A case of Peters Plus Syndrome

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Abstract

Peter’s Plus Syndrome is a rare, autosomal recessive genetic disorder characterized by abnormalities of anterior chamber, short stature, developmental delay, cleft lip & facial dysmorphism. It is listed as a rare disease by Office of rare diseases of the “National Institute of Health” with only less than 70 cases with this condition reported so far. Here we present such a case reported from our institution.

Key Words


Clinical Details

46 yr old female presented with palpitation & breathlessness since 6 months, with history of hard of hearing & decreased vision since childhood. She had normal developmental milestones but was a poor performer at school. There was no significant family history. Her height - 146 cm, Weight - 36 Kg, Arm Span - 136 cm, Height neck ratio - 13.6, & had hypertelorism, flat nasal bridge, low set ears, short neck, low hair line, high arched palate, maloccluded teeth, central cyanosis, Grade I clubbing. Cardiovascular system - Loud S1, wide split S2, loud P2, Ejection systolic murmur in left 2nd & 3rd intercostal space.

Investigations

The electrocardiogram showed right axis deviation with left atrial enlargement and right ventricular hypertrophy by voltage criteria. Her chest X- ray had increased bronchovascular markings with CT thorax demonstrating features of pulmonary artery hypertension. X ray of cervical spine had C3-C4 vertebral fusion. Echocardiogram was suggestive of Lutembacher’s syndrome with pulmonary hypertension. Ultrasonogram of abdomen, tomogram of abdomen, MRI of brain was done to rule out other congenital abnormality and was found to be normal. Audiogram revealed bilateral moderate to severe sensory neural hearing loss. Slit lamp examination was done which showed aphakia in both eyes with scattered vacuoles in cornea, iris atrophy and iridocorneal adhesions in both eyes. There was iridodonesis in left eye and pin pointed pupils in right eye. Karyotyping was suggestive of X chromosome deletion in its short arm (Xp).

Conclusion

This is a case of Peter’s Plus Syndrome with Classical clinical & radiological findings. Ours is unique case in that vertebral fusion, Xp deletion has not been reported so far. The previously reported case was by Dr Seema Kapur et al from Maulana Azad medical college, New Delhi in a 10 year boy. Our case is unique that though the disease was congenital, the patient presented only at 46 years.

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Case Report

Legs showing increased gap between big toe and second toe

Slit Lamp picture showing iris atrophy, corneal opacity and irido-corneal adhesions

Karyotyping – sex chromosome x short arm deletion

C3-C4 Vertebral Fusion

Review of Literature

The term Peters plus Syndrome was first coined in 1984 by a Dutch Ophthalmologist Mary Van Schooneveld, other synonyms being Krause-Kivlin Syndrome, Peter’s anomaly - Short Limb dwarfism syndrome. The genetic defect underlying this condition lies in \( \alpha_1,3 \) galactosyl transferase like gene that codes for the enzyme \( \alpha_1,3 \) galactosyl transferase, which is needed for adequate glycosylation of proteins. The glycosylation of proteins is defective in this rare syndrome. There is no racial / sexual predilection. Peters anomaly is a rare form of anterior segment dysgenesis in which abnormal cleavage of anterior chamber occurs.
It can be associated without cataract (Type I) and with Cataract (Type II). Peter’s anomaly when associated with systemic defects and developmental delay form Peter’s Plus Syndrome. It is important to identify this disorder as there is definite risk of recurrence, so genetic counseling is of great importance & with identification of gene, prenatal diagnosis may be possible.

_Treatment of manifestations:_ consideration of corneal transplantation (penetrating keratoplasty) for severe bilateral corneal opacification prior to age three to six months to prevent amblyopia; Simple separation of iridocorneal adhesions in mild cases; management of amblyopia by a pediatric ophthalmologist; surgical/medical intervention for glaucoma development/educational interventions as needed.

**References**