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OCULAR MANIFESTATIONS IN RUBINSTEIN -TAYBI SYNDROME- A CASE REPORT

Ophthalmology

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ABSTRACT

Rubinstein-Taybi syndrome is an extremely rare genetic condition caused by mutation or deletion in CREBBP and/or EP300 gene located in chromosome 16. It has an incidence of 1 in 1,25,000-3,00,000 births.(1) A multitude of major malformations and clinical complications, though not pathognomonic, amongst which diverse ocular abnormalities are part, are associated with the syndrome.Our patient, an 8 year old boy, with microdeletion of chromosome 16p on Fluorescence In Situ Hybridization(FISH), presented with unilateral iris and retinochoroidal coloboma along with other systemic abnormalities.

KEYWORDS

Rubinstein-taybi Syndrome, Crebbp Gene, Ep300 Gene, Coloboma.

INTRODUCTION

Rubinstein-Taybi syndrome/Broad Thumb Hallux Syndrome is a rare multisystem disorder, initially reported by Rubinstein and Taybi in 1963.[2]

With an equal distribution amongst males and females, the syndrome is associated with distinctive features like Neurodevelopmental abnormalities(Developmental delay, Mental retardation), Craniofacial abnormalities(Microcephaly, Hypertelorism, Epicanthal folds, Low set ears, Beaked nose, Hypoplastic maxilla, High arched palate, Crowded teeth, Talon cusps of incisors, Micrognathia/Retrognathia), Ocular abnormalities(Nasolacrimal duct obstruction, Corneal opacity, Refractory errors, Strabismus, Ptosis, Coloboma, Congenital cataract, Congenital glaucoma).

Other associated features could be Capillary malformations, Syndactyly, Clinodactyly, Broad thumbs, Persistent fetal finger pads, Cryptorchidism, Congenital heart diseases, Scoliosis and an increased risk of tumours that show patterns of neural origin.

Here we present the case report of an 8 year old male patient with various ocular abnormalities including iris and retinochoroidal coloboma.

Case Report

Our patient, an 8 year old male child, known case of Patent Ductus Arteriosis +Arterial Septal Defect on treatment, was brought to the Out Patient Department(OPD) for regular checkup.

Thorough history was taken. FISH analysis showed microdeletion of chromosome 16p. Intellectual disability, Developmental delay, Microcephaly, Hypertelorism, low set ears, limb abnormalities and incoherent speech were noted.

Best corrected visual acuity of 6/60 and 6/36 measured in right eye and left eye respectively. Retinoscopy showed compound myopic astigmatism in both eyes.

Latent nystagmus was observed in the left eye. Anterior segment examination revealed nebulomacular corneal opacity at 5 O'clock and inferior iris coloboma in the right eye.

Fundus examination revealed bilateral myopic fundus picture with inferior retinochoroidal coloboma excluding optic disc in the right eye.(Figure1)

A change of spectacle corresponding to best correction was prescribed and parents were counselled regarding the prognosis of the syndrome and the necessity of remaining in regular followup.

DISCUSSION

Diverse ocular abnormalities occur in majority of Rubinstein-Taybi syndrome cases.

In a study conducted amongst 24 Rubinstein Taybi Syndrome patients by M.M van Genderen et al. showing visual acuity of ≤0.3 in 5 patients, the most frequent abnormalities noted were Nasolacrimal duct obstruction, Corneal abnormalities, Congenital glaucoma, Congenital cataract and Coloboma, amongst which retinal abnormalities were the most observed.[3] Visual Evoked Potential Test(VEP) showed abnormal waveform in 15 patients.[3]

Electroretinography(ERG) abnormalities were noted in 14 of 18 patients.[3]

Our patient presented with unilateral iris and retinochoroidal coloboma along with compound myopic astigmatism and myopic retinal changes in both eyes.

He was adviced VEP and ERG testing but was unfortunately lost to followup.

In Rubinstein-Taybi syndrome, retinal and electrophysiological abnormalities occur more frequently with age, which emphasizes the importance of visual function tests and electrophysiological investigations at regular intervals.[3]

CONCLUSION

In majority of cases, vision is poor and the loss is irreversible because of conduction pathway defects and progressive retinal thinning.[4]

There is no specific treatment available for this plurimalformative syndrome.

Interventional guidance, behavioural training and special learning tools(Braille, audiovisual gadgets) could be of use.

Patients and their families are recommended to undergo genetic counselling.

In this case, the patient is on treatment for associated cardiac abnormality and is undergoing physiotherapy and speech therapy.

The rarity of the syndrome, the nonavailabilty of a specific treatment and the debilitating systemic associations call for the need for early intervention, thorough examination and regular followups.

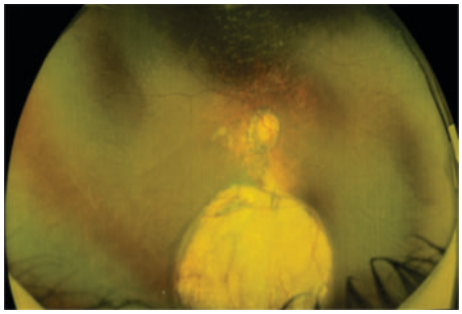

Figure1: fundus Picture Of Right Eye Showing Inferior Retino choroidal Coloboma

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