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Moebius syndrome: A single case study

Gayathri Purushothaman [1*], Chitra Alagusundaram [2], Jennifer Ramesh [3], Karunya [4]
Lecturer, Department of Speech, Hearing & Communication, NIEPMD, Chennai, Tamil Nadu, India
Speech Language Pathologist, Pebbles Rehabilitation Centre, Chennai, Tamil Nadu, India
Audiologist and Speech Language Pathologist, Beyond Multi – Rehabilitation Centre, Chennai, Tamil Nadu. India
Audiologist and Speech Language Pathologist, Chennai, Tamil Nadu. India

* Corresponding Author: **Gayathri Purushothaman**

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Abstract
Moebius syndrome is a rare congenital condition that is characterized clinically by complete or partial facial nerve palsy with or without paralysis of a group of cranial nerves. It is often associated with other malformations of the limbs and oro-facial structures. It presents with diverse manifestations that can have repercussion on the quality of life of the affected individuals. This study presents a clinical case of a child with the moebius syndrome in the perspective of speech language pathologist along with literacy characteristics and highlights the causes, signs and symptoms, milestones development, communication and feeding difficulties faced by the child and multi-disciplinary team approach to enhance the child's speech, language and literacy skills.

Keywords: Moebius Syndrome, speech, language and metaphonological skills, Multidisciplinary approach

Introduction
Moebius syndrome is a rare congenital condition that is characterized clinically by complete or partial facial nerve palsy with or without paralysis of a group of cranial nerves. It is often associated with other malformations of the limbs and oro-facial structures. It is a developmental disorder, characterized by absence or underdevelopment of the nerves that control facial (cranial nerve 7) and eye movements (cranial nerve 6).

Incidence and prevalence
Moebius syndrome affects males and females in equal numbers. The disorder is present at birth (congenital). The exact incidence and prevalence rates of Moebius syndrome are unknown. One estimate indicates the incidence at 1 case per 50,000 live births in the United States.

Causes
The causes of Moebius syndrome remain unknown, although the condition most likely results from a combination of environmental and genetic factors. Researches have not identified any specific genes related to the syndrome. The disorder; however, seems to be associated with changes in particular regions of chromosomes 3, 10 or 13 in some families. Certain medications taken during pregnancy, as well as abuse of drugs such as cocaine, might also be risk factors for Moebius syndrome. Although it appears to be genetic, its precise cause remains unknown and the medical literature presents conflicting theories. It affects boys and girls equally, and there appears to be, in some cases, an increased risk of transmitting the disorder from an affected parent to a child.

Signs and symptoms
Most people with Moebius syndrome have weakness or complete paralysis of the facial muscles.

Children and adults with facial paralysis may be unable to smile, frown, raise their eyebrows, close their eyelids or pucker their lips. This not only results in lack of facial expression but may also result in drooling and difficulty with speech. Infants can have difficulty with sucking and swallowing. Other features of Moebius syndrome can include:

- Motor delays due to upper body weakness
- Strabismus
- Dental problems
- High palate and Cleft Palate
- Hand and feet problems including club foot and missing or fused fingers (Syndactyly)
- Hearing Problems
- Dry eyes and Irritability

Although they crawl and walk later, most children with Moebius syndrome eventually catch up. Speech problems often respond to therapy, but may persist due to impaired mobility of the tongue and lips. As children get older, the lack of facial expression and an inability to smile become the dominant visible symptoms.

Diagnosis

A diagnosis of Moebius syndrome is based upon the characteristic signs/symptoms, a detailed patient history, and a thorough clinical evaluation. There are no diagnostic tests that confirm a diagnosis of Moebius syndrome.

