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Case report

SEVERE PETER PLUS SYNDROME: A RARE CASE REPORT

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ABSTRACT

Severe Peter plus Syndrome is a rare autosomal recessive condition that is characterized by ocular anomaly and associated with other systemic major or minor anomalies. Mutations of B3GALTL gene encoding beta 1,3 glucosyltransferase have been seen in patients with Peter Plus Syndrome.[1] We report a male patient with unusually severe manifestations of Peter Plus Syndrome including prominent forehead, long area between nose and mouth (philtrum), pronounced double curve of the upper lip, Anterior Eye Staphyloma (Bilateral), retrognathia, widely spaced nipples and Fallot's tetralogy. To our knowledge Fallot has not been reported previously in Peter plus Syndrome and bilateral anterior staphyloma, a most severe anterior chamber eye defect is also apparently rare in this syndrome. Our patient might represent a new variant of severe Peter plus syndrome with anterior eye Staphyloma and Fallot's tetralogy.

Key words Severe Peter Plus syndrome, Fallot's tetralogy, Anterior eye staphyloma, Retrognathia

INTRODUCTION

Severe Peter Plus syndrome is a very rare autosomal recessive condition characterized by ocular anolamly, disproportionate short stature, developmental delay, cleft lip/palate and other systemic major or minor anomalies. "Peter plus syndrome" is also known as Krause-van Schooneveld – Kivlin syndrome. [1-3]

We report a male patient with unusually severe manifestations of Peter Plus Syndrome including prominent forehead, long area between nose and mouth (philtrum), pronounced double curve of the upper lip, Anterior eye staphyloma (Bilateral), retrognathia, widely spaced nipples and Fallot's tetralogy.

CASE

A term male neonate (birth weight: 2.9Kg), normal delivery, born to non consanguineous parents at 7th week of gestational age was brought to our hospital on 1st day of life with complaints of protrusion of right eyeball. Examination revealed prominent forehead, long

area between nose and mouth (philtrum), pronounced double curve of upper lip (Cupid's bow), bilateral anterior eye Staphyloma, webbed neck, retrognathia & micrognathia, short stature. Systemic examination revealed a single second sound in pulmonary area, systolic murmur, no cyanosis. 2D- ECHO findings revealed Tetralogy of Fallot.

Fig.1: Widely spaced nipples, depressed nasal bridge, cupid's bow

Fig.2: Large forehead, long philtrum, right eye staphyloma

DISCUSSION

Mutations of B3GALTL gene encoding beta 1,3glucosyltransferase have been seen in patients with Peter Plus Syndrome[4]. Less than 70 people with this condition have been reported worldwide. Patients with this syndrome may also have shortened upper limbs i.e. rhizomelia and shortened fingers and toes (brachydactyly)[5]. It is also noticed that intellectual disability may be present which may be mild to severe, although some individuals may have normal intelligence[6]. We report a male patient (neonate) with unusually severe manifestations of Peter plus Syndrome.[7] Fallot's Tetralogy has not been reported previously in Peter Plus Syndrome. Bilateral Anterior Staphyloma, a most severe anterior chamber eye defect is also relatively rare in this syndrome.[8] Our patient might represent a new variant of Severe Peter plus Syndrome with anterior eye Staphyloma and Fallot's tetralogy.[9,10]

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