<td>Rickets</td>
</tr>
</tbody>
</table>

CONCLUSION

This case report provides an approach in the evaluation of child with evidence of injury but no history of trauma. In the absence of a history of a significant accidental trauma, evaluation of non-accidental trauma should be performed. Determining whether a fracture is due to accidental or non-accidental trauma can be challenging, but the future safety of the child, depends on a timely diagnosis and intervention.

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Author Contributions
Muhammad Waseem – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Evelyn Erickson – Acquisition of data, Drafting the article, Critical revision of the article, Final approval of the version to be published

Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

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REFERENCES

An unusual cause of hypertension

Muhammad Waseem, Evelyn Erickson

ABSTRACT

Introduction: Hypertension is not common in infants and young children. The etiology of hypertension in this age group may be different from older children and young adults. Hypertension may be associated with intussusception in young children. Case Report: A previously healthy 17-month-old, ill-appearing and dehydrated girl was brought to the emergency department with fever and vomiting for three days. The vomiting was non-bilious and non-projectile. Her temperature was 103°F, heart rate was 126/min, respiratory rate was 32/min, oxygen saturation 98% and blood pressure 122/77 mmHg. She had dry mucous membranes. The abdomen was soft with mild tenderness in the left upper quadrant and no palpable masses. She was diagnosed with ileocolic intussusception. Conclusion: Intussusception should be considered a diagnostic possibility in infants who have a history of vomiting and in whom lethargy and hypertension are the presenting features. This case report demonstrates the importance of measuring blood pressure in ill-appearing children.

Keywords: Hypertension, Intussusception, Diagnostic features

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INTRODUCTION

Hypertension in children is defined as blood pressure measurements above the 95th percentile for age, gender and height of the patient. Blood pressure is not routinely measured in infants and young children in the emergency department, though measurement should be obtained in ill appearing young children. Unlike adults and adolescents, hypertension in an infant or young child is usually indicative of an underlying condition, therefore, a careful search should be conducted. Renal disorders and coarctation of the aorta are the two most common causes of hypertension in young children. The association of hypertension and intussusception has been described in the past, being characterized as transient and usually resolving after the reduction of intussusception.

CASE REPORT

A previously healthy 17-month-old girl was brought to the emergency department with fever and vomiting for three days. The vomiting was non-bilious and non-projectile. She had no diarrhea but parents reported her crying more than usual for last two days. On the day of presentation, her parents also noted that “she was less active”. Her parents denied sick contacts and any travel history. Her immunization was current. There was no family history of hypertension.
On arrival at the emergency department she was dehydrated and ill-appearing. Her temperature was 103°F, heart rate was 126/min, respiratory rate was 32/min, oxygen saturation was 98% and blood pressure was 122/77 mmHg. She had dry mucous membranes. There were no meningeal signs. Pupils were equal and reactive, and extra-ocular movements were intact. Her chest was clear. The abdomen was soft with mild tenderness in the left upper quadrant. There were no palpable masses. Her initial stool was negative for occult blood. There was no peripheral edema. No skin lesions or rashes were noted. There were no focal abnormal findings on neurological examination.

Laboratory studies showed a white blood cell count of 8.9x10⁹/mm³ with a normal differential count and a hematocrit of 33.2%. Her urinalysis showed specific gravity of 1.043 and 3+ ketones. Serum biochemistries revealed sodium 141 mEq/L, potassium 4.6 mEq/L, chloride 107 mEq/L, CO₂ 19 mEq/L, urea 15 mg/dL, creatinine 0.6 mg/dL, glucose 74 mg/dL and calcium 9.3 mg/dL. Due to persistent vomiting, a plain X-ray of abdomen was obtained, which showed non-specific pattern and paucity of bowel gas (Figure 1).

The patient was initially treated with intravenous fluids to correct dehydration. Her clinical condition improved after hydration but blood pressure remained 120/76 mmHg. An abdominal ultrasound was obtained which showed a normal urinary tract and intussusception in the mid transverse colon (Figure 2). Subsequently, a barium enema was performed which successfully reduced the ileocolic intussusception. Her blood pressure returned to normal after reduction of the intussusception. She was discharged home in stable condition two days after the procedure.

**DISCUSSION**

The differential diagnosis of a child with fever, lethargy and vomiting is broad. In addition, the presence of hypertension in a young child may make this list more extensive. Since hypertension is not a common problem in a pediatric emergency department, presence of hypertension in a young child may pose a diagnostic challenge.

Unlike adults and adolescents, hypertension in an infant or young child is usually indicative of an underlying condition, therefore a careful search should be conducted. Emergency physicians are often the first to evaluate these children. It is important to recognize the underlying causes of hypertension in this age group.

Hypertension in children is defined as blood pressure measurements above the 95th percentile for age, gender and height of the patient. Standard nomograms, based on the above factors, are necessary for the interpretations of blood pressure values. Inappropriate cuff size is the most common cause of hypertension in children, thus selection of an arm cuff of the right size is necessary for accurate measurement of blood pressure [1]. An appropriate cuff size should have an inflatable bladder width which is at least 40% of the arm circumference at a point midway between the olecranon and the acromion; and cuff bladder length should also cover 80–100% of the circumference of the arm [2]. Recommended cuff sizes are as follows: neonates (2.5 cm), infants (5 cm), 1–8 years (9 cm) and 9–14 years (12.5 cm).

