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An Interesting Diagnosis for a Case of Repeated Aspiration Pneumonia

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Description

A 2-year 6-month-old little girl, presented with recurrent fever with episodes of cough and non-bilious vomiting for last 6 months. She had poor eating habit and had failure to thrive. Examination revealed dysmorphic facies like prominent forehead, deep sunken eyes, malar prominence, hypertelorism, small nose, long filtrum, small contracted mouth, pursed lips (Figure 1). Skeletal features showed contracture of multiple joints especially small joints of hand and foot with prominent muscle, camptodactyly with ulnar deviation of hand, uncorrected club foot and short trunk and limbs (Figure 2).



Figure 1 Typical facial features "whistling face".

She was febrile, coughing relentlessly and chest auscultation revealed bilateral crepitations. Chest X ray revealed features like aspiration pneumonia with predominant involvement of right upper lobe. For better delineation of pulmonary pathology, a HRCT (High Resolution Computerized Tomography) of the chest was done that revealed dilated thoracic esophagus with bowel wall thickening at GE (Gastro-esophageal) junction and patchy consolidation of both upper and lower lobes likely aspiration pneumonia. So, she underwent a GI (Gastro intestinal) Contrast study that showed a massive hiatus hernia with hour glass like stomach due to peristaltic activity. The dilated esophagus on CT chest is now correctly identified to be a part of this large hiatus hernia,

responsible for frequent aspiration (Figure 3). All her physical features and hiatus hernia led to a diagnosis of Freeman Sheldon Syndrome which is Arthrogryphosis Type 2a. A mutation analysis was planned but could not be done.



Figure 2 Physical findings of Freeman Sheldon syndrome.



Figure 3 Contrast study showing large hiatus hernia.

Upper GI endoscopy was not done. She went for a corrective surgery but succumbed after this high-risk procedure due to post-operative complications.

Freeman Sheldon syndrome (OMIM 193700) is a type of cranio-carpotarsal dystrophy associated with limb and spine deformities, multiple joint contracture, typical craniofacial characteristics, and gastroenterological problem [1]. They may

have upper airway obstruction [2] and extra-pulmonary restrictions causing chronic respiratory failure but present case had recurrent pneumonia. These patients usually have swallowing difficulty, dysphagia due to contraction of muscles around and inside oral cavity and muscles of deglutition. They can present with various types of hernia like inguinal hernia, umbilical hernia and hiatal hernias. They have increased risk of gastroesophageal reflux with or without hiatus hernia like above patient [3] and that was the cause of her repeated febrile illnesses.

References

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