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VACTERL association accompanied by facial palsy: A rare paediatric rehabilitation case report

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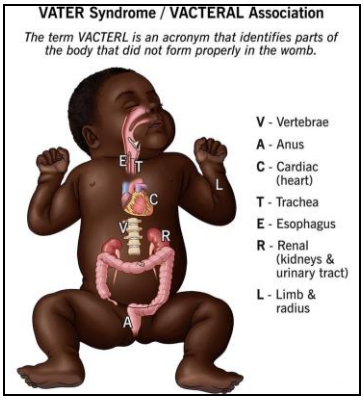
Abstract

Background: VACTERL syndrome (VACTERL association) is a group of growth abnormalities (birth defects) that happen in the early stages of embryo development during pregnancy. The term VACTER is an acronym that identifies parts of the body that didn't form properly during fetal development. Each case of VACTER syndrome is unique. Diagnosis is clinical and requires at least three component features, together with exclusion of overlapping syndromes such as CHARGE syndrome, Fanconi anaemia, Holt–Oram syndrome and oculo-auriculo-vertebral spectrum. The reported incidence is approximately 1 in 10,000 to 1 in 40,000 live births. Physiotherapy documentation of this condition, particularly where craniofacial involvement coexists, is limited. Case Presentation: A 9-year-old male child presented with difficulty in performing facial expressions, non-fluent speech, hearing difficulty and long-standing difficulty in passing stools. He had undergone anoplasty on the first day of life for imperforate anus. Additional findings included right-sided microtia, bilateral pre-axial polydactyly, hemivertebra at the cervicothoracic region and a patent ductus arteriosus with a small atrial septal defect that had resolved spontaneously on follow-up two-dimensional echocardiography. Right-sided hypoplasia of the depressor anguli oris muscle accounted for the asymmetry of facial expression. Conclusion: This case illustrates the value of a systematic physiotherapy assessment in a child with VACTERL association presenting with facial palsy and demonstrates how an impairment-based ICF-structured rehabilitation programme can be designed to address facial, communicative, respiratory and musculoskeletal consequences of the condition.

Keywords: VACTERL association, facial palsy, microtia, polydactyly, anorectal malformation, Sunnybrook Facial Grading System

Introduction

VACTERL syndrome is a group of growth abnormalities (birth defects) that happen in the early stages of embryo development during pregnancy. VACTERL stands for vertebral defects, anal atresia, cardiac defects, tracheooesophageal fistula, renal anomalies and limb abnormalities. A diagnosis occurs if child has at least three parts of their body affected by symptoms. Each case of VACTER syndrome is unique. Affected individuals may have additional abnormalities that are not among the characteristic features of VACTERL association. Frequency - Occurs in 1 in 10,000 to 40,000 newborns. Causes - Different causes in different people. condition is likely caused by interaction of multiple genetic and environmental factors. It is unclear why the features characteristic of VACTERL association group together in affected individuals. Since the cause of VACTERL syndrome is unknown, there is no way to prevent the condition. Genetic testing and prenatal care can detect some symptoms of VACTERL syndrome before your baby is born to help doctor and parent prepare to treat your child on your due date. After your baby is born, your provider will make sure your baby is healthy enough for surgery to correct any symptoms that could be life-threatening. Treatment will be lifelong as symptoms could arise throughout childhood and into adulthood. There is no cure for VACTERL syndrome.



Demographic Data

Table 1: Demographic Data

Parameter	Details	Parameter	Details
Gender	Male	Height	110 cm
Age	9 years 9 months	Weight	22 kg
Date of birth	20/02/2016 (20th Feb 2016)	BMI	18.2 (15–22)
Date of evaluation	11th December 2025	Head circumference	50 cm
Chronological age	9 years 9 months 21 days	Blood group	O +ve

Chief Complaint: Difficulty in doing facial expressions and talking.

Associated Complaints: Difficulty in remembering colours, hearing and passing stools.

Pedigree Chart: Father - 36 years; Mother – 35 years. 3° consanguineous marriage, Children: Girl – 12 years; Boy (patient) – 9 years.

