



DEVIANT SPEECH-LANGUAGE PROFILE AND AUDITORY DYS-SYNCHRONY ASSOCIATED WITH NEUROFIBROMATOSIS TYPE 1: A SINGLE CASE STUDY

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ABSTRACT

The present single case study aims to investigate the myriad of potential speech, language and audiological characteristics along with exploring and getting an insight into the diagnostic procedure of a child with Neurofibromatosis Type 1 (NF-1). Evaluation was conducted by a speech language pathologist, audiologist and psychologist to study different areas of ability (intellect, hearing capability, speech-language skills and motor function) and performance (academic achievement and behavior). Psychological evaluation depicted that the child's verbal IQ is more affected than performance IQ, has problems in attention, social interaction, coordination, manual dexterity and balance. Speech and Language evaluation depicted deviancy in syntactic and semantic skills, errors of substitution and distortions among all the consonants and blends, (due to poor oral motor co-ordination), severely affected speech intelligibility and hoarse vocal quality. Audiological evaluation revealed hearing sensitivity within normal limits bilaterally. DPOAEs (SNR > 6 dB) were present and robust in both ears. Although right ear had normal ABRs, but left ear ABR finding was abnormal (waves I, III and V were unidentifiable), along with presence of large and prolonged cochlear microphone, thereby indicating the presence of auditory neuropathy/dys-synchrony (AN/AD) in the left ear. Thus, deficits in speech, language, hearing, cognition and motor skills occurs in children with NF1.

Key Words: Neurofibromatosis Type 1, Deviant Language, Speech Errors, Auditory Neuropathy/ Dys-synchrony.

Introduction:

Neurofibromatosis Type 1 (NF-1) is an autosomal dominant disorder with variable expression resulting from a mutation in chromosome 17 that has the potential to affect multiple organ systems. A wide variety of clinical manifestations have been documented, ranging from severe cosmetic disfigurement and brain tumors to café-au-lait spots and freckles (Kayl & Moore, 2000). Research is suggestive of the fact that this population is generally associated with low average to average intellectual ability, along with a high risk for developing learning disabilities and attention difficulties. Neurocognitive profile indicates predominant weakness in the development of visuospatial skills, fine motor skills, and executive functioning. (Johnson, Wiggs & Stores, 2005; North, 2000).

A deficit in language is the characteristics of school-aged children with NF1. Studies that have examined language in NF1 reveal a delay in some aspect of language (Dilts et al. 1996; Eldridge et al. 1996; Hofman et al, 1994; Hyman et al. 2005). Dilts et al. (1996) examined the language abilities in 19 children with NF 1 (aged 6-17 Years of age) and compared their performance to age matched unaffected siblings. They found that 58% of their sample failed the expressive language screening test, 26% of whom also exhibited receptive language deficits. Hyman et al. (2005) examined the receptive and expressive language abilities of 81 children with NF1 and found presence of language delay. A few studies have highlighted difficulties with receptive and expressive language, phonological awareness, non sense word decoding, single word reading, and associated linguistic memory (Billingsley et al., 2003) among the NF 1 population. Deficits on tests of picture naming, receptive grammar, written language, and phonological processing have been identified by Hyman et al (2005) among NF1.

Lorch et al (1999) examined the speech characteristics of 30 adults with NF1 and reported rates of speech disorders in spontaneous speech as follows: hypernasality (37%), mild articulation difficulties (17%), atypically fast rate of speech (40%), and atypical volume (27%).

From the perspective of clinical hearing science, it is pertinent that investigation of auditory system function was not systematically addressed in NF1 even as late as 1987 by the medical community (Neurofibromatosis Consensus Statement, 1988). Hearing loss can occur in NF1, but is not common, and deafness is rare. A sensorineural loss may be due to a structural problem in the inner ear, or a neurofibroma may actually block the outer or middle ear resulting in a conductive loss. These types of losses are different than the loss due to acoustic neuromas (tumors of the auditory nerve) in NF2. It has also been noted that children with NF1 have an abnormality in the way their brains receive nerve impulses from the ears, but there is no proof that this causes hearing problems. Although uncommon, but NF1 can frequently involve portions of the auditory system in diverse and subtle ways in which no characteristic audiologic findings can be discerned (Pikus, 1995).

