Jarcho–Levin syndrome with association of unilateral pulmonary hypoplasia and diastematomyelia: A case illustration

Sir,

Jarcho–Levin syndrome (JLS) is a rare distinctive form of short-trunk dwarfism with rib and vertebral anomalies. This is the first case of a newborn exhibiting an unusual combination of spondylothoracic and spndylocostal JLS with lung anomaly and diastematomyelia to be reported. A new born (1-day-old) male baby, born by Lower segment Caesarean section (indication was nonprogression of labor), birth weight 2.35 kg, first child of nonconsanguinous marriage, was referred to our tertiary care hospital with multiple anomalies and recurrent episodes of cough, cold, vomiting after feeds, and rapid breathing. On examination, the baby was afebrile, with tachycardia, and
tachypnea (32 per min). Head circumference was 35 cm, short stature (length is 37 cm), short neck, anteverted nares and dysmorphic features (low-set ears, hypertelorism, high-arched palate, short upper segment). Distal penile hypoplasia with chordee and penile torsion, bifid scrotum, hypertrophy of left hemiscrotum were also present. On further examination, abdomen was distended with visible bowel loops. Imperforate anus and anal pit was noted. Spinal examination showed scoliosis in thoracolumbar region with curvature to right along with sacral agenesis, left calcaneovalgus, and over-riding of feet. Chest radiography showed fusion and fanning of ribs [Figure 1a and b]. Milks scan [Figure 1c] was performed due to vomiting after feeds and images revealed no evidence of gastroesophageal reflux. Computed tomography of thorax showed hypoplastic left lung. MRI spine [Figure 2] showed severe kyphoscoliotic deformity of dorsolumbar spine, agenesis of sacral segments, multiple vertebral segmentation and fusion anomalies in the dorsolumbar region. Bifid ribs were noted. Cervical cord was normal. There was a low-lying cord, split at D5 level by fibrous bar, which is known as diastematomyelia. 99m Tc DTPA (diethylene triamine penta acetic acid) renogram was performed to look for any associated renal anomalies and revealed bilateral normally functioning kidneys [Figure 1d]. Based on the constellation of symptoms and imaging findings, JLS was suspected and was advised DNA isolation.

JLS is a rare distinctive form of short-trunk dwarfism with rib and vertebral anomalies. This is the first case of JLS revealing a rare combination of both subtypes of JLS with lung anomaly and diastematomyelia the other case published from India showed diastematomyelia and spondylodistal dysostosis. Hernias, neural tube defects and anomalies of the anal opening, urinary tract, external genitalia, uterus and lower limbs may be associated. Recently, airway abnormalities have been described in two patients with the JLS similar to our case. Malformations of the spinal cord with a separation into two hemicords have been termed “diastematomyelia,” which has been rarely reported in the literature, which is present in our patient. JLS has been
Cytokines play a complex role in the pathogenesis of asthma and COPD. According to textbook teaching, COPD patients show neutrophilic inflammation while asthmatics show eosinophilic inflammation in their airway. However, a growing body of evidence reports the existence of overlap between asthma and COPD. GINA guidelines have recently defined Asthma-COPD Overlap Syndrome (ACOS) as a distinct entity.

Neutrophilic inflammation does not respond to corticosteroids, while eosinophilic inflammation is corticosteroid responsive. Sputum eosinophilia has been reported in 20-40% of sputum samples from stable COPD patients as well.

Airway inflammation in asthmatics is mediated by Th2 cells, which express signature interleukins (IL) for allergic (eosinophilic) inflammation, such as IL-4, 5, and 13.

Bafadhel et al. reported that increased sputum IL-5 levels were associated with sputum eosinophilia in COPD patients, which was attenuated by oral corticosteroid therapy.

Michael et al. reported three biological clusters based on sputum cytokine profile of 86 asthmatics and 75 COPD patients. Cluster 1 consisted of mainly asthmatics and had elevated sputum Th2 cytokines and eosinophils. Cluster 2 comprised both asthma and COPD patients with sputum neutrophil predominance. Cluster 3 had mainly asthmatics.

Sir,

We read with great interest your article “Value of past clinical history in differentiating bronchial asthma from chronic obstructive pulmonary disorder in male smokers presenting with shortness of breath (SOB) and fixed airway obstruction.” Authors have highlighted the importance of past history in differentiating asthma from chronic obstructive pulmonary disorder (COPD). But, recent insight into current evidence emphasizes the need for correct classification of asthma and COPD patients by recognizing the subgroup with overlap syndrome.

Traditionally asthma is considered a childhood/early adulthood onset disease with reversible airflow limitation while COPD is considered to manifest after 40 years of age and has irreversible or partially reversible airflow limitation. The pathogenesis of COPD has shown that it has much earlier origin than previously thought. In utero/childhood smoke exposure and decreased lung growth in early adulthood are linked to increased risk of COPD at a later age.

At one end, COPD patients may have partially reversible airflow limitation and at the other end, asthmatics may show partially irreversible obstruction due to airway remodeling by long-term disease process.

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