Management

The treatment of Moebius syndrome is directed toward the specific abnormalities in each individual. Usually, these children are managed by a multidisciplinary team, often in a craniofacial center. The multidisciplinary team includes,

- Paediatrician
- Neurologist
- Plastic surgeons
- Opthamalogist
- Orthopedists
- Dental specialist
- Otolaryngiologist
- Audiologist
- Occupational therapist and Physiotherapist
- Speech language pathologist

Case study

Background information

XXX, 2.11 years old male child was accompanied by his parents to the department of Speech and Language pathology with the complaint of not speaking age adequately. Birth history was not significant. The child was born to non-consanguineous parents with no significant family history of communication delay or any other Speech and Language problems.

On observation the child has signs and symptoms of

- Strabismus
- Dental problems
- High palate
- Clubbed feet
- No hearing and vision problems

On evaluation,

Table 1: Developmental, Vegetative, Speech and language evaluation

Developmental milestones	Vegetative skills	Cranial nerve and OPME	Speech and language evaluation
Motor Milestones Motor milestones were reported to be delayed as the child achieved walking only by 2 years 4months.	Vegetative skills of Sucking, biting, chewing and swallowing were reported to be inadequate. Blowing skills were absent. Drooling was reported to be present occasionally.	OPME reveals that the child had puckered lips at rest; high arched palate, misaligned teeth with dental problems; over all, the oral cavity was observed to be smaller in size. Function reveals that the child has inadequate pursuing of lips, restricted lingual movements, inadequate VPC. Jaw movements were restricted.	The child is able to maintain eye-contact and can attend and concentrate to the verbal stimuli and will able to sustain on picture stimuli for 1- 2-minute duration. The child is able to comprehend simple verbal commands, family members, and emotions, able to discriminate edibles and non-edibles, no, bye-bye and few body parts. The child is able to express his needs non-verbally by pointing towards desired object along with the vocalization of /a/ and babbling sounds of /m/ and /n/.
Speech Milestones Speech milestones were reported to be delayed as the child achieved babbling by 11 months and first word is not yet achieved	Also, the parents reported that he had problem of sucking at the time of birth and had been alternated through spoon feeding and feeding tubes.	CN examination reveals that the Cranial nerves V, VI, VII, IX, XI and XII are affected.	The child was observed to have nasal air emission while vocalizing /a/ sound and the loudness was reported to be inadequate as reported.
Social and behavioral history reveals that the child is able to socially smile at others, able to mingle with peer groups, temper tantrums were present, and toilet indication was partially present. Parent child interaction was inadequate both quantitatively and qualitatively.			On formal evaluation the child's RLA: 12-14 months and ELA: 4-5 months according to REELS. Com-DEALL reveals GM:12-18 months FM:12-18 months ADL:12-18 months COG:12-18 months EMO: 12-18 months RL:12-18 months EL:0-6 months SOC:12-18 months

Diagnosis

The diagnosis was given as Mixed Receptive and Expressive Language Delay consequent to Moebius Syndrome as diagnosed by the Pediatrician and Neurologist.

Management

Hence the management will include team approach; where the Speech and language goals will focus on:

- To improve the child's feeding skills along with expression of speech sounds.
- Improving upper lip mobility is a primary goal for clients with Moebius syndrome since the inability to close or round the lips affects feeding safety as well as speech clarity.
- The initial goal is generally lip closure. In addition to horns, a series of graduated lip closure exercisers has been used successfully.
- Once lip closure has been achieved, the **"Straw Hierarchy"** can be introduced.
- This is used to teach the coordination necessary for safe swallowing and to improve jaw stability, lip rounding and tongue retraction. Oral-motor/feeding therapy has been used effectively for improving lip closure and lip rounding as a means of improving feeding skill levels and feeding safety.

Conclusion

Moebius syndrome is a rare congenital condition that is characterized clinically by complete or partial facial nerve palsy with or without paralysis of a group of cranial nerves. The cause is known. The case study highlights the characteristics of a syndrome, diagnosis and management. Since the condition is characterized by a group of features, a multidisciplinary team approach is essential in the diagnosis and efficient management of the condition

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