This case report raises the question whether blood pressure is measured routinely in children less than three years of age. Generally, blood pressure is not routinely measured in infants and young children in the emergency department [3, 4]. However, blood pressure

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**Figure 1:** Plain abdominal X-ray showing non-specific pattern and paucity of bowel gas.

**Figure 2:** Abdominal ultrasound showing intussusception in the mid transverse colon.
measurement should be obtained in all ill appearing young children.

Essential hypertension is very rare in children and should be considered only after exclusion of other causes. Renal disorders and coarctation of the aorta are the two most common causes of hypertension in young children. Blood pressure should be obtained in the upper and lower extremities to rule out coarctation of the aorta. In addition, a transient rise in blood pressure may be seen in the presence of stress, crying or pain. Therefore, the diagnosis of hypertension in a child should be made only after resolution of these causes.

Hypertension may be associated with intussusception. The association of hypertension and intussusception has been described in the past, but only a few case reports exist in literature [5, 6]. Hypertension is transient and usually resolves after reduction of intussusception. Therefore, intussusception should be considered a diagnostic possibility in infants who have a history of vomiting, and in whom lethargy and hypertension are the presenting features. This case report highlights the importance of measuring blood pressure in ill-appearing children.

CONCLUSION

The association of hypertension and intussusception has been described in the past. Intussusception should be considered a diagnostic possibility in infants who have a history of vomiting and in whom lethargy and hypertension are the presenting features. This case report demonstrates the importance of measuring blood pressure in ill-appearing children in the emergency department.

********

Author Contributions
Muhammad Waseem – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Evelyn Erickson – Acquisition of data, Drafting the article, Critical revision of the article, Final approval of the version to be published

Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

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REFERENCES

A floppy infant

Muhammad Waseem, Joel Gernsheimer, Tae K Park, Fernando Jara, Evelyn Erickson

ABSTRACT

Introduction: Infant botulism is a relatively uncommon but potentially life threatening cause of a septic appearing or lethargic infant. Case Report: A 6-week-old male infant presented to the emergency department with a history of poor feeding and fever for several days. His parents reported that he had been “more sleepy than usual” and had a weak cry. He had not passed any stool for five days. He was receiving a topical home herbal remedy for whitish lesions in his mouth. The rest of his review of systems and past medical history was non-contributory. On arrival to emergency department, he was ill appearing and lethargic. His vital signs were: temperature 101°F, heart rate 152/min, respiratory rate 44/min and oxygen saturation 99%. He had poor muscle tone and generalized weakness. He was diagnosed with infant botulism. Conclusion: It is extremely important that the diagnosis of infant botulism be suspected and appropriately treated when any infant presents with progressive weakness. Since infant botulism is a treatable condition, prompt diagnosis is therefore important in reducing morbidity and mortality.

Keywords: Infant botulism, Clostridium botulinum, Weakness

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INTRODUCTION

Infant botulism is a relatively uncommon but, potentially life threatening cause of a very ill appearing infant. We present the case of a six week old infant who presented to the emergency department (ED) with progressive neurological weakness that ultimately required ventilatory support. The differential diagnosis of weakness in the young infant is discussed and the pertinent literature is reviewed.

CASE REPORT

A 6-week-old male infant presented to the emergency department with a history of poor feeding and fever for several days. His parents reported that he had been “more sleepy than usual” and had a weak cry. He had not passed any stool for five days. He was receiving a topical home herbal remedy for whitish lesions in his mouth. The rest of his review of systems and past medical history was non-contributory.

On arrival in the emergency department, he was ill appearing and lethargic. His vital signs were: temperature 101°F, heart rate 152/min, respiratory rate
44/min and oxygen saturation 99%. His pupils were both equally dilated but reacted to light. His tympanic membranes were normal. Examination of his pharynx was normal. No whitish lesions were seen in his mouth and it is not known what the reported whitish lesions were, although it was speculated that they could have been from candidiasis. His neck was supple. His chest was clear with good bilateral breath sounds. The cardiovascular examination was normal. His abdomen was soft and non-tender, but bowel sounds were diminished. The neurological evaluation revealed that he was lethargic with a weak suck, cry and gag reflex. He had bilateral weakness of his facial muscles. He had poor muscle tone and weakness of his extremities. The deep tendon reflexes could not be elicited. His complete blood count, serum chemistries, urine analysis, chest X-ray and head CT scan were all normal. Lumbar puncture was performed and analysis of his cerebrospinal fluid was normal.

While being monitored in the emergency department, it was noted that he had frequent episodes of oxygen desaturation and apnea. He was intubated and placed on a ventilator. The diagnosis of infant botulism was suspected and subsequently was confirmed by stool studies. *Clostridium botulinum* spores were seen in his stool and botulinum toxin was detected in samples of his stool. He was given intravenous botulism immune globulin. After receiving the immunoglobulin, this infant improved gradually over a period of three weeks. He was able to breathe on his own, and was extubated two weeks after receiving the immunoglobulin therapy. He was discharged without any evidence of residual neurological deficits three weeks after his initial presentation. Unfortunately, this patient was lost to clinic follow-up, and we were unable to contact the family to find out about the patient’s current status.

**DISCUSSION**

The diagnosis of infant botulism should be strongly suspected in any infant with an acute onset of weakness in sucking, swallowing or crying, ptosis, inactivity and constipation. However, because infant botulism is an uncommon disorder it is often missed, leading to disastrous consequences.

