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Dr. Kanika Gulati
Department of Paediatrics, Dr. D. Y. Patil Medical College, Pimpri, Pune, Maharashtra, India

Dr. Renuka Jadhav
Professor, Department of Paediatrics, Dr. D. Y. Patil Medical College, Pimpri, Pune, Maharashtra, India

Dr. Sharad Agarkhedkar
Professor and Head, Department of Paediatrics Dr. D. Y. Patil Medical College, Pimpri, Pune, Maharashtra, India

Correspondence
Dr. Kanika Gulati
Department of Paediatrics, Dr. D. Y. Patil Medical College, Pimpri, Pune, Maharashtra, India

Peters' plus syndrome like phenotype with ring chromosome 21

Dr. Kanika Gulati, Dr. Renuka Jadhav and Dr. Sharad Agarkhedkar

Abstract
Peters' Plus Syndrome is an autosomal recessive disorder characterized by Peters' anomaly, cataract, short stature, dysmorphic facial features developmental delay and intellectual disability. Ring Chromosome 21 is a rare chromosomal disorder in which the affected infant has breakage of chromosome 21 at both ends and ends of chromosome join together to form a ring which manifests as mental retardation, abnormalities of face, eyes and/or internal organs.
A 4year old male child was brought with complaints of inability to see, 3 episodes of seizures and dysmorphic features. Patient also had history of delayed milestones. On Examination all vital parameters were normal. Dysmorphic features included broad nose, low set ears, retrognathia, cupid shaped upper lip. On auscultation there was a systolic murmur. Height less than -2 SD. Ophthalmic examination showed bilateral corneal opacity. MRI Brain showed periventricular leukomalacia and thinning of corpus callosum. 2D ECHO showed 4mm atrial septal defect. BERA suggestive of bilateral mild hearing loss. EEG was normal. Cytogenetic studies showed a ring chromosome 21 and loss of one chromosome 21. I could not carry out the study for B3GALTL mutation which is associated with the syndrome. Thus the diagnosis of Peters' Plus syndrome like phenotype with ring chromosome 21 was considered.

Keywords: peters' plus syndrome, krause-kivlin syndrome, ring chromosome 21, corneal opacity

Introduction
Peters' Plus syndrome is an autosomal recessive disorder characterized by Peters' anomaly, cataract, short stature, dysmorphic facial features, developmental delay and intellectual disability. Ring chromosome 21 is a rare chromosomal disorder in which the affected infant has breakage of chromosome 21 at both ends and the ends of chromosome join together to form a ring which manifests as mental retardation, abnormalities of the face, eyes and/or internal organs.

Case summary: A 4 year old male child was brought with complaints of inability to see, 3 episodes of seizures since 4 months and abnormal facial features and has history of delayed milestones. On examination vitals are within normal limits. Height was less than -2SD. Dysmorphic features include broad nose, low set ears, receding chin, high arched palate, retrognathia, cupid bow's shaped upper lip, long philtrum. On auscultation, there was a systolic murmur. Ophthalmic examination showed bilateral corneal opacity.

Investigations
1. MRI Brain: Periventricular leukomalacia and thinning of corpus callosum.
2. 2D ECHO: 4mm atrial septal defect.
3. BERA: suggestive of bilateral mild hearing loss.
4. Cytogenetic studies: showed a ring chromosome 21 and loss of one chromosome 21.
5. EEG: Normal

Fig 1: Affected patient having dysmorphic features- broad nose, low set ears, long philtrum, cupid bow's shaped upper lip

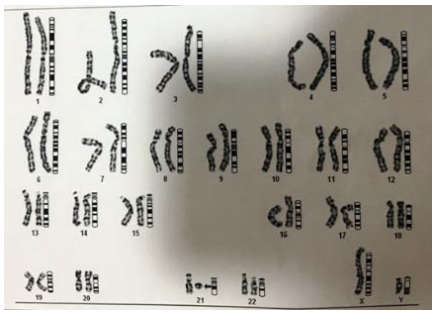

Fig 2: Cytogenetic study showing male karyotype- mos45, XY,-21[04]/46,XY,r(21)(::p11.1→ q21) [16]

Discussion

Peters plus syndrome is an inherited condition that is characterized by eye abnormalities in the anterior segment, short stature, distinctive facial features, and intellectual disability. Developmental milestones, such as walking and speech, are delayed in most children. Mutations in the *B3GALTL* gene cause Peters plus syndrome. The *B3GALTL* gene helps in the synthesis of enzyme called beta 3-glucosyltransferase (B3Glc-T), which is involved in the glycosylation [1].

The symptoms and findings associated with Ring Chromosome 21 may be extremely variable, depending upon the amount and location of genetic material lost from the 21st chromosome. As a result, some individuals with Chromosome 21 Ring may have few or no associated symptoms, whereas others may have severe physical abnormalities and mental retardation [4].

Conclusion

Peters' plus syndrome is a rare disorder; its incidence is unknown. Fewer than 80 people with this condition have been reported worldwide. In this case the ophthalmic examination, the dysmorphic features and presence of developmental delay along with intellectual disability are suggestive of Peters' plus syndrome. However, due to lack of gene mutation study, the diagnosis reached is Peters' plus syndrome like phenotype with presence of ring chromosome 21 as seen in the cytogenetic studies. The patient was treate for seizures with carbamazepine. Behavioral therapy was initiated.

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