Case Presentation

The patient was apparently alright after birth. After 3 to 4 hours of delivery, the doctors noticed anal atresia & also found that the baby was having a congenital heart disease, but the doctors assured the parents that it would get resolved on its own in a few months. Also, the baby had right side ear deformity since birth and is having 12 fingers on hand (Preaxial/ Radial), i.e. 1 extra finger on each hand. After the delivery, since he was not able to defecate, an operation which was anoplasty (i.e. the surgery for anus reconstruction) was performed. He was kept in NICU for 10 days, after which he was brought home. After bringing him home, he was having a history of continuous cold and cough for almost 1½ years, for which he got treatment in a local Hospital intermittently. While the treatment was going on, due to medications the patient used to feel better for some days, but after that the patient used to feel sick again. After 2 years his mother understood that he was not able to speak/talk properly or play properly as the children of his age, so at that time the patient's mother took him to Kshirsagar Hospital at Shirdi. There the doctor told the patient's mother that after some more years, his operation for the ear will be performed, through which the hearing and talking problem will be resolved. So the patient did not take any further treatment for that. The patient was put in school and was learning normally. After he came in 4th standard at the age of 9 years, his mother noticed that his son could talk but not fluently and also his grasping power was less and difficulty in hearing. Also, due to winter he was troubled with constant cough and cold. Concerned about this, the patient's parents took him to PHC, where the doctor referred him to go to SJS Hospital Kokamthan. On the same day, i.e. on 24th November 2025, he was brought to SJS Hospital Kokamthan by his parents, where he was referred to Paediatric PT from the same day itself. All vaccination are done till now.

Birth History

Pre Natal: Age of mother at time of birth – 26 yrs, Consanguineous marriage – Yes. (3^0 Degree)

Peri Natal: Baby was delivered at 9 months of pregnancy, His birth weight was 2.5 kg, Normal vaginal delivery was performed, No H/O: breech presentation.

Post Natal: Baby was having difficulty in passing stools, for which surgery was performed, Congenital heart disease: PDA – 2 mm; ASD – small (left to right shunt), When his 2-D echo was done on 10/10/2022, the patient had resolved PDA and ASD, so he is not having any congenital heart defect presently.

Past Medical History

Tab. Augmentin (Antibiotic), Nasoclear (Nasal drops), Betadine gargles (Cough & cold), Syp. Duphalac (Constipation), Syp. Relux (Constipation), Syp. Sorbiline (asthma & occasional constipation), Glycerine suppositories (Constipation), Syp. Cyopiox (Bacterial infection)

Past Surgical History - Anoplasty was done.

Developmental History

Table 2: H/O Gross Developmental Milestones

Milestone	Age Achieved
Neck holding	5 months
Rolling (supine to prone)	2½ months
Crawling	–
Sitting with support	1 year
Sitting independently	1 year – 1½ years
Standing with support	2 years
Standing independently	3 years
Cruising	3½ years
Walking independently	3 years
Climbing steps with support	3½ years
Climbing steps independently (B/L)	4 years
Climbing steps independently (alternate)	4 years

Table 3: H/O Fine Motor Milestones

Milestone	Age Achieved
Midline B/L activities	5 months
Grasping rattle / cloth	9 months
Releasing rattle / object	9 months
Grasping cube	1 year
Grasping pellet	1½ years
Grasping pencil / pen	3 years
Unbuttoning button	7 years
Buttoning button	8 years

Table 4: H/O Social Development

Milestone	Age Achieved
Smiles at mirror / image	2 years
Waves bye bye	2 years
Play simple ball games	3 years
Knows gender	–

Table 5: H/O Speech & Language

Milestone	Age Achieved
Babbling, cooing, laughing	3 years
Monosyllable words (M, B)	7 years
Bisyllable words (Mama, Baba)	7 years
2 words with meaning	8 years
10 words with meaning	Speech is not fluent, cannot be assessed
Simple sentence	9 years
Telling story	Not achieved

H/O Oromotor Development

Deviation of mouth: Present, Drooling: Absent, Dentition: Present, Consistency of food: Can eat solid food.

