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Case Study

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PETERS' PLUS SYNDROME: A RARE CASE REPORT

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ABSTRACT

Peters' Plus syndrome is an infrequent genetic disease, characterized by low height, peculiar facial appearance, eye anomalies and mental retardation with less than 75 cases reported worldwide. The clinical examination, combined with the comparative pattern technique described, was applied to a 3 year old female child, for requisite diagnosis of this rare syndrome. It is important to know and identify these clinical symptoms in order to diagnose the disorder, starting early rehabilitation and supplying the family with genetic counselling on this little known ailment.

KEYWORDS: Peters' Plus Syndrome, Krause-Kivlin Syndrome, Peters' anomaly, congenital disorder, glycosylation.

INTRODUCTION

Peters-plus syndrome, also known as Krause-Kivlin syndrome, is a rare congenital disorder of glycosylation with an autosomal recessive pattern. It is diagnosed clinically by the presence of Peter's anomaly – a congenital corneal opacity secondary to a defect in neural crest cell migration that causes a malformation of the anterior eye segment, associated with other symptoms. These symptoms are delayed psychomotor development with variable degrees of mental retardation. Disproportionate, short stature is observed along with facial features like cleft lip or palate (seen in approx. 50% cases), hypertelorism, prominent forehead and narrow eyes. Thin cupid's bow-shaped upper lip with elongated philtrum, small ears with hearing loss, broad & webbed neck, and joint hyperextensibility.

The presence of congenital heart defects, genitourinary abnormalities such as cryptorchidism, hypoplastic clitoris and hydronephrosis, as well as structural brain malformations may affect the prognosis of individuals.

This disorder belongs to protein-o-glycosylation defects, subgroup O-flucosylglycan synthesis defects. Glycosylation is known for its abundant and diverse form of post- transcriptional modification. It takes part in every physiologic process which including its role in modulation of IgG. Malfunction of this system is associated with different inflammatory and autoimmune diseases such as SLE, rheumatoid arthritis, inflammatory bowel diseases, cancer and AIDS. Additionally, inflammation pathways play a prime role in endothelial and kidney damage.

Its exact incidence is still unknown, and fewer than 75 cases with this anomaly have been identified in the medical literature.[1]

CASE PRESENTATION

A 3 year old female patient reported to our department with mild, intermittent pain in left maxillary and mandibular molar region since 3-4 months.

Patient presented with eye abnormalities, hearing disability, difficulty in deglutition, fused digits, cleft palate since birth. She got palatoplasty done 1 year back at Chettinad Hospital and Research Institute, Chennai and has been using hearing aids since.

On general examination, dysmorphia is evident with short stature, with developmental milestones delayed. (Fig.1)

Fig.1.

Extraorally, retrograde mandible, depressed nasal bridge and elongated philtrum.bilateral micropthalmia, coroneal opacity, microcornea, sclerocornea and corneal coloboma was observed in both eyes. (Fig.2)

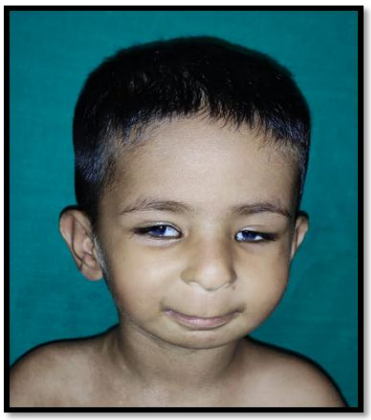

Fig.2.

Both the hands were with partial syndactyly. (Fig.3)

Fig.3.

Hypopigmented patches were covering the right shoulder region. (Fig.4)

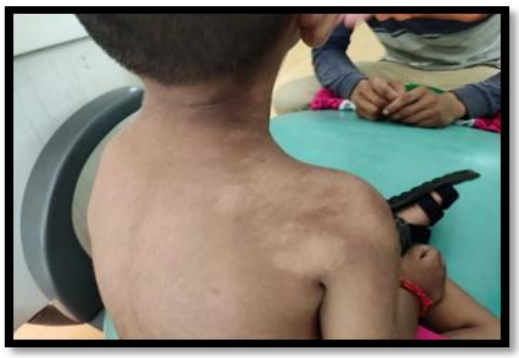

Fig.4.

