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A Clinical Case Report on Goldenhar Syndrome Withoesophageal Stricture.

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ABSTRACT:

Multiple congenital conditions known as Goldenhar syndrome include recognizable features in the face, ears, and eyes.Early finding then effective treatment can lead to positive results.A 8-year-old male child brought by parents with case of 1 episode of GTCS type convulsion. convulsion was aborted after 5 mins without any medications. After that shifted child to hospital Child was given inj levera, inj mannitol and on admission patient was kept NBM. Physical examination was performed and external auditory canal is not visualized bilaterally. Bilateral pinna appears tiny and deformed mandibular condyle appears smaller in the left as compared to the right. Endoscopy was done proximal oesophageal stricture are presented and paediatrician diagnosed Goldenhar syndrome with oesophageal stricture. started on oxygen by Nasal prongs, IV fluids, Inj Ceftriaxone, Inj Pan top, inj Emset, Inj Levera, Inj 3% NaCl. Stricture noted at 22 cm from the incisor teeth and advice for Barium swallow. Barium swallow was done stricture from 22 cm from incisor teeth up to entry of stomach. Paediatric and Anaesthesia fitness was given and oesophageal dilatation was done in coordination with paediatric surgeon. Graded dilatation was performed with Savary Gillard dilators from 5 mm up to 11mm and was advised for repeat dilatation after 4 weeks. Pt was vitally stable during and post procedure and was started on liquid diet for the night and soft diet from next morning. Patient had mild pain while swallowing hence Sucran- O syrup was added, post which complaints of pain reduced. Repeat EEG was done s/o normal. Patient is currently vitally and hemodynamically stable and hence is being discharged with advice to follow up after 4 weeks.

Keywords: Goldenhar syndrome, Genetic, Endoscopy, Dilation, oesophageal stricture, oesophageal anastomosis.

INTRODUCTION

A rare congenital disorder called Goldenhar syndrome is characterized by faulty eye, ear, and spine development.(*Goldenhar Syndrome: An Atypical Presentation With Developmental and Speech Delay - PMC*, n.d.). Goldenhar syndrome, also called OAV or oculo-auriculo-vertebral spectrum, was initially identified by ophthalmologist and general practitioner Maurice Goldenhar in 1952.Only one in every 3,500 to 25,000 newborns has Goldenhar at birth.Additionally, various internal organs and facial structure may be impacted by this illness.(*OculoAuriculo Vertebral Dysplasia; Goldenhar Syndrome - IJOOO*, n.d.)Each person's anomalies and symptoms are different in severity.This could indicate ear tags, small ears, missing ears, small eyes, missing eyelids, growths on the ears, or loss of hearing. In the spine, it can result in fused or missing vertebrae, as well as inadequate vertebral development.The syndrome frequently results in scoliosis, or a crooked spine, in its victims.There are additional, less prevalent, and less obvious signs.(*Goldenhar Syndrome: Symptoms, Causes, and Treatment*, n.d.)

The percentage of Goldenhar patients who have intellectual disabilities ranges from 5 to 15%.Additionally, some have anomalies in their internal organs, most frequently congenital heart problems. Kidney and limb defects are quite rare. Microtia, a hearing loss that often affects one ear and can result in partial or complete deafness on the affected side, abnormalities in the cardiovascular system, lungs, kidneys, and genitalia, and the central nervous system.(*Goldenhar Syndrome: A Case Report with Review - PMC*, n.d.)The aberrant narrowing of the oesophageal lumen is known as an oesophageal stricture, and it frequently manifests as dysphagia, which is frequently reported by patients as trouble swallowing.An aberrant constriction of the

oesophageal lumen is referred to as an oesophageal stricture, and it frequently manifests as dysphagia, which is frequently reported by patients as trouble swallowing.(Desai & Moustarah, 2023). When one considers all the various potential etiology for oesophageal strictures, it is probable that they can affect any age range or community.Patients often appear with one or more of the following symptoms, regardless of the stricture's type: dysphagia, food impaction, odynophagia, chest pain, and weight loss.The most important symptom is dysphagia that progresses to solid food and occasionally extends to semisolid and liquid foodsThe majority of patients undergo endoscopic evaluation since it can offer comprehensive knowledge of oesophageal architecture, establish not only the diagnosis of a stricture but also enable mucosa biopsy.(*Oesophageal Strictures: Symptoms, Causes & Treatment*, n.d.)

When necessary, endoscopy offers the chance to therapeutically enlarge the stricture. Only patients with complicated strictures or when endoscopy is insufficient due to significant lumen narrowing should undergo contrast fluoroscopy.Endoscopic dilators are frequently used to treat benign strictures, followed by disease-specific therapy strategies.(Everett, 2019).

CASE PRESENTATION:

A 8-year-old male kid brought by parents with One episode of GTCS type convulsion. As narrated by mother child was apparently alright but suddenly developed convulsion which is GTCS types involving all 4 limbs with up rolling of eyes with no deviation of mouth. Convulsion was aborted after 5 mins without any medications. Child was taken to treatment metro superspecialist hospital. Child received the inj levera inj mannitol and referred to tertiary care of hospital for further management. On admission patient was kept NBM, started on oxygen by nasal prongs, iv fluids, inj ceftriaxone, inj pan top, inj Emset, inj levera, inj 3% NACL. Investigations done CBC,Hb 11, TLC 6300, PLT 3.5 HCT 31.8. Left, KFT done and was normal. Physical examination performed and result was swelling below eyes Ophthalmic call done and no evidence of disc oedema.CT face was done and no obvious abnormality in brain parenchyma. External auditory canal is not visualized bilaterally. Bilateral pinna appears tiny and deformed mandibular condyle appears smaller in the left as compared to the right. Asymmetrical pneumatization of mastoid air cells. The left side appears more pneumatized than the right. Patient had one episode of fever spike which subsided on medications. Lumbar puncture was done. No evidence of meningitis. Usg local site done parotitis with submandibular gland adenitis. MRI brain done no abnormality. Child was treated symptomatically.Patient was maintaining saturation with nasal prongs. Was accepting feeds hence Iv fluids tapered to 70%. 2d Echo done normal echo. Patient was shifted from injectable levera to oral levera. Patient was maintaining saturation and was stable and has been shifted to ward. ENT call was done I/V/O pre auricular swelling and was advised to get gastro call done i/v/o odynophagia. Gastro call was done and planned for gastroscopy. Patient was stable with no fever. Endoscopy was done proximal oesophageal stricture present. paediatrician diagnosed Goldenhar syndrome with oesophageal stricture**.** Noted at 22 cm from the incisor teeth and advice for barium swallow. Barium swallow was done s/o stricture from 22 cm from incisor teeth up to entry of stomach. Review gastro call done and advised for oesophageal stricture dilatation after fitness. USG Abdo pelvis was done s/o loaded bowel loops with gaseous distension. Paediatric and anaesthesia fitness was given and oesophageal dilatation was done in coordination with paediatric surgeon. Graded dilatation was performed with savary Gillard dilators from 5 mm up to 11mm and was advised for repeat dilatation after 4 weeks. Pt was vitally stable during and post procedure and was started on liquid diet for the night and soft diet from next morning. Patient had mild pain while swallowing hence sucran- o syrup was added, post which complaints of pain reduced. Repeat EEG was done and normal. Patient is currently vitally and hemodynamically stable and hence is being discharged with advice to follow up after 4 weeks.

DISCUSSION

Oculo-auricular-vertebral syndrome (OAV), also referred to as Goldenhar syndrome, is a congenital condition marked by Facial asymmetry, ear malformations such as preauricular fistula (microtia), or abnormalities of the eyes (bulbar dermoid cyst, microphthalmia), as well as vertebral anomalies.Maurice Goldenhar originally wrote about this case in 1952.(*Maurice Goldenhar Originally Wrote about This Case in 1952 - Google Search*, n.d.)

Its genesis is currently unknown. Goldenhar syndrome is a congenital condition, which means that infants are born with it. A chromosomal abnormality is the root of the problem.However, the syndrome is inherited as a hereditary issue in just 1% to 2% of instances. It is either autosomal dominant or autosomal recessive in these situations. This phrase indicates that the gene or genes responsible for it are located on a chromosome unrelated to Gender. Genetic factor can be recessive or dominant, but dominant is more prevalent.Nobody has yet identified the precise genes that are to blame. Although GS is typically sporadic, there have been cases of autosomal dominant or, less frequently, autosomal recessive genetic transmission in the literature.A multifactorial etiology has been proposed, affecting the embryological development of the first and second branchial arches and vertebral elements during the first three to five weeks of development. This etiology includes chromosome abnormalities, single gene mutations, vascular disruption, teratogens (thalidomide, retinoic acid, vasoactive drugs, etc.), maternal infection with rubella and influenza, maternal diabetes, maternal smoking, and multifetal pregnancy.There are numerous reported disorders involving different organ systems that are connected to GS. The name Expanded Goldenhar Complex should be used when there are related uncommon additional facial deformities because GS is the most severe variant of OAV spectrum.The diagnosis of these syndromes is made through a thorough clinical examination supported by the skeletal survey, USG abdomen, and echocardiography, and depending on the systems implicated, may also require other modalities such as CT and magnetic resonance imaging.The imaging findings will probably be used to confirm the diagnosis because many of these diseases are caused by sporadic rather than hereditary factors.(Hernandez et al., 2006)

When pulmonary, cardiac, or genitourinary systems are involved, along with radial deficits, vertebral segmentation anomalies, and other close differentials, the idea of an expanded Goldenhar complex should be raised.In order to regulate the pathology's etiology and stop further stricture formation as well as its advancement, the right therapy approaches must be used.Long-term PPI medication is the most effective way to treat peptic strictures. The clinical response to the PPI trial implies GERD linked eosinophilia rather than eosinophilic esophagitis, hence eosinophilic esophagitis should go through the trial.It is advisable to use vaporized glucocorticoids like fluticasone or budesonide to treat genuine eosinophilic esophagitis if a repeat endoscopy still reveals eosinophilia.In order to prevent pill-induced oesophageal stricture, certain precautions must be taken when taking pills. Patients need to be reminded to drink lots of water while taking their medications, take one pill at a time, and to avoid lying down for 30 minutes after taking them. Such actions aid in preventing additional mucosal deterioration and stricture recurrence.(*Water, Hydration and Health - PMC*, n.d.)

CONCLUSION

Patients with Goldenhar syndrome should undergo a full multidisciplinary examination because they may have several congenital defects. A finding must be made in conjunction with other multidisciplinary experts thus that proper therapeutic planning and ongoing assessments of the child's growth and development can be made.The abnormalities and severity found determine the prognosis, and a periodic review every six months is required to gauge the condition's development. The baby's prognosis in this example is excellent; there are no significant facial defects or presence abnormalities, such as heart or kidney abnormalities. The eyes, ears, and heart of the patients described here have a variety of issues. Patients are currently receiving medical attention and being monitored for additional potential issues.

CONFLICT: - No any conflict.

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