**Pathophysiology and Epidemiology:** Botulism is a rare but potentially fatal paralytic disorder caused by a neurotoxin produced by *Clostridium botulinum*. This toxin, which is one of the most lethal poisons, causes an irreversible block of stimulation induced pre-synaptic cholinergic transmission. The toxin mainly affects the peripheral cholinergic nervous system. Because it does not affect adrenergic neural transmission and it does not readily cross the blood brain barrier. Botulism can be acquired in multiple ways, such as ingestion of spores that colonize the gastrointestinal tract and produce the toxin, ingestion of contaminated food that already contains the toxin such as sea food, sausages and canned foods, and infection of a wound by *Clostridium botulinum* which then produces the toxin in the wound. An important example of this is the injection of contaminated “Black Tar Heroin” [1, 2].

Infant botulism is due to colonization of the gastrointestinal tract of the infant by *Clostridium botulinum* that then produces the neurotoxin which spreads throughout the body via the circulation. Infant botulism was first recognized in 1976, and since then many cases have been reported in the United States making it the most frequently recognized form of botulism [1–3].

Infant botulism affects infants between one week and one year of age. Most cases occur within the first six months of life with a peak incidence at 3–4 months of age [3]. The majority of cases in the United States probably are caused by ingested spores that are present in dust that becomes contaminated by activities such as construction. The soil in some states such as Pennsylvania, Utah and California are particularly rich with *Clostridium botulinum*. Although cases of infant botulism from ingestion of *Clostridium botulinum* spores in raw honey or home canned foods have been reported, this is less common than previously thought [4]. It was initially postulated that the home made remedy that this baby was given may have contained raw honey or corn starch that was contaminated with *Clostridium botulinum* spores, but the parents denied that these ingredients were in this home remedy and we were unable to get a sample of it to test. It is much more likely that the infection in this infant was from *Clostridium botulinum* spores in the soil that were released into the air from local construction, and entered the baby’s gastrointestinal tract. At the time of this case, there was a lot of construction being done in the South Bronx, where this infant’s family lived. Ingestion of spores that are released into the air from soil due to construction is the most common method of infants developing botulism in the northeastern parts of the United States.

The incubation period for Botulism is thought to be at least three days. There are eight *Clostridium botulinum* toxin types. The majority of the infant botulism cases are caused by type A and B [4]. Interestingly, breast fed infants appear to be protected from botulism.

**Table 1: Important Causes of Weakness in an infant**

<table>
<thead>
<tr>
<th>Sepsis</th>
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<tbody>
<tr>
<td>Meningitis/encephalitis</td>
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<tr>
<td>Hypothyroidism</td>
</tr>
<tr>
<td>Pompe disease (Glycogen storage disease type II)</td>
</tr>
<tr>
<td>Electrolyte disturbance</td>
</tr>
<tr>
<td>Spinal muscular atrophy</td>
</tr>
<tr>
<td>Neonatal myasthenia gravis</td>
</tr>
<tr>
<td>Botulism</td>
</tr>
<tr>
<td>Guillain-Barré syndrome</td>
</tr>
<tr>
<td>Congenital myotonic dystrophy</td>
</tr>
</tbody>
</table>
Clinical Presentation: The clinical presentation of infant botulism includes progressive neuromuscular weakness which can be mild to severe. This may be misinterpreted on examination as lethargy. Cranial nerves are affected first by the muscles of the trunk, extremities and diaphragm [4]. This may cause respiratory failure. Lethargy and poor feeding are often the initial presenting symptoms of infant botulism [1]. Constipation and weak cry are other historical features. Occasionally, a history of ingestion of raw honey or canned food may be present.

This initial presentation is followed by progressive descending weakness and hypotonia, so that the infant appears to be “floppy”. Bulbar involvement, often but not always, presents with poor sucking and gag reflexes, dilated pupils with poor response to light and accommodation, decreased eye movement, ptosis and facial paralysis. Absent deep tendon reflexes, especially with type B, toxin often occurs [5]. Occasionally, young infants may present with only a history of poor feeding followed by rapid collapse or deterioration [6].

Differential Diagnosis: The differential diagnosis of the weak and floppy infant is extensive (Table 1) and includes both neurologic and systemic diseases such as sepsis, hypothyroidism, ingestions and metabolic disorders. Because many disorders can mimic infant botulism, which is a relatively rare condition, it is often not considered initially and is then missed [6].

Sepsis is the most common diagnosis that mimics infant botulism. Most patients with infant botulism are usually afebrile. In addition, sepsis does not have cranial nerve and other neurologic findings that are often present in patients with botulism. Electrolyte disturbances, including hypoglycemia may cause lethargy and weakness.

Several disorders that cause neuromuscular weakness deserve special mention. Tick paralysis is caused by a neurotoxin secreted by a wood or dog tick that prevents liberation of acetylcholine at the neuromuscular junction. The patient usually presents with ataxia and then develops a rapidly progressive ascending flaccid paralysis that can cause respiratory failure and death. Bulbar findings, including dysphagia, dysarthria, facial paralysis and ocular muscle weakness can occur late in the course of this illness as compared to botulism where bulbar findings occur early and there is descending rather than ascending paralysis. Whenever a patient presents with rapidly progressive paralysis, tick paralysis should be suspected and a careful search should be made for the presence of a tick which should then be removed if found, and this removal often results in rapid improvement.

