Asia Pacific Journal of Research
A peer reviewed international Journal
IMPACT FACTOR : 6.58
Print -ISSN -2320 - 5504
Online-E-ISSN - 2347 - 4793

AUDIOLOGICAL, SPEECH-LANGUAGE ASSESSMENT AND MANAGEMENT IN NOONAN SYNDROME: A LONGITUDINAL CASE STUDY

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ABSTRACT

Noonan Syndrome (NS) is characterised by short stature, typical facial dysmorphology and congenital heart defects. The incidence of NS is estimated to be between 1:1000 and 1:2500 live births. It is associated with hearing loss and speech problems. In approximately 50% of cases the disease is caused by mutations in the PTPN11 gene (www.betterhealth.vic.gov.au).
Case Report
We present longitudinal case report of a boy, 13 years old with complaint of hearing and speech problem. The case was under observation and rehabilitation since last eleven years. The general examination of the head and face revealed that the ears were low-set. Audiological and speech language examination revealed a profound sensorineural hearing loss bilaterally with deficient speech and language skills. Genetic analysis confirmed the diagnosis of Noonan syndrome. An attempt has been made in this paper to highlight on one of the associated problems that is hearing loss and consequent speech-language delay. Early and regular monitoring through Audiological and Speech language assessment and management can lead to better rehabilitation.

Key words: multiple congenital anomalies, audiological manifestations, speech manifestations

Introduction

Noonan Syndrome (NS) is a rare autosomal dominant inherited disorder which means that the gene (and the condition) can be transmitted from parent to child. It was known under various names such as male Turner's syndrome, Turner-like syndrome or Turner syndrome with normal karyotype, when in 1963 Dr. Jacqueline Noonan defined these changes as a specific syndrome which was named after her (Noonan and Ehmke, 1963). The incidence of NS is different and ranges from 1/1000 to 1/2500 live births (Allanson, 1987 and Sharland, Burch, Patton, 1997). Sharland et. al. studied 151 patients with NS and found incidence of hearing loss in them as 40 %. The disease is characterized by short stature, facial abnormalities, congenital heart defects and urogenital malformations. Less frequently the disease may be accompanied by delayed psycho-motor development. Also, in these patients, especially in childhood, disorders of coagulation factors (VIII, XI, XII) and thrombocytopenia are common. There is a great variability in expression and the phenotype becomes less pronounced with increasing age (Allanson , Hall , Hughes, Preus, Witt, 1985). It is a genetic condition caused by a fault in one of at least seven different genes. In genetic testing, a mutation in the PTPN11 gene causes Noonan syndrome

in about 50 per cent of affected people *(www.betterhealth.vic.gov.au)*. It was once believed that most cases of Noonan syndrome were sporadic, which means the child's gene spontaneously changed. However, researchers now suspect that Noonan syndrome is inherited in up to 75 percent of cases *(www.betterhealth.vic.gov.au)*.

Case Report

We present here the case of a 13year old boy who had reported to AYJNIHH (ERC) at the age of 2yrs2months with a complaint of speech and hearing problem. A test battery of audiological tests comprising of BOA (Behavioural Observation Audiometry), Impedance Audiometry, OAE (Otoacoustic Emission), ABR (Auditory brainstem evoked audiometry) were done. The Behavioural Observation reports were indicating normal hearing which did not co-relate with the OAE (TE) findings indicating refer for both ears. The child was recommended for observation speech therapy. The child underwent another BOA which revealed moderate to moderately severe hearing loss. An ABR assessment was recommended. Immittance revealed bilateral 'A' type tympanogram with absence of reflexes bilaterally. A complete speech and language evaluation was done for the case. The Receptive expressive emergent language scale (REELS, Bzoch and League,1971;Bzoch,League and Brown,2003) test was administered which reveals a Receptive Language age (RLA) of 12-14 months and Expressive Language age (ELA) of 7-8 months. Child has an average developmental progress and average intelligence as per the psychological reports. The child had some typical features like short stature, facial abnormalities, congenital heart defects. Child was sent to Medical college, Kolkata for karyotyping. A normal male karyotype type (46 XY) was determined, but the clinical examination of the child pointed to Noonan syndrome. Genetic testing of the child and parents confirmed PTPN11 gene mutation. The ABR testing done revealed no identifiable peak V at 109 dBnHL indicating profound sensorineural hearing loss. The child was recommended a Body Level Hearing Aid pseudobinaurally fitted.

