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Roya Samadi, Ali Akhoundpour Manteghi, Mehri Baghban Haghighi, Shervin Assari

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Case Report: A 25-year-old male with usual symptoms of OI, mild mental retardation and a psychotic feature due to complex partial seizures, after an experience of head trauma. He was treated with risperidone, aripiprazole, oxcarbamazepine, alendronate and vitamin D3.

Conclusion: It seems that seizure in OI is more common than general population. This may happen probably as a result of the complications of the head trauma in background of osteogenesis imperfecta. Moreover, clinical manifestations of complex partial seizure, can be mistaken with primary psychosis. Mental retardation and hearing loss might complicate this manifestation. So, psychiatric counseling and neurological evaluations should be carried out in OI patients.

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CASE REPORT

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Introduction: Osteogenesis imperfecta (OI) is an uncommon hereditary connective tissue disorder affecting collagen type I. The most common manifestations are frequent bone fractures and deformities, blue sclera, dental abnormalities and hearing loss. Seizures and mental retardation are not so common. **Case Report:** A 25-year-old male with usual symptoms of OI, mild mental retardation and a psychotic feature due to complex partial seizures, after an experience of head trauma. He was treated with risperidone, aripiprazole, oxcarbamazepine, alendronate and vitamin D3. **Conclusion:** It seems that seizure in OI is more common than general population. This may happen probably as a result of the complications of the head trauma in background of osteogenesis imperfecta. Moreover, clinical manifestations of complex partial seizure, can be mistaken with primary psychosis. Mental

retardation and hearing loss might complicate this manifestation. So, psychiatric counseling and neurological evaluations should be carried out in OI patients.

Keywords: Complex partial seizures, Lobstein syndrome, Osteogenesis imperfect, Psychosis

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INTRODUCTION

Osteogenesis imperfecta (OI) or Lobstein syndrome is a hereditary connective tissue disorder affecting collagen type I quantity or quality. It has 8 main types. Type 1 is the mildest and most common one. The pattern of inheritance is often autosomal dominant, although autosomal recessive and new mutation cases have also been reported [1]. Diagnosis is often made based on clinical manifestations and confirmed by DNA analysis. A range of clinical features can be seen in the skeletal system, such as osteopenia, frequent fractures, bone deformities, triangular face, short stature, disproportional body, hypotonia and laxity of skin and joints. Also, blue sclera, dental abnormalities and hearing loss are usually observed. The intellect is often normal [1]. Prevalent neurological complications include macrocephaly, communicating hydrocephalus, cerebral atrophy, basilar invagination, brainstem compression and skull fractures [2]. However, seizures are rather uncommon [2, 3].

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CASE REPORT

The patient was a 25-year-old male, from a remote village in North Khorasan, Iran. When he was 21, he experienced a motorcycle accident, resulting in humerus fracture and head trauma, but without any seizures, amnesia, or coma. Brain CT-scan did not show any complication at the time. However, within 1–3 months after that, he developed a mental dysfunction, including intermittent psychotic periods, presenting with disorganized behavior and thought, disturbed form of thought, visual hallucinations, self-talking and laughing, aggressiveness and reduced need to sleep. He would have no recollection of these episodes afterwards. He also had states of left hand clawing, headache, vertigo and blurred vision just before the beginning of these episodes. He had been an easygoing as a child and had reached milestones normally. Family psychiatric history was negative.

He had been admitted in psychiatric centers twice previously and diagnosed with schizophrenia. The treatment following discharge from hospital had not been regular and effective. Duration of each episode was variable from several days to one or several months-based on information on past admissions or history taking from family. In his most recent admission in our hospital in Mashhad, his abnormal appearance, history of frequent bone fractures and skeletal deformities as a result of mild to moderate traumas were noticed. One of his five siblings had similar facies, skeletal deformities and frequent atypical fractures which led us to consider OI. Nevertheless, parental history of frequent fractures and deformities was negative. Therefore, more precise examinations, laboratory tests and genetic counseling were performed.

In general appearance, short stature, kyphoscoliosis (Figure 1), triangular face and macrocephaly were evident. Easy bruising and laxity of skin and ligaments, decreased muscular mass and strength, flat foot (Figure 2) and genu varum were observed. Deep tendon reflexes were normal.

In X-ray studies of extremities and vertebra, beside the osteoporosis (Figure 3), previous fractures were seen in atypical locations, such as compressed fracture of thoracic vertebra (T_7) (Figure 3A–B), shaft of the left femur (Figure 3C), left wrist and ankle and right elbow (Figure 4). Subsequent deformities included decreased height of the vertebra (Figure 3A–B), external rotation of left ankle and foot and flexion deformity of the elbow joint (Figure 4). Also, bone healing following the fractures seemed impaired as evidenced by malunion of the femoral shaft and elbow joint (Figure 3C–D). In skull X-rays, there were wormian bones (Figure 3E).

Audiometry indicated a unilateral conductive hearing loss. IQ test, he was not cooperative. However, mild mental retardation was suggested clinically based on his educational background: He had barely finished elementary school, with poor grades. Electroencephalogram (EEG) showed a generalized epileptiform spike and waves.

Figure 1: Kyphoscoliosis.

Figure 2: Flat foot.