Sacral dimple was evident on the lower back. (Fig.5)

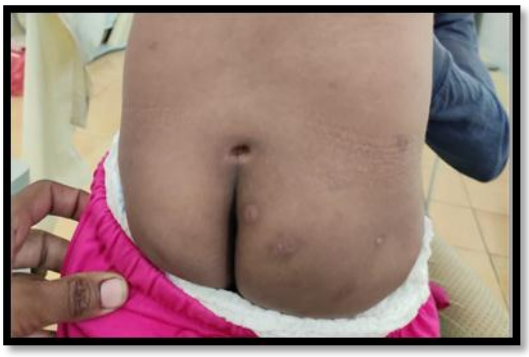

Fig.5.

Intraoral findings revealed deep dentinal caries and root stumps in multiple teeth. Scar on the palate was observed due to palatoplasty. (Fig.6)

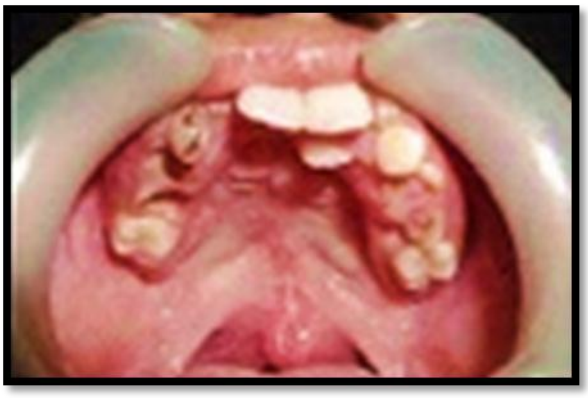

Fig.6.

A provisional diagnosis of Early Childhood Caries in a case of Peters' Plus Syndrome was established in the patient.

Apart from routine investigation for dental condition, hand and wrist radiograph was taken which showed no bone involvement in partial syndactyly. (Fig.7)

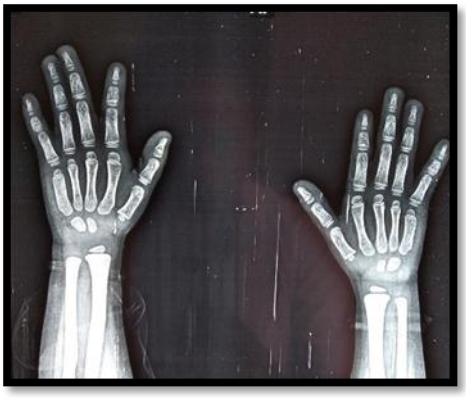

Fig.7.

Skeletal survey of long bones and thorax and blood investigations were normal.

Dental management for the patient was done in our institute.

DISCUSSION

Peters' anomaly could be inherited either as autosomal dominant or autosomal recessive mode. It occurs sporadically (with no other associated family members).[1] It is known to be associated with chromosome 4 abnormalities.[2] Systemic abnormalities may include congenital heart defects, hydrocephalus, and renal dysgenesis, none of which were in our case.[3] Midline body structures seem to be selectively involved in some cases but skeletal survey of our patients showed normal long bones and thorax. There was no endocrinal anomalies result from association of pituitary dysfunction.[4] Patients can have cleft lip and palate such as present in this patient, genitourinary abnormalities, pilonidal cyst, spina bifida, sacral hypoplasia which was prominently visible in our case, anal atresia or vesicocolonic fistula. The exact prevalence of this syndrome is unknown. Fewer than 75 affected individuals have been reported in the literature; they come from varied ethnicities.

Dental treatment in a patients vary according to their health condition. Early detection of the disease is vital to proceed for early treatment to ensure good development of the general and oral health of the child. In this case, patient underwent surgical correction for congenital defect observed in this syndrome i.e. cleft palate at the age of 1. The high level of dental decay detected could hold accountability to on-demand bottle feeding, poor oral hygiene, high sweet consumption, lack of use of fluoride prevention and lack of regular dental visits. For treatment of carious teeth several invasive dental procedures were required which could not be performed under general anaesthesia since patient suffers from Renal Parenchymal Disease Grade 3. Thus treatment is ongoing under local anaesthesia with required protocol. In case patient presents with Gingival hyperplasia or hypertrophy induced by anticonvulsants/ antiepileptics/ Ca channel blockers prescribed for symptoms associated with Peters Plus, it is difficult to maintain oral hygiene and, often, the masticatory function. To treat gingival hyperplasia, the medication may be replaced, after consulting the patient's physician, and conservative periodontal treatment may be provided, including supra and subgingival scaling, prophylaxis and oral hygiene. Corticosteroids are contraindicated in these patients for the risk of developing glaucoma.

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