LAURENCE MOON BARDET BIEDL SYNDROME – A RARE CASE PRESENTATION

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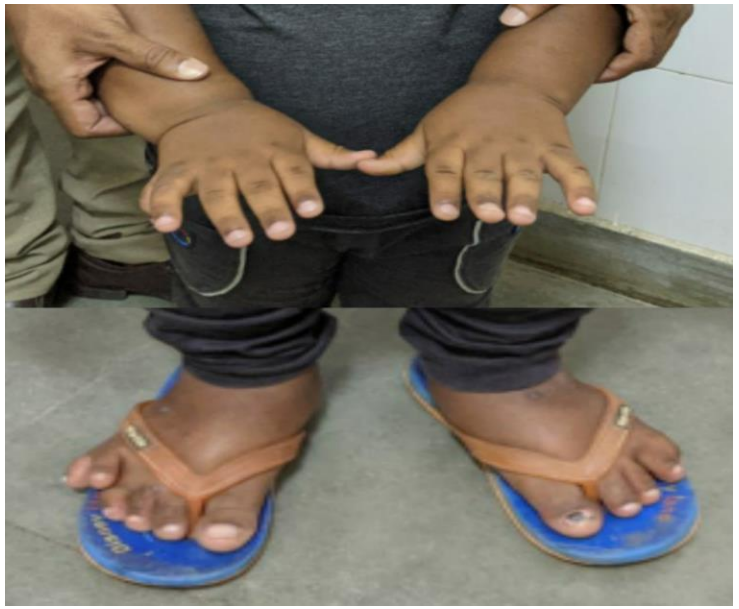
INTRODUCTION

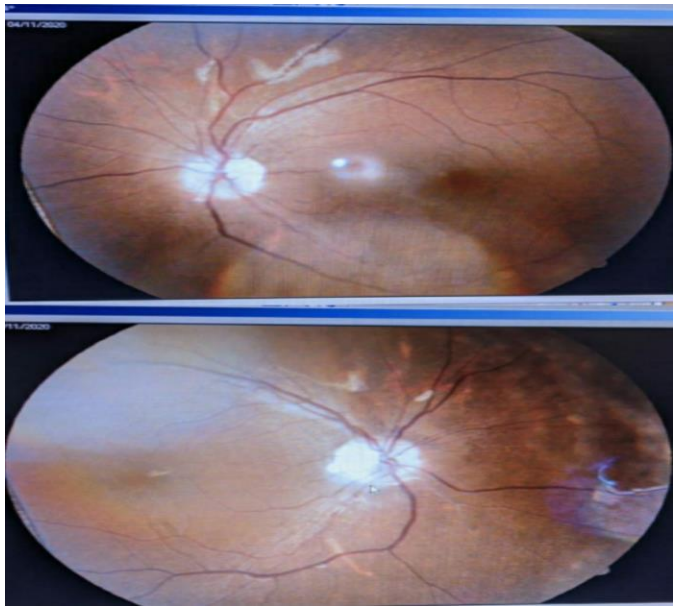
- Laurence moon bardet biedl syndrome is an autosomal recessive genetic disorder with variable expressivity and a wide range of climical variability observed both within and between families.It includes Laurence moon syndrome with features of retinitis pigmentosa(rod-cone dystrophy), mental retardation, hypogonadism in males. We report a typical case of Laurence moon bardet biedl syndrome in a male showing all ocular features, most of the general features and a typical family history. There is a typical onset of blindness in this case.

Case report

- Here is a case of 4 year old male who came to our OPD, ophthalmology department, PESIMSR kuppam, with complaints of blurring of vision, jerky eye movements , night blindness since 1month which progressed gradually.
- He is born of consanguineous marriage , had developmental delay and mentally retarded. The other sibling and parents were normal but there is family history of similar presentation in one of their relatives.
- Systemic examination revealed obesity, polydactyly(hexadactyly) and syndactyly in both upper and lower limbs, ataxic gait and hypogonadism, epicanthus inversus in both eyes.
- Ophthalmic examination revealed only perception of light in both eyes with nystagmus. On slit lamp examination there is posterior subcapsular cataract in both the eyes.

- Fundus examination revealed pale, waxy optic disc with attenuation of arterioles and bony corpuscular pigment seen at periphery giving the impression of primary typical retinitis pigmentosa with consecutive optic atrophy.





Discussion/ Conclusion

- Laurence Moon Biedl Bardet syndrome was first defined by Bardet in 1922 as an autosomal recessive disorder characterized by structural and functional abnormalities of organs and tissues with diverse embryonic derivation. The five cardinal features of syndrome include polydactyly, pigmentary retinopathy obesity mental retardation and hypogonadism. Other systemic features include short stature congenital heart block deafness and neurological disorders Laurence Moon Biedl Bardet syndrome is a rare genetic disorder. Biedl in 1922 added mental deficiency and genital hypoplasia to this syndrome.
- We report a typical case of laurence moon bardet biedl syndrome in a male of 4 years old with significant family history. He presented with an early onset blindness and all ocular features like retinitis pigmentosa, optic atrophy, posterior subcapsular cataract and nystagmus. He also showed characteristic general features of obesity, polydactyly, syndactyly. He also had mental retardation and developmental delay.

Thankyou