He underwent speech therapy for 2years and the outcome was a significant improvement in speech as per the REELS. An RLA of 27-30 months and ELA of 24-27 months.

At 4yrs of age another complete audiological, speech & language evaluation was conducted comprising of puretone audiometry which revealed a severe degree of hearing loss correlating with the previous findings. The child this time was recommended and fitted with a semidigital BTE Hearing Aid binaurally.

The child underwent speech therapy for 4yrs after which at the age of 9yrs, complete speech and language was re-evaluated and was diagnosed as Delayed speech and language development secondary to hearing loss. Three dimentional language acquisition test (3DLAT, Herleker and Karanth,1994) was performed in which ELA, RLA and cognition improved to above 33-36 months. The Weiss and Curtis Development of Language & Speech (1936) revealed an expressive behavior of 66months and receptive behavior of 72 months. The Linguistic Profile Test (LPT, Suchitra and Karanth,1993) was also done that shows RLA of 6-7yrs and ELA of 5-6 years. The Communication Deall (Karanth, 2007) reveals expressive vocabulary of 60-66 months and receptive vocabulary of 54-60 months.

At 13 years of age the child underwent puretone audiometry once again and the audiological findings revealed a bilateral profound sensorineural hearing loss. The child was recommended BTE Hearing aid binaurally which the child is presently using. Case was diagnosed as Deficient Speech (misarticulation) and Language skills secondary to hearing loss. The Linguistic Profile Test was also done that shows RLA of 6-7yrs and ELA of 5-6 yrs. Immittance audiometry revealed bilateral 'A' type tympanogram with absence of reflexes.

Discussion

Eleven years of observation and rehabilitation of the case indicates that the degree of hearing loss has progressed through moderate to severe to profound sensorineural hearing loss as the case has grown from 2years to 13 years of age. This is depicted in figure.1.

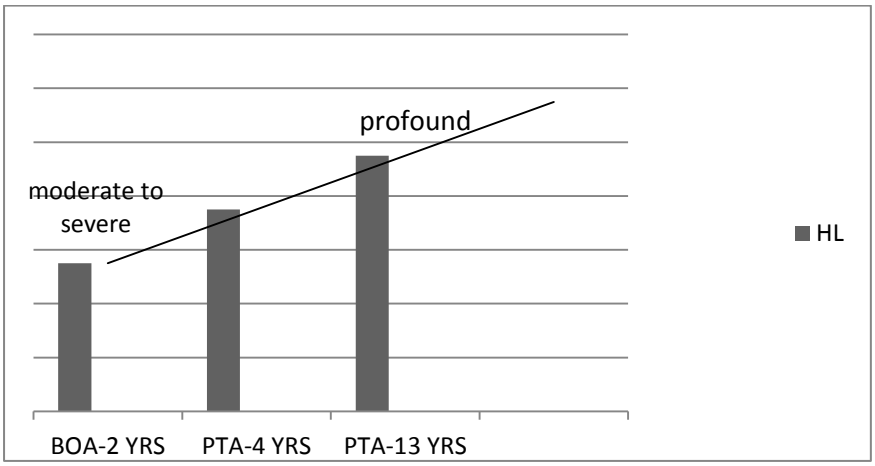

Figure.1: Increase in degree of hearing loss from 2 to 13 years of child's age.

BOA- Behavioral Observational Audiometry. PTA- Pure Tone Audiometry.
HL- Hearing Loss

On the other hand the speech-language developmental history reveals that the expressive and receptive language age was 7-8 months and 12-14months respectively at 2years 2months of age whereas at 4years of age the expressive and receptive language age was 24-27 months and 27-30 months respectively. At 9 years of age the expressive and receptive language age has reached to 5-6 years and 6-7years respectively in LPT and when the child has reached 13 years of age, the ELA and RLA was same as it was at 9 years of age. This indicates that though there is valuable increase in expressive and receptive language age it seems it has reached a plateau since last 2years of rehabilitation as it is depicted in figure.2. This may account for other physiological abnormalities which the child is developing nowadays and because of which he also has to skip speech therapeutic sessions.