H/O Bowel & Bladder Control

As the patient has undergone surgery of anoplasty (i.e. surgery for anus reconstruction), after which enema was done at hospital for 10 days and also was done by parents at home for 6–7 months, which helped in defecation. Even now the patient is not able to pass stools properly.

- Ability to inform before defecation: 8 years.
- Ability to inform after passing urine or faeces: 8 years.

On Observation

Standing Posture

Facial expressions - deviated on right side, 1 extra finger on each side, Microtia present, Kyphosis and scoliosis present in upper thoracic region.

On Examination

7th cranial nerve affected (motor), Tone, Higher Mental Function, Range of motion and Manual Muscle Testing (MMT) are normal. Serratus anterior and Infraspinatus graded 4 - winging of scapula present. Gross motor and fine motor development is typically atypical, proximo-distal.

Facial Muscle Assessment

As the patient has facial palsy, strength of facial muscles is assessed by Jacqueline Montgomery scale.

Table 6: Facial Muscle Assessment Jacqueline Montgomery scale

Muscle	Grade	Description
Extra ocular muscles	F	Immediate tracking in smooth motion over full range
Levator palpebrae superioris (LPS)	F	Completely normal range of movement, holds against therapist's light manual resistance
Orbicularis oculi	WF	No resistance to eye closure; closure may be incomplete but only small amount of sclera and no iris is visible
Corrugator supercilii	NF	Slight motion detected
Occipitofrontalis	NF	Slight motion detected
Nasalis	WF	Shallow crease, patient yields to any resistance
Orbicularis oris	WF	loss of lip closure, impaired speech articulation and drooping at the corner of the mouth
Buccinator	WF	Food gets stuck in the side of the mouth (vestibule) during meals. Inability to build up pressure in the mouth makes actions like whistling, sucking through a straw, or playing wind instruments difficult.

Key: F – Functional; WF – Weak functional (moderate impairment); NF – Non-functional (severe impairment); O – Absent.

Sunnybrook Facial Grading Scale

Resting Symmetry

Table 7: Sunnybrook Facial Grading Scale (Resting Symmetry)

Component – Compared to normal side	Score
Eye	1– Narrow
Cheek	1– Less Pronounced
Mouth	1– corner pulled up
Total	3 × 5 = 15

Table 7: Sunnybrook Facial Grading Scale (Symmetry of Voluntary Movement)

Standard Expression	Symmetry of Voluntary Movement – Compared to normal side Score	Synkinesis – Involuntary muscle contraction associated with each expression Score
Brow lift (FRO)	2– slight movement	2– Moderate: Obvious but not disfiguring synkinesis
Gentle eye closure (OCS)	4– movement almost complete	1– Mild: Slight synkinesis
Open mouth smile (SYG/RIS)	1– Unable to intiate movement	2– Moderate: Obvious but not disfiguring synkinesis
Snarl (LLA/LLS)	1– Unable to intiate movement	2– Moderate: Obvious but not disfiguring synkinesis
Lip pucker (OOS/OOI)	2– slight movement	2– Moderate: Obvious but not disfiguring synkinesis
Total	10 × 4 = 40	9

Composite score: 40 – 15 – 9 = 16

Balance - Paediatric Balance Scale – Total score: 56.

Gait - Normal.

Respiratory System Examination

Table 8: Respiratory System Examination

Parameter	Findings
Chest symmetry	Asymmetrical
Chest expansion – Axilla level	2 cm
Chest expansion – Nipple level	1 cm
Chest expansion – Xiphoid level	1.5 cm
Breathing pattern	Abdominothoracic
Thoracic configuration (AP: TV ratio)	18: 20 = 1: 1 (which indicates barrel shaped chest)