Guillain-Barré syndrome is another differential diagnosis. It presents as a progressive symmetrical ascending paralysis which starts in the lower extremities. Its progression is comparatively slower than tick paralysis. Cranial nerves are rarely affected, although a Miller-Fisher variant causes facial paralysis. The pupils are not affected. Lumbar puncture shows cells and high levels of protein in the CSF, whereas CSF analysis is normal in botulism.

Poliomyelitis, which is now rare but may occur in unvaccinated children, presents with high fever, meningeal signs, asymmetrical weakness and lymphocytosis in the CSF.

Myasthenia gravis is the most common disorder of the neuromuscular junction in children. It can occur as transient neonatal myasthenia gravis in infants who are born to mothers who have myasthenia or as congenital myasthenia. Infants with myasthenia have generalized weakness and hypotonia; however, deep tendon reflexes are present. Facial weakness and bulbar paralysis may often occur causing poor suck and swallowing and a weak cry. Ptosis may also be seen. Respiratory failure necessitating ventilatory support may occur. Myasthenia usually responds well to treatment with anticholinesterase inhibitors, such as neostigmine or edrophonium.

The diagnosis of infant botulism is clinical. Treatment should not be delayed pending laboratory confirmation. Although not pathognomonic and not always present early in the disease, electromyographic findings consistent with infant botulism strongly support this diagnosis in the presence of appropriate clinical setting. A clinical diagnosis is supported by the identification of C. Botulinum spores in the stools and confirmed by the identification of the toxin in the stool. Serum samples are often negative in patients with infant botulism. Our patient had both spores and toxin in his stool. Serum testing was not available at our institution. Toxin may be detected in contaminated food, if a specific food is involved.

Management: Supportive care, especially ventilatory support as needed, with very close monitoring is paramount in management of botulisms. Botulism Immune Globulin IV (BIG-IV) which is a botulinum antitoxin derived from humans is very safe and effective. It should be administered immediately in the presence of a reasonably certain clinical suspicion. Botulism Immune Globulin Intravenous (Human) (BabyBIG, USA) was administered at a dose of 50 mg/kg to our infant, which at that time was the recommended dose. It should be noted that since March of 2012, the recommended dose of BabyBIG is 75 mg/kg. BabyBIG interrupts the progression of weakness by blocking the accumulation of toxin in the nerve terminals. It reduces complications, relapses, length of intubation and hospitalization. Antibiotics have not been shown to assist in the treatment of infant botulism. In fact, in the past it was recommended that antibiotics should not be used in infant botulism because lysis of C. botulinum in the gut would release more toxins into the gut and then into the circulation. However, BIG-IV therapy destroys all of the toxins in the gut and will block absorption of toxin for at least six months. If needed antibiotics can then be safely used to treat secondary infection. Antibiotics and debridement, as needed, are definitely indicated to treat wound botulism. Penicillin or metronidazole can be used, but aminoglycosides should not be used as they worsen the effects of botulinum toxin [6]. Given the age and septic appearing presentation, many of these infants usually receive broad spectrum antibiotics.
CONCLUSION

We presented a case of an infant with botulism. It is extremely important that the diagnosis of infant botulism be suspected and appropriately treated when any infant presents with progressive weakness. Since infant botulism is a treatable condition, prompt diagnosis is very important in reducing morbidity and mortality. Early recognition allows expeditious and appropriate treatment, which saves lives.

********

Author Contributions
Muhammad Waseem – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Joel Gernsheimer – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
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REFERENCES
Intimal angiosarcoma of the thoracic aorta

Michelle Forman, Michael E Mulligan

ABSTRACT

Introduction: Sarcomas of the great vessels are uncommon, with aortic being the rarest. Only 30 cases of true intimal aortic sarcomas (IAS) are documented. They tend to occur in the abdominal aorta, with less common occurrences in the thoracic aorta. Their growth patterns, predispose them to a propensity for metastases and cause embolic phenomenon. Case Report: A 58-year-old male presented with chest pain and dyspnea and was evaluated for pulmonary embolus and coronary artery disease. Computed tomography angiography (CTA) demonstrated no pulmonary emboli; however, there was severe atherosclerosis/thrombosis of the aortic arch. The process extended centrally, nearly filling the entire lumen. The surgery consultant advised anticoagulation and strict blood pressure control, recommending that the patient come to the outpatient department for surgery. Due to personal reasons, the patient failed to return at the recommended time. Three months after initial presentation the patient was admitted for surgical replacement of the aorta. The surgeon reported the aorta as “chock-full of fibro-fatty material nearly obstructing its course”. The pathology report was aortic sarcoma of intimal origin. Conclusion: Aortic sarcomas are rare tumors, with the intimal subtype in the thoracic aorta being even rarer. Delay in diagnosis of these tumors often occurs, since the imaging features are nearly identical to atherosclerotic disease. Since atherosclerotic disease is clearly more frequent than intimal sarcoma of the aorta, it is not difficult to understand that this diagnosis is not usually made until after surgical resection or at autopsy. At presentation, nearly all of the patients have metastatic disease.

Keywords: Aorta, Sarcoma, Computed tomography

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INTRODUCTION

Primary sarcomas of the great vessels (aorta, pulmonary artery and vena cava) are extremely uncommon tumors, with aortic sarcomas being the rarest type; about 26% occurring in aorta compared to about 37% each for pulmonary and vena caval sites. They are sub-classified as either mural or intimal tumors, with intimal tumors characterized as poorly differentiated on histology [1, 2]. Latest reviews, using the strict histological definition of intimal sarcoma reported that only about 21–30 cases of true intimal aortic sarcomas (IAS) have been documented, with the mean age of presentation being 62.2 years [3, 4].

