A novel presentation of Hecht-Beals trismus pseudocamptodactyly syndrome: Review of literature and holistic management

Partha Chattopadhyay, Cyriac Abby Philips, Debargha Dhua, Prabir Kumar Josh, Prasanta Mukhopadhyay

ABSTRACT
Introduction: Hecht-Beals Trismus Pseudocamptodactyly syndrome, a rare autosomal dominant inherited disease encompass a variety of genotypic and phenotypic verges that affects members of a family. Case Report: Here we report a patient, who presented with congenital coal black hyperpigmentation of the skin with progressive trismus. Conclusion: With emergence of new reports, the mutational and expressive variations of this disease is becoming more enigmatic. Our tidings shed light on a novel clinical association with coal black skin pigmentation in whom trismus was dealt sufficiently through operative methods. Further mutational analysis and clinical probing would help in concretizing a definite categorization of these diseases that lie between these gamuts.

Keywords: Trismus, Masseter Spasm, Hypermelanosis


INTRODUCTION
Trismus is a condition in which there is inability to normally open the mouth due to varied reasons, commonly tetanus, sub mucosal fibrosis, condylar fracture, temporo-mandibular joint dysfunction, neuroleptic syndromes, peritonsillar abscess and ankylosis of temporomandibular joint. Trismus can also be part of a rare syndrome complex known as the Trismus-Psuedo-Camptodactyly Syndrome [1]. Here we present a patient who presented to us with Trismus-Psuedo-Camptodactyly Syndrome, in whom congenital progressive diffuse coal black hyperpigmentation was a striking feature, with mild affection of his elder brother (coal black pigmentation to a lesser degree with mild trismus). Corrective surgical procedure of the trismus and microstomia resulted in betterment and quality of life of our patient without any recurrence even after nine months.

CASE REPORT
A 17-year-old male, presented to us with progressive difficulty in opening the mouth for two years, associated with bony deformities of skull and extremities and
progressive hyperpigmentation of the skin since birth. There was no relevant history of infections in childhood, drug intake by the mother of the patient during pregnancy, drug intake in the patient, trauma, surgeries or similar complaints in the family members. On examination, the patient was found to have features of macrocephaly, micrognathia with microstomia, arachnodactyly, trismus, pseudo-camptodactyly (flexion contractures of the distal joints of the hands on complete dorsiflexion), a broad stout neck, with low set ears, scoliosis and diffuse melanotic hyperpigmentation of the whole body, including palms and soles, with linear melanonychia, without any pigmentary changes of the oro-genital mucosal surfaces (figures 1A, B). Systemic examination revealed no abnormalities, except for an irregularly irregular pulse rate. A complete hemogram, liver function test, renal function test including serum electrolytes along with thyroid function testing fasting blood glucose levels and a routine urine analysis were well within normal limits. Roentgenograms of the skull showed features of diploic space expansion along with premature sinuses with frontal and occipital bossing and that of the peripheral bones and the knee joints revealed cortical widening with delayed fusion of the epiphyses. An oral pantomogram revealed features suggestive of a probable mandibular condylar dysplasia, as interpreted by our consultant radiologist (figure 2). The electrocardiogram revealed features suggestive of ventricular premature beats. A Computed Tomography of the abdomen and a Magnetic Resonance Imaging of the Brain revealed no significant abnormalities. An Echocardiogram performed was within normal limits.

Based on the specific clinical findings, a diagnosis of Arthrogryposis, distal type 7, also known as Hecht-Beal's Trismus - Pseudocamptodactyly Syndrome was made. The patient had painless restriction of mouth opening beyond 20 mm. A Computerized Tomography of the skull bones revealed normal condyles and the temporomandibular joints on either side with bilateral mandibular coronoid hyperplasia (figure 3). These enlarged coronoids was impinging on the posterior aspect of the zygoma causing trismus and hence a bilateral coronoidectomy procedure was planned.