Differential Diagnosis

Table 9: Differential Diagnosis

Condition	Why it is considered (Overlap with VACTERL)	Why it is less likely / Distinguishing features
Alagille syndrome	Vertebral anomalies (butterfly vertebrae), congenital heart disease, renal anomalies may occur	Cholestatic liver disease, bile duct paucity, posterior embryotoxon, characteristic facies; TEF and anal atresia uncommon
CHARGE syndrome	Congenital heart defects, tracheoesophageal fistula (TEF), ear anomalies, growth delay	Coloboma, choanal atresia, cranial nerve dysfunction (including facial palsy), genital anomalies
Fanconi anaemia	Radial ray, vertebral, renal and cardiac anomalies cardiac defects may mimic VACTERL	Bone marrow failure, pancytopenia, café-au-lait pigmentation, short stature, positive chromosome breakage test
Holt–Oram syndrome	Limb malformations with congenital heart disease	Cardiac conduction defects; vertebral, anal, TEF and renal anomalies uncommon
Oculo-auriculo-vertebral (Goldenhar) syndrome	Vertebral anomalies, microtia, hemifacial microsomia, cardiac defects	Epibulbar dermoids, mandibular hypoplasia, facial asymmetry; anal atresia and TEF uncommon
Townes–Brocks syndrome	Anal atresia, ear anomalies, thumb/radial defects, renal anomalies	hearing loss, triphalangeal thumb; vertebral defects and TEF uncommon
Feingold syndrome	Esophageal/duodenal atresia, digital anomalies, microcephaly	Syndactyly, learning disability, vertebral and anal anomalies are less frequent
22q11.2 deletion syndrome	Cardiac, renal and vertebral anomalies	Palatal defects, immune deficiency, hypocalcaemia, characteristic facies
VACTERL association	≥3 major anomalies: Vertebral, Anal, Cardiac, TEF/EA, Renal, Limb. Facial palsy and microtia are rare associated findings.	Diagnosis of exclusion; absence of defining features of other syndromes.

Investigations

2-D Echo & Colour Doppler: 2-D echo was done on 10/10/2022, the patient had resolved PDA and ASD, so he is not having any congenital heart defect presently, Artificial Rupture of Membrane: Imperforate anus, operated on Day-1, single stage surgery.

X-Ray: Diaphragm – Elevated dome of diaphragm, right lobe slightly above than left, Cardiothoracic ratio – Normal 14:5 (1 to 1.5 up Diaphragm), B/L hands pre-axial polydactyly, Hemivertebra of cervicothoracic, 3rd and 4th ribs are fused – left side (scoliosis), No homogenous opacity noted.

Diagnosis

9 year old male, is a known case of VACTERL association associated with facial palsy.

Problem List

- Difficulty in facial expressions.
- Difficulty in talking fluently.
- Difficulty in hearing.
- Scapular muscle weakness.

Short Term Management

A. To improve facial expressions

1. Facial PNF

- For frontalis, orbicularis oculi, orbicularis oris, buccinator — give an initial stretch prior and opposite to muscle action.
- Give assistance to the muscle with less strength and resistance to the muscle with more strength.
- Initially ask the patient to do the movement voluntarily, how much ever he can; this will help in facilitation of muscles.
- Facial PNF: given for 3 repetitions × 3 sets.

2. Electrical Muscle Stimulation

- Since the muscles are partially innervated, hence to stimulate the muscles, EMS is given in interrupted galvanic mode and trained for 30 contractions at intensity where palpable / visible contractions are seen, at frequency of 100 Hz.

3. Rood's Facilitation Techniques

- Such as fast stroking, brushing, icing can be given in order to facilitate the muscle in order to make them functional again.

B. Reduce the difficulty in talking fluently

- Auditory and sensory stimulation testing.
- Teach the patient to recite a single word, then two words and then full sentence.
- Teach the patient to read and write simultaneously.
- Counsel the patient's parents for speech therapy and also refer them to a speech therapist.
- Ask the patient's parents to take keen interest in the learning of patient and to devote their time to the child.

C. Improve hearing

- Sound therapy and aural rehabilitation.
- Blindfolding the patient and making the patient recognise the sounds of animals, birds etc.
- Show the patient picture cards so that he can understand and remember both the voice and the animal / bird.