Here, we present a case of undifferentiated intimal aortic sarcoma which is unusual both in location and presentation. This patient’s tumor was located within the thoracic aortic arch, and he presented with chest
pain, instead of the more typical secondary symptoms of embolic phenomena.

**CASE REPORT**

The patient was a 58-year-old African-American male with a past medical history significant for type two diabetes mellitus. He presented with chest pain and dyspnea on June 4, 2010 and was subsequently evaluated for a pulmonary embolus as well as coronary artery disease. Cardiac enzymes were negative for indication of ischemic changes. Computed tomographic angiography (CTA) of the chest demonstrated no pulmonary emboli; however, the imaging revealed severe supposed atherosclerosis/thrombosis of the aortic arch extending to the descending thoracic aorta, stopping at the level of the diaphragm. The process extended centrally, nearly filling the entire aortic lumen. The adrenal glands were free of involvement (Figures 1–2).

The vascular surgery consultant placed the patient on anti-coagulation therapy (warfarin 5 mg) and strict blood pressure control (metoprolol 50 mg and lisinopril 20 mg), recommending that he return as an outpatient for surgery on the aortic arch and descending thoracic aorta. Due to personal and other preoperative medical reasons (full mouth dental extraction), the patient’s presumed elective aortic surgery was delayed until September 2010.

As a result of chest pain, he visited our emergency department (ED) twice in the month of August 2010 and each time was ruled out for cardiac ischemia. Both times he was continued on his regimen of anti-coagulation and blood pressure control. A computed tomography (CT) scan was performed on both ED visits, demonstrating a new right adrenal mass (2x2 cm) that was not present on initial CT scan, in addition to the previously identified severely diseased aorta (Figure 3). The second time he presented to the ED, he was admitted for one week.

Three months after initial presentation, on September 23, 2010 the patient was admitted for surgical replacement of the descending thoracic aorta with a tube graft utilizing a right axillary artery cannulation site and left femoral artery and vein cannulation site. The surgeon reported the thoracic aorta as “chock-full of fibro-fatty material nearly obstructing its course, blending to a more normal appearing aorta at the diaphragm and in the proximal aortic arch”.

The pathology report described the specimen as “multiple fragments of opaque, yellow-tan to pink-gray friable soft tissue in aggregate measuring 6.2x5.2x3.4 cm. Sectioning revealed markedly friable, partially laminated, yellow-white to gray-brown cut surfaces”. The final pathology report unexpectedly revealed undifferentiated aortic pleomorphic sarcoma of intimal origin (Figure 4).

The patient was discharged from the hospital two weeks later. However, he returned to the ED after one week, complaining of neurologic symptoms (confusion, aphasia) and was admitted on October 13, 2010. CT scan and magnetic resonance imaging (MRI) of the brain demonstrated two intra-axial masses (4x3 cm in left temporal region and 2x2 cm in right occipital region). CT scan of the chest, abdomen and pelvis showed increased size of the right adrenal mass (3x2 cm), a new left adrenal mass (1x1 cm), indication of tumor thrombus in the IVC, and a peripherally enhancing paravertebral soft tissue mass involving the musculature which, in hindsight, was present on the initial CT of the chest from June 2010 (Figures 5–8). A positron emission tomography (PET) performed a few days later showed increased metabolic activity in the above lesions as well as in the T3 and T4 vertebral bodies. After consultations with medical oncology and radiation oncology specialists, a decision was made to first treat the brain lesions with external beam radiation therapy (XRT). A course of systemic chemotherapy was planned to follow. The brain lesions were treated with 6 mV photon therapy for a total of 30 gray in 10 fractions. Two weeks after completing the initial XRT, because of worsening pain, the patient had his paraspinal lesion treated with 6 mV photon therapy for a total of 25 gray in 5 fractions. Finally in mid December 2010, he was able to begin three cycles of chemotherapy with gemcitabine and taxotere.

With complaints of pain, nausea and vomiting from the metastatic disease and subsequent treatment, he was admitted multiple times in the following months. November 2010, January 2011, February 2011, and March 2011 admissions involved multiple follow-up imaging studies, all showing progression of the bilateral adrenal masses, invasion of the inferior vena cava (IVC), paravertebral soft tissue mass, and vertebral body involvement (Figures 9–10). New findings included right lower lobe pulmonary emboli. A follow-up MRI of the spine in March 2011 showed epidural metastases at T3, where he also had bony disease. His MRI of the brain in March 2011 showed marked decrease in size of the brain metastases and no new foci of metastasis.

Early in presentation, the patient already demonstrated metastatic disease. Metastatic disease was confirmed in a soft tissue lesion on the patient’s back, again with pathology of metastatic undifferentiated pleomorphic sarcoma/malignant fibrous histiocytoma.

As of March 2011, the patient’s diagnosis was stage IV metastatic, undifferentiated, pleomorphic sarcoma. He received three cycles of the chemotherapy regimen of gemcitabine and taxotere from December 2010 through February 2011, as well as supportive treatment for the neurological symptoms from the temporal and occipital lobe brain metastasis but died in July 2011 at hospice care.