The patient was successfully intubated using an adult fibreoptic bronchoscope under intravenous anesthesia. Difficulty in intubation was anticipated and equipment pertaining to emergency jet ventilator and tracheostomy was kept ready. The intra-operative inter-incisor (distance between the upper and lower incisors) mouth opening was found to be 35 mm. The access was through a preauricular incision. The hyperplastic condyles and the normal temporomandibular joint, was exposed and a coronoidectomy, performed on either side. After proper haemostasis, the operative site was closed with drains in place. The post-operative period was uneventful. An intraoral device (figure 4) was used in the immediate postoperative period in order to maintain mouth opening with physiotherapy sessions to prevent secondary scarring and fibrosis. In the subsequent days, the inter incisor mouth opening gradually improved to 45 mm (figure 5). Afterwards, he was discharged from the hospital with an advice to continuing orthopedic rehabilitation and maintenance of oral hygiene. At consequent monthly follow up even after ten months revealed maintenance of mouth opening with no features of a relapse.

**DISCUSSION**

Trismus − Psuedocamptodactyly Syndrome is a rare autosomal dominant disease affecting multiple family members, which manifests as inability to open the mouth (most likely due to masseteric fibrous bands and coronoid hyperplasia, which lead to molding deformities and developmental hypoplasia of the bones of

![Figure 1: A, B) Features of pseudocamptodactyly (flexion contractures of the distal joints of the hands) with dysmorphic facial features including micro-agnathia, low set ears and macrocephaly. Features of Trismus (difficulty in opening the mouth), with diffuse hypermelanotic pigmentation of palms along with arachnodactyly.](image-url)
mandible), flexion contractures of the fingers (apparent on dorsiflexion), with facial dysmorphism in the form of macrocephaly, facial asymmetry, downslanting palpebral fissures, micrognathia, long philtrum, low set ears, arachnodactyly, and chest wall deformities [1, 2].

Trismus – pseudocamptodactyly Syndrome can also be associated with the Carney Complex Variant in which spotty hyperpigmentation or lentiginosis occurs along with distal arthrogryposis and cardiac myxomas. Hecht and Beal in 1969, described this rare occurrence in a father and his four children (two sons, two daughters) who presented with problems in mastication and short finger flexor shortening leading to camptodactyly [3]. Jong (1971), Ter Haar and Vanhoof (1974) and Mabry et al (1974) associated the complex of Trismus with Pseudocamptodactyly in a large number of families from Denmark and also in a kindred with severe features who was traced back to a Dutch girl who migrated to Tennessee soon after the American Revolution, and hence came a new nomenclature, the Dutch-Kentucky Syndrome [4, 5, 6].

Mabry et al. 1974; Lefaivre and Aitchison, 2003 [7], Veugelers et al. 2004 [8] identified an arginine 674-to-glutamine mutation in the MYH8 gene (which was in fact previously found to be associated with the Carney complex) leading to this autosomal dominant disease. Toydemir et al. (2006) identified the R674Q mutation in the MYH8 gene in all affected members of four families with trismus-pseudocamptodactyly syndrome, including descendants of the Dutch family reported by Ter Haar and Van Hoof (1974) [9]. Bamshad et al. (1996) in a revised and extended classification scheme of the distal arthrogryposis, referred to this disorder as distal arthrogryposis type 7 (DA7) [10].

In 1992, Chen et al. and in 2008, Minzer-Conzetti et al. broadened the phenotype of Trismus-Pseudocamptodactyly Syndrome by adding further findings into the clinical spectrum of the disease and also showed that trismus and camptodactyly can occur even as isolated events in some cases [11, 12].

Hence, these case reports suggest that Trismus-Pseudocamptodactyly Syndrome is an autosomal dominant disease that runs in many generations of the family.