D. Improve the strength of scapular muscles and reduce the shortening of neck muscles

- Scapular PNF: can be given in posterosuperior to anteroinferior direction and posteroinferior to anterosuperior direction.
- Stretching the muscles with theraband.
- Wand exercises in all directions will help in increasing strength of scapular muscles and will also help in increasing the range of motion.
- Scapular setting exercises: 4 times a week.

Long Term Management

1. Maintaining the quality of speech and also progress till recitation of small poem, paragraph or a story.

- This can be done by dual tasking — listening and also talking.
- Can ask the mother to recite a paragraph and then ask the child to recite along with her / after her.
- Pay attention that the child speaks the correct words, and if not then train the child to recite the syllables correctly.
- Listening, seeing, recognising, reading, writing.

2. Maintaining the facial muscle training and adding resistance to the achieved strength in order to increase the strength.

- If the affected side has some strength then facilitate the facial muscles and then motivate the patient to do the movement, i.e. wrinkling, frowning, closing of eyes, smiling, puffing of cheeks against manual resistance.
- Continue EMS until there is no muscle contraction at a very low intensity and until the SD curve shows normal innervated muscle graph.

3. To maintain scapular muscle strength.

- Resistance exercises by using theraband.
- Scapular strengthening exercises 4 times a week using weights / weight cuffs.

Discussion

VACTERL association is a rare congenital condition characterized by the coexistence of vertebral, anorectal, cardiac, tracheoesophageal, renal, and limb anomalies, with diagnosis requiring at least three component defects after exclusion of overlapping syndromes. The present child demonstrated vertebral anomalies, imperforate anus requiring neonatal anoplasty, congenital cardiac defects and bilateral pre-axial polydactyly, fulfilling the diagnostic criteria for VACTERL association. Additional findings of right-sided microtia and congenital facial palsy represent uncommon craniofacial manifestations that have been reported only rarely in VACTERL association. [1, 4, 9] The child showed marked facial muscle weakness with a Sunnybrook Facial Grading Scale score of 16, along with speech difficulty, hearing impairment, postural abnormalities and mild respiratory involvement. These findings highlight the functional burden associated with multisystem congenital anomalies and emphasize the need for comprehensive rehabilitation beyond surgical correction. [10, 11] The proposed physiotherapy program was structured according to the International Classification of Functioning, Disability and Health (ICF) framework and included facial proprioceptive neuromuscular facilitation, electrical muscle stimulation, speech-oriented exercises and scapular strengthening. Standardized assessment tools such as the Sunnybrook Facial Grading Scale provide reliable documentation of facial nerve dysfunction and can be useful for monitoring rehabilitation outcomes. [14, 16, 20] This case demonstrates that children with VACTERL association may present with atypical craniofacial involvement and benefit from early multidisciplinary rehabilitation aimed at improving facial function, communication, posture and participation in daily activities. [10, 20]

Conclusion

This case report describes a rare presentation of VACTERL association accompanied by congenital facial palsy, right-sided microtia, bilateral pre-axial polydactyly, vertebral anomalies, repaired anorectal malformation and resolved congenital cardiac defects in a 9-year-old child. The case demonstrates that VACTERL association may present with atypical craniofacial manifestations that significantly affect facial expression, speech, hearing, posture and respiratory function. A comprehensive physiotherapy assessment using standardized outcome measures such as the Sunnybrook Facial Grading Scale and Pediatric Balance Scale facilitated the identification of impairments across multiple body systems. An individualized ICF-based rehabilitation program targeting facial muscle function, speech-related activities, scapular stability, posture and respiratory mechanics was proposed to address these deficits. Early multidisciplinary intervention and long-term rehabilitation are essential to maximize functional independence, communication ability and participation in children with complex congenital conditions such as VACTERL association.

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- Conflict of Interest: No conflict of interest
- Informed Consent: Informed consent was obtained from the patient.
- Ethical Approval: The study was approved by the Institutional Ethical Committee

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