**DISCUSSION**

Aortic sarcomas are rare tumors, with the intimal subtype occurring in the thoracic aorta, as the case reported here, being even more uncommon. Delay in diagnosis for these tumors often occurs as the imaging
features appear nearly identical to atherosclerotic disease. Seeing that atherosclerotic disease is clearly a more frequent diagnosis than intimal sarcoma of the aorta, it is not difficult to understand that this diagnosis is not usually made until after surgical resection or at autopsy.

Sarcomas of the aorta tend to occur mostly in the abdominal aorta, with less common occurrences in the thoracic aorta. For example, in one case series, four out of 21 cases of sarcoma presented in the chest. By definition, in contrast to the mural aortic sarcomas, intimal aortic sarcomas actually grow within the vascular lumen. Because of this growth pattern, they have a greater propensity for fragments to dislodge and be carried in the blood stream and metastasize [5]. Additionally, these tumors commonly present with sequelae from embolic phenomenon with symptoms ranging from absent peripheral pulses to mesenteric occlusion [1–4, 6, 7]. In one case series of 11 patients, all the patients with aortic sarcoma, died within 16 months of diagnosis [1].

Due to the rarity of this disease, there have been no randomized trials for the definitive treatment of IAS, with the therapies that are currently used being based solely on observational studies. Definitive treatment methods for IAS have not been delineated. The literature suggests that resection of the affected portion
of the aorta with placement of a graft and subsequent chemotherapy and radiation is the best approach [2]. Endarterectomy is also a reported treatment choice [4]. At presentation, nearly all of the cases of IAS have evidence of metastatic disease.
CONCLUSION

Our case of intimal aortic sarcoma illustrated a number of interesting facts. First, the tumor may occur in the thoracic aorta, more specifically at the aortic arch, which is an uncommon location. Second, the clinical presentation may not be the classic one of symptoms from embolic phenomenon; instead patient may present with symptoms of chest pain likely from the primary tumor. Third, patients with intimal sarcoma of the aorta tend to die shortly after presentation. However, despite the evidence of metastatic disease early in presentation, patient may lived for many months after diagnosis and treatment.

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Michelle Forman – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Michael Mulligan – Conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published

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REFERENCES
Neuroradiological imaging features of infratentorial cranial fossa tumors in a child

Muhammad Yunus Amran, Meryana Pauline, Andi Kurnia Bintang, Muhammad Akbar

CASE REPORT

An 11-year-old boy was admitted to our hospital with the chief complaint of a generalized tonic seizure which was preceded by projectile vomiting, six hours prior to his admission. There was no previous history of such complaint. The patient’s right arm and right leg were rigid and extended while his left arm and left leg were rigid and flexed. Five years before admission, the patient had complained of chronic headaches. One year before admission, the patient had complained of blurred vision in his left eye followed by the same complaint in the right eye. The blurred vision became progressively chronic, accompanied by diplopia. Three months before presenting to us, he demonstrated truncal ataxia. The patient had no problems with either urination or defecation. There was no history of fever, growth disorders or developmental disorders. Neurological examination showed neck stiffness. His pupils showed isochore mydriasis with decreased light reflexes and papilledema; the patient's visual acuity in both left and right eye was 1/300 with convergent strabismus in the left eye, and left peripheral facial paresis. On motor examination, we found dysmetria and disdiadochokinesia in the extremities of the left side as well as hypotonia on both the sides. Sensory and autonomic functions were within normal limits. Routine and blood chemistry analyses revealed the following results: Hemoglobin 12.5 g/dL, hematocrit 38.3%, WBC counts 8x10^3/mm^3, platelet count 3.13x10^5/mm^3, blood sugar (random) 156 mg/dL, SGOT 26 IU/L, SGPT 53 IU/L, urea 15.53 mg/dL, creatinine 0.32 mg/dL, sodium 135 mmol/dL, potassium 4.2 mmol/dL and chloride 101 mmol/dL. Axial head computed tomography (CT) scans of both noncontrast and contrast (with administration of intravenous gadolinium (Gd-DOTA)) were performed. The CT scan revealed a mass in the infratentorial cranial fossa which suggested a medulloblastoma with a differential diagnosis of an ependymoma (Figure 1A–B). After post-contrast the mass density was enhanced. This result suggested an ependymoma with non-communicating hydrocephalus (Figure 1C–D). The patient was scheduled to undergo ventriculoperitoneal (VP) shunting to relieve the increased intracranial pressure due to mass effect from the tumor in the fourth ventricle. Subsequently, an magnetic resonance imaging (MRI) scan was performed with and without administration of intravenous Gd-DOTA. The MRI scan showed a round lesion with a clear border, irregular edges, hyper intensity in the axial and sagittal T1-weighted images, and hypointensity in the T1WI noncontrast images, (Figure 2A–B) as well as hyper intensity in the T1WI post–contrast images (Figure 2C–D). Simultaneously, T2WI revealed hyper intensity, heterogeneous density and ring/rim enhancement, which is consistent with the description
Figure 1: Axial head computed tomography scan images (A, B) Noncontrast showing, a lesion with hyper density and calcified spots, demarcated, with a density of 44.2 Hu in the infra tentorial fossa pressing on the anterior aspect of fourth ventricle causing dilatation of the lateral and third ventricles and, (C, D) Post-contrast with intravenous gadolinium (Gd-DOTA) showing, a mass in the infra tentorial fossa, which is suspected to have arisen from the fourth ventricle. The mass is lobulated and round shaped, demarcated with irregular edges and enhanced density of 54.7 Hu post-contrast, causing dilatation of the lateral and third ventricle.

