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Speech and language profile of Apert syndrome: A single case report

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Abstract
Introduction: Apert syndrome is a rare congenital condition with a midface hypoplasia, symmetric syndactyly of the hands and feet, and autosomal dominant inheritance. Individuals with Apert syndrome had a high prevalence of language disorders, speech sound disorders and hearing problems.
Aim: The aim of the study was to document the speech and language profile of a client with a diagnosis of Apert syndrome.
Method: A detailed speech and language assessment was done for the client using informal and formal methods of evaluation.
Results and Discussion: The client was diagnosed to have expressive language disorder with speech sound disorder secondary to Apert syndrome. The most frequent and consistent articulatory error was substitution of lingua-dental sounds for lingua alveolar and lingua palatal sounds.
Conclusion: The present study would help students and professionals understand the speech and language profile of a client with Apert syndrome and assist in the assessment procedures to be followed and additionally contribute to rehabilitation strategies.

Keywords: Apert syndrome, speech, language, articulation

Introduction
Apert syndrome is a rare congenital condition with a midface hypoplasia, symmetric syndactyly of the hands and feet, and autosomal dominant inheritance as its defining features [1]. Since the infant mortality is high, the prevalence of Apert syndrome in the general population is lower and majority of cases are sporadic, with an incidence of 1 in 160 000 [2]. Reduced upper jaw size (maxillary hypoplasia), Angle Class III malocclusion, skeletal anterior open bite, dental crowding, unilateral crossbite, cleft palate, improper lip posture, uvula bifida in 30% of palates, upper and ogival palate, macroglossia, supernumerary teeth, and thick gums, retained teeth, delayed and ectopic tooth eruption are some oral characteristics of individuals with Apert syndrome [3, 4]. Individuals with Apert syndrome become mouth breathers as a result of nasopharyngeal and oropharyngeal attenuation, resulting in an anterior open bite [5].
Peterson (1973) [6] reported that many of them have severe oral structural abnormalities, significant hearing loss, and low intelligence quotient, all of which are known to negatively impact speech and language development. Elfenbein *et al*. (1981) [7] mentioned that the psychological effects of craniofacial dysmorphology may hinder social engagement, which would have a detrimental effect on speech and language development. Also, in this study, they reported that three children had difficulties with receptive and expressive language skills. Shipster *et al*. (2002) [8] described a case series of 10 children with Apert syndrome ranging in age from 4 years 1 month to 5 years 11 months, and reported that 8/10 of the children had moderate to severe language impairments. Due to their unusual oral structures, all of the children had articulation difficulties. Also, they reported that communication participation in individuals with Apert syndrome may be affected by the presence of a language disorder, speech sound disorder and/or hearing loss, social communication difficulties, or communication difficulties associated to hearing loss, in addition to any underlying cognitive deficits. Kilcoyne *et al*. (2022) [9] found that individuals with Apert syndrome had a high prevalence of language disorders, speech sound disorders and hearing problems.
As per our knowledge, only few published studies described the speech, language, hearing and communication deficits in Apert Syndrome in western context.

There is a scarcity of research studies related to the speech and language characteristics of Apert syndrome in the Indian context. Therefore, in this study, we are trying to highlight the speech and language-related issues in Apert syndrome, which will aid in assessment and intervention planning.

Case report

The client was a 13-year-old boy who was reported by his parents with the complaint of not speaking age appropriately and reduced speech clarity. He was exposed to both Tamil and English languages. He predominantly communicates in his mother tongue, Tamil. The client was exposed to English while attending the integrated school. He is right-handed. Medical history revealed that the client was diagnosed with Apert syndrome with bilateral syndactyly of fingers and toes. Radiological testing using Computerized Tomography revealed brachycephaly with midface hypoplasia. History of seizure was also reported. Developmental history revealed delayed motor and speech-language milestones. He had normal muscle tone. Psychological evaluation revealed average intellectual functioning. His mental age was found to be 11 years 4 months using Binet Kamat Test. Hearing evaluation revealed bilateral hearing sensitivity within normal limits.

Speech and language profile

A comprehensive speech and language evaluation was carried. Structures of the oral peripheral mechanism were examined, which revealed Angle class III malocclusion, bifid uvula, a high arched palate, crowding and disorganized teeth. However, functional examination of the oral peripheral mechanism indicated adequate movement of jaw, lips, tongue during non-verbal tasks Responses to verbal tasks indicated slightly constrained articulatory movements in terms of speed and range. No swallowing problems were reported. Vegetative skills such as sucking, chewing, biting, and blowing were present.

Diadochokinetic syllable rates and articulation tests were used to investigate the effects of articulatory deficits on a verbal task. The diadochokinetic syllable rate include alternating motion rate and sequential motion rate. The alternating motion rate of /pʌ/, /tʌ/, and /kʌ/ was 3.5, 3.5, and 3.3 syllables per second. The sequential motion rate of /pʌtʌkʌ/ was 1.6 syllables per second. The client's oro-motor deficit could be the basis of his poor diadochokinetic syllable rates. Tamil articulation test [10] was used to assess his articulation skill. Articulation test results revealed the substitution of /θ/ for /d/, /t/, /s/, /ʃ/, /tʃ/ and /dʒ/; substitution of /l̪/ for /l̠/; and the distortion of /r/. Also, during spontaneous speech, cluster reduction was observed. Client demonstrated good interpersonal sound discrimination and was stimulable in all modalities. His Maximum phonation duration was found to be age adequate (/a/-14s, /i/- 14s, /u/-15s).

Speech intelligibility of the client was assessed using speech intelligibility rating scale provided by AYJNIHH. He received a score of 2 which indicates that “speech can be understood with little effort, repetitions needed occasionally”.

Speech-language milestone of the client was reported to be delayed. The client said first word at the age of 2 years. The client was able to follow and execute multi-step verbal commands and identifies personal pronouns, adjectives, prepositions, descriptive words, and time. The client was able to express his needs verbally using three to four words sentences. The client can answer different question forms. The client uses adjectives, prepositions, pronouns during conversation. and can narrate events. He can use all question forms in conversation. Formal language assessment was carried out using Assessment of Language development (ALD; Lakkanna, Venkatesh, & Bhat, 2008) [11]. The result of ALD revealed that he passes the receptive language age criteria of 8 - 10 years, and the expressive language age corresponds to 7-8 years.

Diagnosis and intervention

Based on a thorough evaluation of speech and language skills using both informal and formal tests, the client was diagnosed with expressive language disorder with speech sound disorder secondary to Apert syndrome. Parents were counseled and recommended to consult a plastic surgeon and prosthodontist for correction of malocclusion and misalignment of teeth. They were recommended to attend speech and language therapy three times a week and follow a home training programme in order to improve his speech intelligibility and language skills.

Discussion

The client had Angle class III malocclusion, high arched palate, and bifid uvula, which is complemented by literature evidence [3, 4]. Also, this client had language difficulty and exhibited speech sound errors which is in line with previous studies as well. In this study, the client consistently substituted /θ/ for /d/, /t/, /s/, /ʃ/, /tʃ/, and /dʒ/. This could be because of malocclusion, dental misalignment, and maxillary hypoplasia; the client is substituting lingua-dental sounds for lingua-alveolar and lingua-palatal sounds.

Conclusion

The present study highlights the speech and language profile of a client with Apert syndrome. Students and professionals could benefit from this study by having a better understanding of the speech and language skills of Apert syndrome. Documentation will assist in understanding the nature, signs, and characteristics of Apert syndrome, as well as the assessment procedures to be followed and additionally contribute to rehabilitation strategies.

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