Available online on 15.06.2024 at ijmspr.com

International Journal of Medical Sciences and Pharma Research

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Open Access **Case Report**

A case of inherited metabolic disorder with airway difficulties in intensive care- challenges faced

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Article Info:

Article History:

Received 02 April 2024
Reviewed 04 May 2024
Accepted 26 May 2024
Published 15 June 2024

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Abstract

Uncommon disease like MPS imposes great challenges to intensivists in case of emergency due to their disease nature which involves GAG accumulation leading to cellular dysfunction and various clinical abnormalities. Airway management in Pediatric patients is extremely demanding even with expert hands. So, this case report discusses mainly the disease progress, and anticipated anesthetic challenges with its skillful handling strategy that has to be contemplated in case of emergency situation.

Keywords: intensive care, cellular dysfunction, clinical abnormalities

Cite this article as:

Krishna Prasad T, Soundarya P, Sheik Abdullah K, Varun SK, A case of inherited metabolic disorder with airway difficulties in intensive care- challenges faced, International Journal of Medical Sciences & Pharma Research, 2024; 10(2):1-4 *DOI: http://dx.doi.org/10.22270/ijmspr.v10i2.94*

Introduction:

A genetic mutation affecting lysosome hydrolase results in a set of disorders known as mucopolysaccharidosis (MPS), in which mucopolysaccharides cannot be digested normally and buildin the body, leading to a variety of symptoms. [1] Rough facial characteristic, corneal opacity, hepatosplenomegaly, anomalies of the heart valves, skeletal deformities, lumbar kyphosis or gibbous, and elevated mucopolysaccharide levels in the urine are some of the prominent clinical symptoms. The most accurate method for identifying MPS is enzyme activity measurement, and identifying IDUA mutation sites is a crucial step in MPSI diagnosis [2,3]

Glycosaminoglycan accumulation in the upper airway results in hypertrophy of adenoids, tonsils, tongue, and laryngopharynx, which may all pose difficulty for anesthetic airway management.

Because altered anatomy of the airway and facial structures can complicate airway management [2-5,] During the mask ventilation and endotracheal intubation. So tracheostomy is preferred in place of a definite airway.

In 56–63% of cases, the respiratory system has been demonstrated to be involved in the pathogenic process of MPS. In MPS, morphological changes such as shortness and joint instability of the neck, high epiglottis, hypoplastic mandible, hypersalivation, mucoid hypersecretion, gingival hyperplasia, hypertrophy of the adenoids and tonsils, narrowed nasal airway, flattened nasal bridge, macroglossia, hepato-splenomegaly, tracheobronchomalacia, as well as skeletal deformations such as kyphoscoliosis, ribcage narrowing, temporomandibular joint ankylosis and chest wall deformity, decreased thoracic dimensions due to short stature, diaphragmatic weakness from spinal cord compression as a result of narrowed craniovertebral junction.

Airway blockage is primarily caused by active GAG deposition in upper airway tissues, which causes tongue distension and adenoids/tonsils to enlarge. Increased mucoid secretion makes the problem worse. Due to GAG buildup in the tracheobronchial cartilage, lower respiratory airway collapse typically manifests as laryngomalacia and tracheobronchomalacia. Clinical signs of respiratory impairment include frequent respiratory tract infections (bronchitis, pneumonia, sinusitis, rhinitis, otitis, and bronchitis) and obstruction of the upper airways. Pathologies of the central nervous system, such as hydrocephalus and neuronal degeneration, may also cause upper airway dysfunction. Moreover, active GAG storage causes the craniovertebral junction to narrow, which compresses the spinal cord. [4 5.] So this case presentation emphasizes on meticulous management of such patients with multiple challenges.

Case report:

A 13 years old female patient previously diagnosed with MPS-1 (Hurler syndrome) at the age of four, was admitted to our hospital with complaints of shortness of breath for half a day along with fever and cough with expectoration for 3 days. The patient is a second-born child to a healthy second-degree consanguineous married couple with a family history of MPS -1 in her elder brother. The child was a full-term baby with normal developmental milestones till the age of 3. Then she had an episode of fever of unknown cause followed by which there was a progressive central nervous system degenerative manifestation like developmental regression including loss of toilet training, language, motor skills, intellectual disability, and sleep disturbances. Which slowly worsened over years with several episodes of seizures along with musculoskeletal manifestations like joint stiffness, contracture, scoliosis, and recurrent events of respiratory tract infections on and off. Karyotyping was done which was within normal limits. Enzyme analysis was done which showed reduced activity of α- L iduronidase which is highly suggestive of Hurler's Syndrome (MPS - 1disease)