Additional concerns include social problems, anxiety, depression, withdrawal, and obsessive- compulsive behaviors (Dilts et al, 1996).

Need of the Study:

A given practitioner is unlikely to see more than one or two children with such rare disorders throughout the professional life. There is thus a critical need of sharing the information gathered with community practitioners and other professionals, with encouragement to collect local data about the feasibility and effectiveness of the suggested diagnostic protocols. In this way, better advantage of individual creativity and innovation can be taken, while at the same time contributing towards evidence based research and knowledge.

Aim of the study:

To investigate the myriad of potential speech, language and audiological characteristics along with exploring and getting an insight into the diagnostic procedure of a child with NF-1.

Methodology:

A 10 year old, non right handed male case was considered as the subject of this study. Based on the phenotypic characteristics, the case was diagnosed as NF-Type 1 by the Paediatricians, from where they were referred to the department of Speech Language Pathology with chief complaint of being unable to communicate age appropriately as reflected by predominance of using words (which are slurred in quality) accompanied with defined gestures. Medical History revealed significantly relevant peri and post natal history. Perinatally the child had preterm normal delivery at Hospital with low birth weight and postnatally the child underwent surgery of neurofibromas on left Gluteal Region at the age of 5 years. No positively significant family history had been reported. Educational History revealed that the child was attending in regular school, with poor scholastic performance. Auditory behaviors were indicative of absence of any suspected hearing loss. The child was screened negative for presence of any sclianemna and visual

acuity problem by the Ophthalmology Department, while on the other hand was diagnosed as hemiplegic C.P with neurofibromatosis by Physiotherapy Department.