Figure 2: (A) Axial and (B) Sagittal head, Magnetic resonance imaging (MRI) scan images (noncontrast) post VP-shunting showing a round lesion with clear borders, irregular edges, size 4.87x5.43 cm in the area of the fourth ventricle and mass with hypo intensity on T1WI; (C) Axial and (D) Sagittal head MRI scan images (with contrast) post VP-shunting showing a round lesion with clear borders, irregular edges, size 4.87x5.43 cm in the area of the fourth ventricle and mass with hyper intensity on T1WI.

Figure 3: (A, B) Axial head magnetic resonance imaging scan T2-weighted image; post VP-shunting, showing a mass with hyper intensity and heterogeneous density on T2WI signal, especially at the edge (ring/rim enhancement) which is consistent with the description of an ependymoma, (C) Sagittal head MRI scan T2-weighted image, post VP-shunting, also showing a mass with hyper intensity and heterogeneous density on T2WI signal.

Figure 4: Sagittal head FLAIR-MRI images post VP-shunting, showing an intermediate flair signal relative to both gray and white matter.

of an ependymoma (Figure 3A–C) Fluid-attenuated inversion recovery (FLAIR) MRI further showed the mass with hyper intensity and a heterogeneous signal density (Figure 4). After VP-shunting, the headache was relieved and no more seizures occurred, after which a tentative plan for tumour resection followed by radiotherapy was made. Unfortunately, the patient was discharged against medical advice from the hospital before the surgical procedure could be carried out.

DISCUSSION

Ependymoma is a rare tumor derived from the neuroepithelia, constituting about 3–9% of all central nervous system (CNS) tumors. In children, approximately two-thirds of such tumors occur in the infratentorial compartment while in adults the distribution is normally supratentorial. Ependymomas can be located either intracranially or intraspinal. Intracranial ependymomas occur more commonly in children, while intraspinal ependymomas predominate in adults. Sixty percent of tumors are located in the fourth ventricle of the infratentorial cranial fossa, which develop from the floor of the ventricle and may further extend into the foramen of Luschka and Magendie. Here we reported a case of ependymoma in the infratentorial
cranial fossa with a differential diagnosis of medulloblastoma on head CT scan and MRI scan.

In 1863, Virchow was the first to properly identify an ependymoma derived from ependymal cells [1]. This was followed by Bailey et al. in 1926, who described an ependymoma and an ependymoblastoma, which have been considered the most primitive tumors of the nervous system. This tumor arises from the medullary epithelial plate [2, 3]. Recently, researchers have proposed that radial glial cells may be the candidate stem cells of this tumor [4, 5]. The annual incidence in the countries of Central and South America as well as Asia is less than two per million in childhood [6]. In North America, Oceania and most of Europe, between 2–4 cases per million was reported, while in Denmark, Sweden, Finland, former East Germany and Slovenia, at least four cases per million are reported. Recently, McGuire et al. reported a comparison of the mean age of tumor incidence by tumor site in 237 children. Children with 5.0–0.4 years of age had only 4.2–5.9% of tumor located infratentorially while those of 12.2±0.9 years of age had 10.6–13.7% of tumor located in spinal column [7]. This indicated that the tumors would most likely be located in the spinal column when diagnosed in children with the average of age around 11 years; nevertheless, herein we have reported an 11-year-old child with a tumor located in the infratentorial space. According to WHO classification, ependymomas are classified into three categories:

(i) WHO grade I: subependymoma, myxopapillary ependymoma;
(ii) WHO grade II: classic ependymoma (with the variation of cellular, papillary, clear cell, and fibrillary or tanyctic);
(iii) WHO grade III: anaplastic ependymoma [8]. The symptoms of ependymomas depend on the tumor location, rate of tumor growth, etc.

In our patient the chief complaint was a generalized tonic seizure, followed by several symptoms which are associated with the location of the tumor in the infratentorial cranial fossa, such as vomiting, headaches, double vision and unsteadiness. Fundoscopical examination and neuroradiology imaging indicated an increase in intracranial pressure. Ependymomas are the third most common pediatric brain tumor (10.1%) after astrocytomas (47.3%) and medulloblastoma (16.3%) [9]. In the above list, atypical teratoid-rhabdoid tumor (ATRT) should be considered as well. It is very difficult to differentiate among these tumors, especially between a medulloblastoma and an ependymoma since both normally present as midline tumors, whereas astrocytoma and ATRT are generally eccentric. In order to distinguish between these tumors, additional examinations (CT scan and MRI) need to be performed. Based on the results from a non-contrast head CT scan, we made an initial diagnosis of a medulloblastoma with a differential diagnosis of ependymoma, although after an intravenous injection with contrast the results suggested that what the patient had was in fact an ependymoma. Moreover, head MRI scans revealed the ring/rim enhancement that is very typical for ependymomas. With regards to therapy, the symptoms should be relieved by the use of anti-convulsants and anti-edema agents. VP-shunting must be done to release the increase in intracranial pressure, followed by tumour resection and radiotherapy. All of this is essential to improve the patient’s outcome [10].

CONCLUSION

Detailed history and physical examinations are very important to diagnose an infratentorial cranial fossa occupying lesion. Radiological imaging by computed tomography scan and magnetic resonance imaging scan is essential to support the diagnosis of ependymoma and to distinguish it from a medulloblastoma. Moreover, improved patient outcome requires total tumor resection whenever possible, followed by radiotherapy.