Upon admission, a physical examination revealed that the patient was tachypneic, irritable, can't stand or sit, has a poor oral response, highly uncooperative. Her general findings showed short stature, frontal bossing, coarse facies, low set ears, depressed nasal bridge, thick lips, macroglossia and gingival hyperplasia, short neck, joint contracture, and respiratory symptoms including mouth and concave breathing with coastal margin inversion. Abdominal examination revealed an umbilical hernia. palpation of the abdomen shows hepato-splenomegaly. Chest x-ray revelated bilateral broncho vascular markings increased, abnormal morphology in vertebral height and central depression, and a few suspicious opacities in the right perihilar region suggestive of pneumonitis. (figure 1) CT brain showed cerebral atrophy along with multiple B/L chronic lacunae infarcts of the ganglion-capsular region and corona radiata. B/L small vessel ischemia. J-shaped sella, mild thickening of the inner and outer table with widening of diploic space.

In the emergency ward, the patient developed more tachycardia and desaturation, and in view of the anticipated difficult airway in view of MPS and reduced Atlanta- occipital joint mobility, we tried normal intubation without paralyzing the patient with injection. Midazolam 1mg, injection. glycopyrrolate 0.2mg, inj. propofol 20mg iv stat with fiber optic bronchoscope back up and later paralyzed the patient as successful intubation to reduce the work of breathing and impending respiratory failure. The patient was shifted to the critical care unit and was ventilated in pressure control mode with other supportive management. The patient had a course of 7 days in ventilation and was extubated on the 8th day and supported with intermittent CPAP. (figure 2) Regular ABG and serial x-rays was taken and treated accordingly. The patient was started on Ryle's tube feeding on 3rd day and passed stool on 5th day after intubation. Passive physiotherapy and angioedema measures were given. The patient was shifted to the ward on the 9th day and was monitored. The patient doing well without any further respiratory problems.

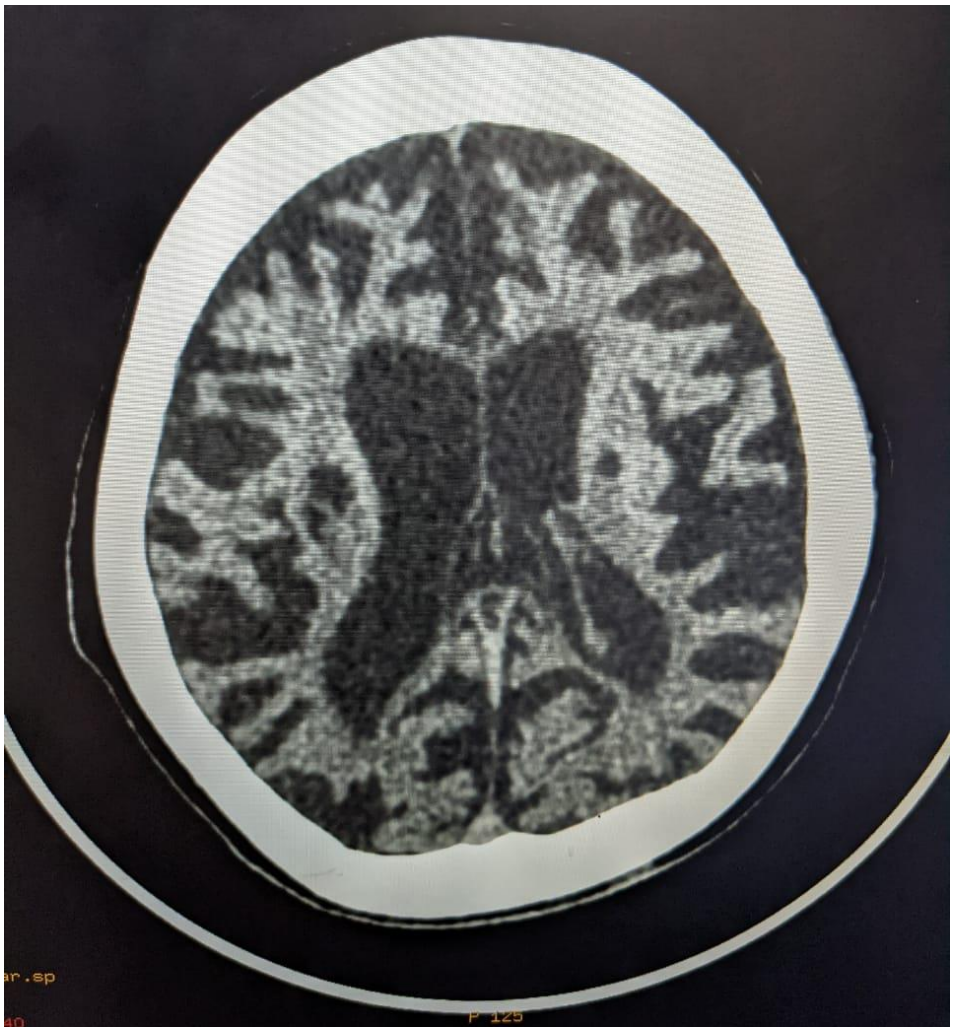

Figure 1: Computed Tomography picture.