Considering the intermittent pattern of his episodes, clear inter-episodic phases, the EEG findings and the non-deteriorating nature of his disease (he had had a constant level of function during these 4 years, in spite of not receiving sufficient treatment), and a favorable response to anticonvulsants; the symptoms of hand clawing, headache, vertigo and blurred vision prior to attacks were altogether considered as sensory auras

Figure 3: (A) Anteroposterior radiograph from dorsal spine shows anterior Wedge fracture in T7 and spinal mal alignment in the form of Kyphoscoliosis. Diffused reduction in spinal density due to Osteoporosis is seen, (B) Lateral radiograph from dorsal spine shows anterior Wedge fracture in T7 and spinal mal alignment in the form of Kyphoscoliosis. Diffused reduction in spinal density due to Osteoporosis is seen, (C) Elbow joint radiograph shows severe decrease in joint space (fusion article), flexion deformity and malunion due to fracture following previous trauma suggestive of severe secondary Osteoarthritis. Bowing deformity of Radius is seen. Relative reduction in bones' cortical thickness is suggestive of Osteoporosis. Metal fixator is observed in the distal of Humorous, (D) Anteroposterior radiographs from left Femur bone shows malunion of Femoral shaft following previous fracture along with Bowing deformity of Femur. Relative reduction in bone's cortical thickness is suggestive of Osteoporosis, and (E) Lateral radiograph from skull shows small bones with irregular border among Parietal and Occipital bones (Lambdoid suture) suggestive of Wormian bones.

(simple partial seizures), subsequently complicating with psychotic symptoms (complex partial seizures).

Magnetic resonance imaging scan of brain showed wide fissures, dilated ventricles and a sort of cortical atrophy, disproportionate with age. Serum levels of calcium and phosphorus were normal (9.7 and 3.7 mg/dL

Figure 4: Flexion deformity of elbow joint.

respectively). However, serum alkaline phosphatase rose to 382 IU/L, probably due to frequent bone fractures. Thyroid function tests were normal. Genetic counseling indicated autosomal recessive OI.

Therapeutic plan was based on an antipsychotic (aripiprazole, 10 mg/day) an antiepileptic (carbamazepine, 200 mg/day initially and increased to 400 mg/day, 2 days later) and bone enhancing agents (alendronate, 70 mg weekly, and vitamin D₃ injection, 300000 IU, monthly). After admission aripiprazole was started and next day carbamazepine was added on. As the patient experienced the side effects of carbamazepine (dizziness and diplopia), it was changed to oxcarbazepine 150 mg/bid. This treatment satisfactorily prevented the episodes and thus proving that the symptoms were secondary to CPS. Loosening of associations and other psychotic symptoms improved after 10 days. Table 1 gives the differences between primary psychosis and CPS with psychosis.

Eventually, he was diagnosed with mild mental retardation and psychosis due to CPS. History of head trauma is thought to be involved in development of CPS in this patient, as OI may predispose patients to severe complications of minor traumas.

He was referred to an endocrinologist after discharge to better treat his OI. He has been followed-up in three visits in the last 2.5 years and is under treatment with oxcarbamazepine 150 mg, twice a day. No neuro-psychiatric symptoms have been reported or observed in this period.

DISCUSSION

Seizures are not frequently comorbid with connective tissue disorders such as osteogenesis imperfecta [3, 4]. However, in a study of the neurologic profile of OI patients by Charnas et al. [5], 5 cases out of 76 patients (6.5%) suffered from seizures, while the prevalence of seizures in the general population of diverse countries has been reported 1.5 to 57 per 1000 [6]. Therefore, it seems that seizures in OI are not absolutely rare and at least are more common than the general population.

Table 1: Differentiation between primary psychosis and CPS with psychosis.

	CPS with psychosis	primary psychosis
Pattern of episodes	The intermittent pattern	Not specific
Inter-episodic	clear inter-episodic phases	Not specific
EEG findings	Epileptiform spike/sharp and waves	Not specific
Deterioration	Usually not	Maybe depends on psychotic disorder type
Streohtypic auras	+	-
Hallucination type	Visual hallucinations are more common	Auditory hallucinations are more common
Response to treatment	Rapid response to anticonvulsant	Response to Antipsychotics within days to months- depends on psychotic disorder

It seems that OI patients (as well as our patient) have an increased tendency for bleeding during surgeries and even minor traumas, as a result of impaired collagen, capillary fragility, impaired platelet retention and aggregation and factor VIII deficiency [7–9]. These factors may lead to brain hematomas and, thereby, seizures and psychomotor retardation in OI patients [10].

This case report is an example of clinical manifestations of patients with CPS, which can be mistaken with primary psychosis. This was probably a complication of the head trauma in a patient with underlying osteogenesis imperfecta. Mental retardation, hearing loss, poor socioeconomic situation and low level of education, might have worsened the capability of the patient to cope with these problems and affected the outcomes as well. Therefore, besides orthopedic surgeries, additional interventions such as lifestyle modifications, physical rehabilitations and psychological counseling should be done in OI patients [1].

CONCLUSION

Clinical manifestations of complex partial seizure, can be mistaken with primary psychosis. As it seems that seizures in Osteogenesis imperfecta (OI) are more common than the general population, neuropsychiatric evaluation can also be beneficial in achieving the best therapeutic results and quality of life in these patients.

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Author Contributions

Roya Samadi – Substantial contributions to conception and design, Acquisition of data, Revising it critically for important intellectual content, Final approval of the version to be published

Ali Akhoundpour Manteghi – Substantial contributions to conception and design, Acquisition of data, Revising it critically for important intellectual content, Final approval of the version to be published

Mehri Baghban Haghighi – Acquisition of data, Drafting the article, Final approval of the version to be published

Shervin Assari – Acquisition of data, Drafting 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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