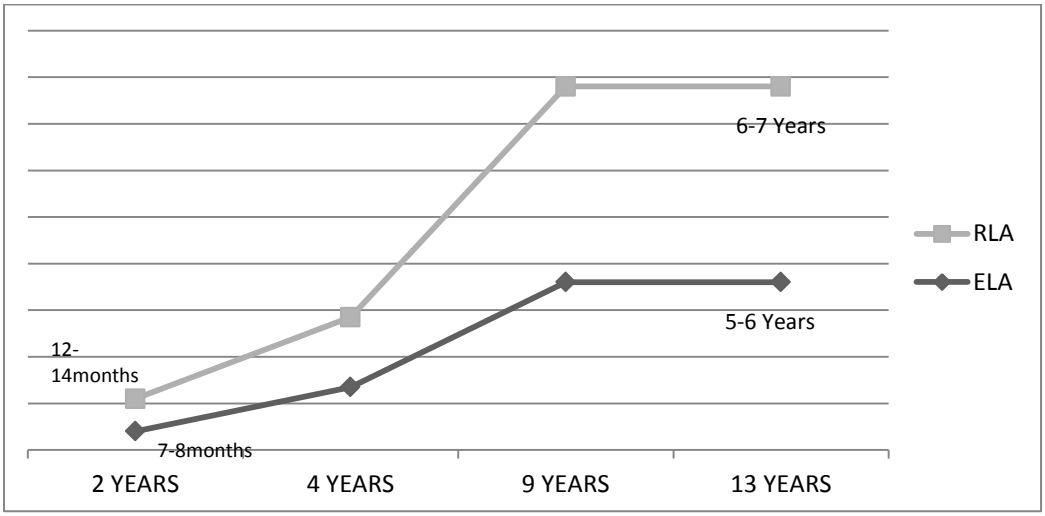

Figure.2: Development in receptive and expressive language age (RLA & ELA) across 11 years of time span

Noonan Syndrome includes a cluster of multiple congenital anomalies, which are easy to recognize clinically. It has been associated with conductive hearing loss as well as sensorineural and vestibular anomalies (Foster and Dyhrkopp, 2016). The clinical diagnosis becomes more and more difficult as the changes are not easily noticed as the child grows, and the risk of consequences of untreated anomalies such as heart defects, chest wall deformities, blood disorders, delayed psychomotor development, ocular defects etc increase during the growth. Hearing loss due to otitis media is a frequent complication (15%–40%). Sensorineural hearing loss is less

common, but involves the low frequency range in 10% of patients and the high frequency range in 25% of patients (Qiu, Yin and Stucker 1998). Structural anomalies of the inner ear have occasionally been reported (Cremers , van der Burgt and CJAM,1992), (Naficy, Shepard and Telian, 1997) and vestibular abnormalities have been described in a single case (Martinez, 1997). Qiu, Yin and Stucker,1998, reviewed 20 cases of this syndrome with regard to hearing sensitivity and middle ear status. An incidence of progressive sensorineural hearing loss at the high frequencies is found for 50% of the ears. It is emphasized that early audiologic management may improve the quality of life for patients with Noonan syndrome.

Given such a high incidence of hearing anomalies, the audiologist and speech therapist can be a very important link in the diagnostic chain. It is important to do genetic testing whenever possible. In approximately 50% of these patients there is a mutation of PTPN11 gene.

Conclusion

This is a rare syndrome with heterogeneous group of changes in various organs. It requires teamwork in order to provide early diagnosis and treatment of numerous malformations. Hearing loss is one of the anomalies associated with this syndrome which can be diagnosed at the earliest. Although the loss might not occur at infancy but as the child grows it gradually develops. A proper test battery approach in relation to audiological tests, speech language assessments and appropriate guidance and counseling can bring optimal rehabilitation if the child is identified at an early age. Rehabilitation requires an ongoing regular checking and monitoring on all the domains of the disorder. The prognosis of these cases is satisfactory as they can lead a normal life if they get proper assessment and treatment through life.

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