Figure 2: Patient in the CCU recovered from ventilatory support

Discussion:

In mucopolysaccharidosis type I, lysosomal hydrolase defects result in an accumulation of glycosaminoglycan in lysosomes, which damages many organs and causes facial dysmorphism, bone malformations, hepatosplenomegaly, and intellectual impairments. The current recommended course of treatment for kids with MPSI is -L-iduronidase replacement therapy.

Several clinical signs of MPS, including as range of motion, visual acuity, hearing, cardiopulmonary function, face distinctive roughness, upper airway obstruction, poor respiratory function, and hepatosplenomegaly, can be improved with enzyme replacement treatment and HSCT. Enzyme replacement therapy, on the other hand, can enhance the body's readiness for transplantation and increase the likelihood that the transplant will be successful. Enzyme replacement therapy is typically given to MPSI children before transplantation in order to improve their physical state before to transplant and lower their risk of morbidity and mortality.

A group of lysosomal storage diseases known as MPS includes active GAG accumulation in all tissues, particularly in connective tissue structures. All kinds of MPS include respiratory difficulties,which frequently result in a child's death from this condition. The typical morpho-phenotypes of hypertrophy of the adenoids and tonsils, laryngomalacia, tracheobronchomalacia, obstructive and restrictive lung disease, small chest volume, skeletal deformation, and hepatomegaly are just a few of the factors that are linked to pulmonary systemimpairment in MPS patients.

It should be emphasized that these factors make respiratory

diseases worsen with age. The causes ofrespiratory limitation are diverse and include chest wall constriction, upper and lower airway blockage, and cervical myelopathy. Because of the common skeletal dysplasia and short height, changes in growth and development offer an additional route for respiratory impairment.

Restrictive lung disease affected the majority of the patients. Spirometry and CT scans of the respiratory system should be used to monitor children and adolescents with MPS since pulmonarydisease poses a high risk of mortality in individuals with MPS and this pathology worsens with age.

Short stature combined with enlarged liver and/or spleen can result in displacement of the diaphragm into the thoracic cavity, further impairing respiratory function. In addition to the defect of the chest wall already mentioned, MPS IVA patients frequently experience atlantoaxial instability and spinal cord compression, which can cause respiratory muscle weakness (Tomatsu et al. 2011). When there is decreased chest wall compliance, spinal cord compression can cause phrenic nerve dysfunction and inspiratory muscle weakness, which may make it more difficult for the patient to maintain ventilation.

Moreover, thoracic and/or lumbar involvement affects the strength of the respiratory muscle affects coughing and secretion clearance, making patients more susceptible to infections.

Even though tracheostomy is a treatment option for refractory progressive upper airway obstructionor for emergency airway management. Nevertheless, in these individuals, the distorted and loose trachea can make it challenging to implant the tracheostomy tube. Moreover, stomal narrowing, granulation formation, intrastromal tracheal stenosis, wound infection, and tracheomalacia may be linked to tracheostomy[6]

Patients become more and more reliant on carers as daily tasks become more and more challenging due to declining vision, hearing, oral health, respiratory and cardiac function, muscular strength, and endurance. On the internet, there is further information that describes the difficulties with enduranceand mobility that MPS IVA sufferers encounter (S1). In the end, as the disease worsens without therapy, the patient's quality of life gradually declines.[7]

Mask ventilation and Intubation are the most common challenges faced by patients with MPS which is usually accompanied by Cardiopulmonary Dysfunction. Intensivists also have additional challenges as a result of spinal involvement [7]. Every elective surgery necessitates the availability of a variety of airway management tools and a preoperative assessment of such risk factors and patient conditions. A team with experience using cutting-edge airway equipment and understanding MPS diseases should administer intubation and ventilation. Therefore, taking into consideration all these challenges, it is necessary for the intensivist to be competent enough to handle such disease processesalong with adequate knowledge of Newer Cutting Edge Airway instruments handling. This documentation of the case report would be beneficial to the clinical impact of early therapy in view of intensivists.

Conclusion:

To mitigate the risks of airway mismanagement in patients with MPS, anesthetic planning should include experienced anesthesiologists and expertise in using a full array of advanced airway devices.

Declaration of patient consent: All the appropriate consents for sharing his/her clinical informationand photographs have been obtained from the patient's parents. The parents understand that their names and initials will not be published and all attempts to conceal patient identity will be made.

Financial support and sponsorship: Nil.

Conflicts of interest: There are no conflicts of interest.

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