Research Article

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Delayed Speech and Language in Bardet - Biedl Syndrome - A Case Report

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ABSTRACT

Bardet-Biedl Syndrome (BBS) is a human genetic disorder that produces many effects and affects many body systems. The wide clinical spectrum observed in BBS is associated with significant genetic heterogeneity. This article includes a case of Bardet- Biedl syndrome in a 2 years 11 months old girl from Hubli, a town in north Karnataka region of India. A detailed speech, language and hearing profile based on systematic evaluation have been highlighted in the present study.

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Introduction

Bardet-Biedl syndrome (BBS) sometimes known as Laurence-moon- Bardet Biedl syndrome, is a rare autosomal recessive ciliopathy characterized by rod-cone dystrophy, learning disability, polydactylism, obesity, genital malformations, and renal abnormalities [1]. It is one of the most well studied conditions in the family of diseases caused by defective cilia collectively known as Ciliopathies. It is a pleiotropic disorder and has a prevalence of around 1:100,000 in North America and Europe [2].

The symptoms of Bardet Biedl syndrome includes, progressive vision loss due to deterioration of the retina, Extra finger next to the pinky finger, Kidney problems, Obesity that develops around 2-3 years of age, abnormalities of the genitalia, infertility due to hypogonadism, learning disabilities, diabetes, speech and language problems, high blood pressure, heart defect, behavioral disorders, distinctive facial appearance, dental anomalies [3-5]

A study described 32 patients with this disorder [6]. 28 patients had severe retinal dystrophy. Polydactyly was present in 18 of 32 patients, but syndactyly and brachydactyly were present in all. Obesity was present in all. Only 13 of 32 patients were considered to be mentally retarded. Another study on a 14-year-old boy reported to the skin outpatient department showed clinical features of severe obesity, increased blood pressure, mental retardation, lower limb weakness. Visual acuity was 6/36 in both eyes. The child also had bilateral conductive deafness of moderate degree [7].

Previous studies do not discuss in detail the speech language and hearing characteristics of children with BBS which have been included in the present study and also there are no studies or case reports from India reporting about Bardet Biedl syndrome cases in children. The present study defines the speech language, communication and hearing aspects of BBS in detail which gives an insight to the communication deficit associated with BBS.

Case Presentation

The patient was a 2 year 11 months old female from Hubli, a town in North Karnataka region of India. Parents approached JSS Institute of Speech And Hearing, Dharwad with the complainant of limited speech output.

Medical History

The child was diagnosed as Bardet-Biedl syndrome due to the presence of night blindness and severe obesity by the pediatrician. History of hypoxia was also noticed.

Speech and language evaluation

Detailed case history included a structured interview with the parents, clinical interaction with the patient and physical examination. Parents were first cousins indicating second degree consanguinity. Pedigree analysis was carried out where it was found to be an autosomal recessive disorder (Figure 1.)

Figure 1: Showing second degree consanguinity

The child was born after full term of pregnancy. Parents reported a history of miscarriage. Developmental history revealed delayed motor and speech and language milestones development.

Language and communication

Detailed speech and language assessment was carried out by administering several standardized tests such as Receptive Expressive Emergent Language Scale (REELS) and 3 Dimension Language Acquisition Test (3D-LAT). Results of REELS showed

delay in both receptive and expressive language skills. 3D-LAT also showed delay in receptive, expressive and cognition skills.

Hearing Evaluation

Otosopic examination was done and it showed Bilateral normal tympanic membrane Tympanometry and reflexometry was done to assess the functioning of middle ear. Bilateral 'A' type tympanogram indicated normal middle ear functioning. Presence of ipsilateral and contralateral reflex thresholds signified normal reflex pathway.

Transient Evoked Oto Acoustic Emission (TEOAE) test was done to evaluate outer hair cell functioning. Bilateral presence of TEOAE's indicated normal outer hair cell functioning.

Diagnosis and treatment

Based on history collected from the patient's family, physical examination, findings of other disciplines and results of the both objective and subjective tests, the client was diagnosed as Delayed speech and language secondary to Bardet Biedl syndrome. Parents were counseled about continuous speech language therapy so as to improve the overall communication and development.

Discussion

A study done on a 3 year old boy with Bardet Biedl Syndrome showed decreased renal function, hypertension, obesity and developmental delay. Electroretinography showed reduced retinal function suggesting a diffuse retinal disorder [8]. Another study revealed that 46% of 13 pediatric patients with BBS showed progressive loss of visual acuity, ophthalmoscopic signs of pigmentary retinopathy and speech and language delay [9]. The above studies are in agreement with the results of the present case report.

Conclusion

The current study showcases a detailed speech language and communication profile of a child with BBS. As BBS is a multifaceted disorder it is important to know in detail about all the deficits and also to treat them accordingly. Speech and language deficit is also one of the major manifestations of BBS and should be provided the appropriate rehabilitation so as to reduce the communication difficulties the child may face in the society. More research and documentations are the need of the hour which will further help us to understand the disorder better.

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