Hypermelanisation can be of localized and diffuse type. Diffuse hyperpigmentation occurring since birth is a very rare abnormality and can be seen with Congenital Adrenal Hypoplasia (coal-black pigmentation), and with other rare dermatological conditions like Melanosis Universalis Hereditaria, Diffuse Congenital Melanosis, Familial Progressive Hyperpigmentation, Generalized
Acanthosis Nigricans [13]. Lentigens have been described with Trismus-Pseudocamptodactyly Syndrome before, in the form of spotty pigmentation along with Carney Complex.

The treatment options for Psuedocamptodactyly Syndrome are bleak. The main target is alleviation of symptoms, mainly Trismus, by operative procedures, since pharmacological management is of little help. Even then, any possible surgical therapy must be undertaken in these patients to relieve trismus so much so, that most of these patients suffer from complications of malnutrition.

Our patient presented with typical features of trismus-pseudocamptodactyly syndrome and previously unidentified phenotype of diffuse cutaneous melanocytic hyperpigmentation, but lacked the Carney Complex abnormalities. His elder brother had same features but to a much lesser degree and was asymptomatic and thus oblivious of the potential peril. Diffuse cutaneous coal black pigmentation without carney complex associated with trismus-pseudocamptodactyly syndrome has not been documented before in peer reviewed literature.

Surgical management of Trismus-Pseudocamptodactyly syndrome has been glorified very seldom in medical literature. Almost all the cases reported before involved correction of coronoid hyperplasia by a respective procedure, followed by orthopedic rehabilitation and timely oral device application. Lefaivre and Aitchison in 2003 first reported the surgical management of this disease in their seminal report that highlighted the correction of trismus in a 28-month-old man who had a mouth opening of 6 mm. The man had initial endoscopic corrective procedure resulting in an early relapse at six weeks, which was then definitively corrected by extensive sub periosteal mandibular resection and coronidectomy with a final mouth opening at 25 mm at 12 month follow up Gasparini et al. (2008) reported the surgical correction in a girl at four years of age by means of coronidectomy which subsequently relapsed after six years and was then managed with a second definitive surgery without further relapse [14]. Another case report highlighted a single corrective bilateral coronidectomy that provided normal mouth opening in a patient with an initial opening of 25 mm [15]. Prompt and complete early correction of coronoid hyperplasia in Hecht-Beal Syndrome along with timely application of oral devices, orthopedic physiokinetic therapy and maintenance of oral hygiene proves to be gladdening in management of this curious and difficult to treat disease entity.

CONCLUSION

Hecht Beal Syndrome encompasses a variety of genotypic and phenotypic verges in a patient. As more awareness transpire, with an extensive armada of nonpareil diagnostics, the mutational and expressive variations of this disease is becoming more curious and obvious. In our patient, trismus was a main concern, which was dealt successfully through surgery. The case under scrutiny here brings forth avant-garde clinical association debut of coal black skin pigmentation and trismus in a patient of Hecht Beal Syndrome in whom surgical correction of lock jaw (addressed less in other reports) resulted in an improvement in quality of life, pushing further insights into novel representations of this group of diseases.

**********
Author Contributions
Partha Chattopadhyay – Analysis and interpretation of data, Drafting the article, Final approval of the version to be published
Cyriac Abby Philips – Conception and design, Analysis and interpretation of data, Drafting the article, Critical revision of the article, Final approval of the version to be published
Debargha Dhua – Acquisition of data, Critical revision of the article, Final approval of the version to be published
Prabir Kumar Josh – Analysis and interpretation of data, Drafting the article, Final approval of the version to be published
Prasanta Mukhopadhyay – 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.

Copyright
© Partha Chattopadhyay et al. 2012; This article is distributed under the terms of Creative Commons attribution 3.0 License which permits unrestricted use, distribution and reproduction in any means provided the original authors and original publisher are properly credited. (Please see www.icaserportsandimages.com/copyright-policy.php for more information.)

REFERENCES
6. Jong JGY. A family showing strongly reduced ability to open the mouth and limitation of some movements of the extremities. HUMAN GENETICS 1971;13(3):210-7.