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Author Contributions

Muhammad Yunus Amran – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Meryana Pauline – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

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Authors declare no conflict of interest.
REFERENCES

Extensive maxillofacial and oral myiasis
Felipe P Daltoé, André Ricardo Nosé, Rodrigo C Mosca,
Andrea Mantesso

CASE REPORT
A 28-year-old homeless man was brought to a police officer to the emergency service of the Regional Sul Hospital (São Paulo, Brazil) for evaluation of an extensive destruction of the oral and maxillofacial tissues. The patient was a heavy smoker (three packs of cigarettes per day) and according to his medical records he had a previous diagnosis of oral squamous cell carcinoma, but decided not get it treated. Three years later, the surface of the swelling revealed an extensive necrotic ulcer extending to the mouth, lips, nose and neck with live maggots visible and moving. Around 110 larvae were surgically removed and the necrotic tissue was debrided (Figure 1A–B). The patient was sent to the oncology service for a whole body evaluation, however, he passed away two weeks later due to systemic complications.

DISCUSSION
The term myiasis is applied to the injurious action that a parasites of the order Diptera causes to the living or dead tissue in which they grow in vertebrates organisms [1]. It is more common in animals and it has been rarely reported in humans [2]. Moreover, considering that myiasis develops by direct infestation of tissues by larvae (maggots) laid by flies [1], the mouth is not a common place for its development compared with dermis or other tissues.

Oral myiasis is usually associated with poor hygiene, wound healing, mouth breathing, mental impairment or senility [3]. In our case, the patient was clearly predisposed to the infestation considering the fact that he was a homeless, had unhygienic living condition, and had a previously untreated oral carcinoma.

The treatment of oral myiasis in most cases includes only surgical exploration to remove the larvae and necrotic tissue [3]. Alternatively, use of medicines such as ivermectin has also been proved efficacious, by enhancing parasitic death and their emergence to tissue surface [4]. Initial infestation can easily mimic gingival inflammations. Likewise, some cases of myiasis in association with oral tumors have also been reported [5].
CONCLUSION

The most effective action for prevention of human myiasis is by education and improvement of general sanitary conditions. Unfortunately, in underdeveloped or developing countries like Brazil, some people still live in poor environment associated with compromised hygiene and lack of information which can leads to human myiasis.

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Author Contributions
Felipe P Daltoé – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

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REFERENCES
Lesser-trélat sign in a patient with neoplasia of upper eyelid

Satyaki Ganguly, Kranti C Jaykar, Sambeet Kumar Mallik

CASE REPORT

A 40-year-old female presented to the dermatology OPD with an ulcerating growth of the left upper eyelid for the last three years. The growth was gradually increasing in size. On examination, there was an irregular erythematous swelling involving the whole of left upper eyelid with ulceration and areas of necrosis in the lateral part. A provisional diagnosis of sebaceous gland carcinoma was made. A biopsy of the eyelid growth was advised to confirm the diagnosis. Along with eyelid lesion, numerous asymptomatic, darkly pigmented papules were discovered over the face, trunk and extremities. These were more over the flexures like the neck, axilla, sub-mammary area and groin (Figures 1, 2). On being questioned about the lesions, the patient said that these lesions have appeared rapidly over a period of last six months. Detailed haematological, biochemical investigations, chest X-ray, upper gastrointestinal endoscopy, lower gastrointestinal endoscopy, mammography, abdominal ultrasound and bone marrow examination failed to reveal evidence of any systemic malignancy. Based on the clinical findings, a diagnosis of Lesser–Trélat sign in association with skin malignancy was made. The patient was referred to ophthalmology department for further management of the growth in the eyelid. Unfortunately, before a biopsy could be done the patient was lost to follow-up.

DISCUSSION

Seborrhoeic keratosis is a benign tumor, frequently pigmented, more common in elderly and composed of epidermal keratinocytes. The sudden appearance of numerous seborrhoeic keratoses in an adult may be a cutaneous finding of internal malignancy. Internal malignancy associated with the sudden development of numerous seborrhoeic keratoses in an eruptive fashion, with or without pruritus, is known as the sign of Lesser–Trélat [1]. Weakened subepithelial matrix—from the effects of neoplasm on the extracellular matrix of the host—has been postulated as a possible cause of Lesser–Trélat sign. To be considered a case of Lesser–Trélat, the keratoses should begin at approximately the same time as the development of cancer and run a parallel course in regard to growth and remission.
Common malignancies associated with this sign are adenocarcinoma of stomach (most common), lung, colon, breast, prostate, lymphoma, leukemia, ovarian cancer, nasopharyngeal carcinoma and transitional cell carcinoma of the bladder [2]. It has been associated with skin malignancies like maligna melanoma [3], lymphocytoma cutis [4] Paget’s disease [5], and Sézary syndrome [6]. A sudden eruption of many seborrheic keratoses may follow exfoliative erythroderma, erythrodermic psoriasis, erythrodermic drug eruption, lepromatous lepromyos and HIV infection [7]. Sebaceous gland carcinomas are very rare tumors, usually arises from the meibomian glands and majority of lesions affect the upper eye lid. The lesions are nodular and appear like a chalazion which lasts for more than six months.

CONCLUSION

Internal malignancy associated with the sudden development of numerous seborrheic keratoses in an eruptive fashion, with or without pruritus, is known as the sign of Lesser-Trélat

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