Bardet-Biedl Syndrome: An Underreported Genetic Syndrome

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Abstract- We report a case with Bardet-Biedl syndrome. The presenting features and Its diagnosis are discussed. The need for prompt diagnosis and also the need to report such rare cases was discussed

Index Terms- bardet-biedl syndrome, retinitis pigmentosa, developmental delay

I. INTRODUCTION

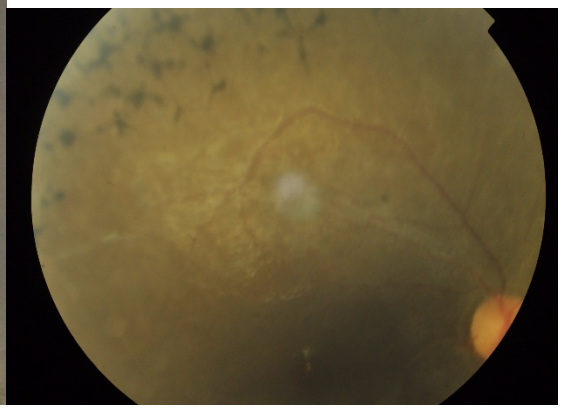
Bardet-Biedl syndrome is a rare autosomal recessive genetic disorder.

The prevalence of BBS has been estimated in different populations, ranging from 1 in 160,000 in European populations to 1 in 13,000 in Bedouins from Kuwait[1]. Less than 15 cases have been reported from India.The incidence is higher in some populations with a high level of consanguinity or geographically isolated population, with disease incidence of 1 in 13,000 in the isolated populations of Newfoundland(Canada) and Kuwait, 1 in 17,000 births. It is characterized by clinical manifestations of primary features like Rod-Cone Dystrophy, Polydactyly, Obesity, Genital abnormalities, Renal Defects, Learning Difficulties. Secondary characteristics are Developmental Delay, Speech Deficit, Brachydactyly, Ataxia, Olfactory Deficit, Diabetes Mellitus, Congenital Heart Disease[2]

II. CASE REPORT

A 13 year 8 months old male child born to a couple with 2nd degree consanguineous marriage presented to Paediatric OPD of Dr.PSIMS & RF with alleged complaints of Small testes (since birth), Decreased vision in both eyes (since 1 year of age) with excessive weight gain (since 7 years of age) and learning difficulties (since 8 years of age).

On Examination he had Hexadactyly on all four limbs, Absent Pubic Hair with Small Testes, Decreased visual acuity bilaterally, Fundus Examination: Bilateral Retinitis Pigmentosa, Ejection systolic murmur present over pulmonary area, developmental delay



III. Conclusion

1)Early diagnosis is delayed as presenting features appear at different point of time & Reporting of case can get delayed

2)Multiple specialists see along course of disease & follow up with single doctor may not be present.

3)Awareness about reporting and its impotance needs more emphasis

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