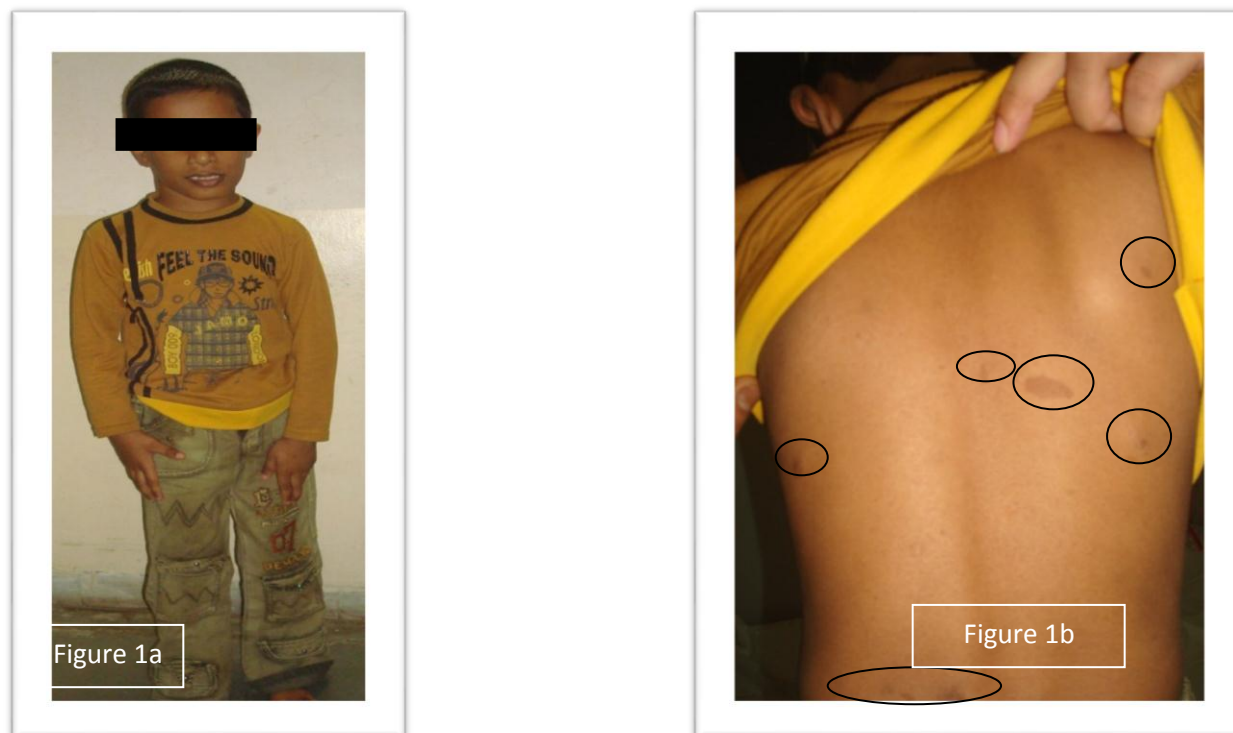


Figure 1(A) and 1(B): Showing features of NF-I.

Taking into consideration the phenotypic characteristics of the subject, we decided to do a detailed multidisciplinary investigation by a team of Speech Language Pathologist, Audiologist and Psychologist, so as to get an overview of such rarely reported cases of NF 1.

A Speech Language Pathologist evaluated receptive and expressive language skills, articulation, voice, speech intelligibility and academic achievement using Hindi version of Linguistic Profile Test: LPT and Picture Articulation Test: PAT (Ali Yavar Jung National Institute for the Hearing Handicapped and Regional Rehabilitation Training Centre Madras, India, under UNICEF sponsored project, 2004), Dr. Speech Version 4 (Vocal Assessment for Windows, Version-4.30; 1998, Tiger DRS, Inc.), and Percentage of Consonants Correct (PCC; Shriberg & Kwiatkowski, 1994) respectively, along with investigation of structure, function and control systems of the speech mechanism.

Alongside, an audiological evaluation by an audiologist included otoscopy, pure-tone audiometry (air and bone conduction), immittance audiometry (tympanometry, acoustic reflex thresholds), otoacoustic emissions (OAEs), and auditory brainstem response (ABR) testing.

Puretone audiometry was performed using MAICO MA 53 in a soundproof room. Hearing impairment was defined as the level of Puretone thresholds averaged at 0.5, 1, and 2KHz. Hearing loss of 26–40 dBHL was considered mild, 41–60 dBHL moderate, 61–70 dB moderately severe, 71-90 dBHL severe more than 90 dB profound. Tympanograms and acoustic reflex thresholds(ART) were measured using a Madsen Zodiac 901 Middle-Ear Analyzer. Tympanometry was obtained using a 226 Hz probe tone, with a sweep pressure start point of +200 daPa and an end point of -400 daPa. Transient Evoked Oto-Acoustic Emission (TEOAE) and distortion product Oto-Acoustic Emission (DPOAE) were recorded separately by NEUROSOFT systems. TEOAE was measured using click stimuli with a nonlinear mode of stimulus presentation and was considered “present” when the signal-to-noise ratio was ≥ 6 dB and the confidence ratio was 80%. DPOAE was recorded with a fixed ratio of $f2/f1 = 1.2$ and fixed levels of 65 dB SPL and 55 dB SPL. A DPOAE pass criterion was presented at a particular frequency region when the signal-to-noise ratio was ≥ 6 dB.

The 100 μ sec click-evoked Auditory Brainstem Response (ABR) was recorded using a band pass from 100 to 3000 Hz and in a 10 ms time window with 2000 averages. Click stimuli were rarefaction and condensation clicks presented at the rates of 11.1 per second. The ABR was undertaken using a NEUROSOFT system and was considered absent if there was no repeatable response to click stimulus at 100 dB nHL, the maximum intensity of the equipment, and normal if a response was present at lower than 30 dB nHL. Cochlear Microphonic (CM) was obtained simultaneously with ABR tests. As reported by literature review, the most direct method of separating the CM and ABR was to compare responses obtained with rarefaction polarity stimuli with those obtained with condensation stimuli (Berlin et. al., 1998). CM follows the characteristics of the external stimulus; thus, the direction of the CM reverses with changes in polarity of the stimulus. ABR, however, may show only slight latency shifts with polarity changes and does not invert. Child was sedated before the test.

A psychologist evaluated the intellectual ability and behavior using Wechsler Intelligence Scale for Children-Revised (WISC-R; Wechsler, 1974), and Child Behavior Checklist (CBC; Achenbach, 1991) respectively. Motor abilities were assessed in two areas: visual motor integration and motor coordination by administering Berry-Buktenica Developmental Test of Visual Motor Integration (VMI; Berry, Buktenica, & Berry, 2010) and Henderson Test of Motor Impairment (TOMI-H; Scott et al., 1984) respectively.

Results & Discussion:

At presentation, the child had short stature [Figure 1 (A)], multiple café-au-lait spots [Figure 1 (B)], skin freckling, and multiple palpable plexiform neurofibromas along nerve roots along with history of operation of neurofibromas on left gluteal region.

Subjective examination of Speech Mechanism revealed normal appearance of all the structures except the tooth, which were broken along with presence of distocclusion and crossbite. The functionality (for both speech and non speech tasks) of lips and tongue along with the motor control systems of speech, were also affected. LPT suggested a receptive and expressive language age of 5.5 years and 4.5 years respectively, with deficits in both syntactic and semantic skills indicating delay in language development. PAT revealed predominant errors of substitution and distortions among all the consonants and blends, which were supposed to appear mainly due to poor oral motor co-ordination. Apart from that the speech intelligibility was also severely affected, reflected by a PCC of 42%. Dr Speech analysis revealed a hoarse vocal quality, with lowered fundamental frequency and increased perturbations. According to a study conducted within preschool children with NF 1, 68% exhibited delays in speech and/or language. Delays were demonstrated by 32% in articulation, 37% in receptive and expressive language each. 16% exhibited a voice disorder and 42% were judged to have a resonance problem (Thompson et al., 2010).

Results of audiological evaluation were quite interesting. The pure tone average (500 Hz, 1 KHz & 2 KHz) was 11.6 dBHL and 16.6 dBHL in right and left ear respectively with air-bone gap of lesser than 15 dBHL suggestive of hearing sensitivity within normal limits bilaterally. Tympanogram of 'A' type along with presence of ipsilateral ART bilaterally, suggested of absence of middle ear pathology for both the ears, though the contralateral ART of left ear was absent. DPOAEs (SNR > 6 dB) were present and robust in both ears for all test frequencies indicative of a good functional integrity of the cochlear outer hair cells bilaterally. On the other hand, although right ear had normal ABRs, but left ear ABR finding was abnormal (waves I, III and V were unidentifiable), along with presence of large and prolonged cochlear microphonic, thus indicating presence of auditory neuropathy/dys-synchrony (AN/AD) in the left ear. Although auditory dysfunction is characteristic of Neurofibromatosis Type 2; however, some limited evidence suggested that the auditory system may also be affected in 2.5% of subjects with NF1 (Barcelos Corse, 2005).

Findings of psychological evaluation were also remarkable. Result of WISC-R revealed a lowered full scale Intelligence Quotient (IQ), with verbal IQ being more affected than performance IQ. On CBC, the parents reported the child as having attention problems and difficulties with social interaction. VMI was indicative of average performance in both visual perception and motor coordination tasks while on TMI, the child had moderate problems with coordination, with main problem in manual dexterity and balance. Studies have also documented problems with dexterity, coordination, balance and gait as well as psychomotor slowing (Rosser & Packer, 2003).

Conclusion:

This study found that speech and language delays along with hearing, cognitive and motor deficits occurs in children with NF1, contributing towards later difficulties in academic achievement and social development. Two vital implications can thus be deduced. First, all children with NF 1 should have a thorough assessment with follow up intervention as needed and second a diagnostic protocol for this population should include contribution from multidisciplinary team members. Although results of a single case study can not be generalized across whole of the NF 1 population, so future research should replicate the protocol across a larger population, in order to determine the efficacy and document the variability.

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