A Rare Association of Congenital Cystic Adenomatoid Malformation with Down syndrome

Rohit Khandelwal¹, Leeni Mehta Khandelwal², Karnail Singh³.
1. Resident, Department of Paediatrics, Govt. Medical College, Amritsar, Punjab. Currently working as Assistant Professor, Department of Pediatrics, Vydehi Institute of Medical Sciences and Research Centre, Bangalore.
2. Department of Internal Medicine, Vydehi Institute of Medical Sciences and Research Centre, Bangalore, Karnataka.
3. Professor and Head, Department of Paediatrics, Govt. Medical College, Amritsar, Punjab.
Email: docrl09@gmail.com


Keywords: Congenital cystic adenomatoid malformation, Down syndrome, CECT

Abstract:-
Congenital cystic adenomatoid malformation (CCAM) is an unusual condition characterized by immature malformed lung tissue with cystic appearance, which results from an abnormality of branching morphogenesis of the lungs. An adenomatous overgrowth of terminal bronchioles and alveoli leads to large masses which are communicating with tracheobronchial tree and have feeding vascularisation from pulmonary(bronchial) circulation. We hereby report a 2 year old male child with CCAM with features of down's syndrome. Congenital cystic adenomatoid malformation is a rare pulmonary anomaly. It consists of hamartomatous or dysplastic lung tissue mixed with normal lung. The lesion probably results from an embryologic insult, before the 35th day of gestation, with development of terminal bronchiolar structure¹. It was first acknowledged as a separate entity and introduced into English literature by Ch'in and Tang in 1949².

Case Report
A 2 year old male child, second in birth order, born of non consanguinous marriage, was admitted with a history of dry cough, intermittent fever (100-101.F) for 4 days, respiratory distress, lethargy and decreased intake for 1 day. The child had features
suggestive of Down’s syndrome with developmental delay.

On examination, right hemithorax was dull on percussion with decreased air entry and bronchial breathing in right mammary and inframammary area. Chest x-ray (CXR) showed right sided heterogenous opacity.

Hemogram revealed anemia with a Hb of 6.2 g/dl, leukopenia with a TLC of $2.5 \times 10^3/\mu L$ with (neutrophils 30% of the total, lymphocytes 67% of the total) and thrombocytopenia with platelets of 1 lac/mm$^3$. Peripheral Blood film showed that RBCs were both normocytic and microcytic with mild hypochromia with anisopoikilocytosis. Platelets were just adequate. He was treated with 3 weeks of parenteral antibiotics but continued to have cough with respiratory distress. CXR showed right sided heterogenous opacity with multiple cystic shadows with rightward shift of mediastinum with collapse of right upper lobe. Contrast enhanced computed tomography (CECT) showed multiple cystic lesions in right lower lobe with air fluid levels in them with collapse/consolidation of left lobe with mediastinal lymphadenopathy. Patient was diagnosed as Type I CCAM. The child remained well for 1 week and was discharged.

After 2 months, the child again developed fever for 15 days with cough and respiratory distress for 2 days. On examination, breath sounds were diminished on right hemithorax and there were crepitations. Hemogram showed anemia with a Hb of 5.5 g/dl, neutrophilic leucocytosis with TLC of $11 \times 10^3/\mu L$ with (neutrophils 79% of the total). CXR revealed right sided heterogenous opacity with collapse/consolidation of right upper lobe with multiple cystic shadows. After treatment with antibiotics, child got well and was discharged.

Discussion
CCAM is rare and usually presents before the age of 3 years. It is more common in boys than girls and is usually unilateral. Patients can present with respiratory distress, recurrent respiratory infections and pneumothorax. Some cases are asymptomatic until mid-childhood and are discovered as an incidental finding on radiography. Differential diagnoses include congenital lobar emphysema, pulmonary sequestration, congenital diaphragmatic hernia, bronchogenic cyst and cystic bronchiectasis. CCAM association with Down’s syndrome, which is seen in our case, has not been reported yet.

CCAM is best diagnosed with CT scan and classified as:

**Type I** (50%) : This is the most common type and composed of variable cysts with atleast 1 dominant cyst >2 cm lined with ciliated pseudostatified epithelium. Prognosis is excellent.

**Type II** (40%) : Composed of smaller uniform cysts upto 2 cm with similar histology. This form is commonly associated with anomalies (especially renal, cardiac, intestinal and skeleton).

**Type III** : Least common type and is composed of microcyst appearing solid upon visual inspection. Poor prognosis is secondary to respiratory compromise and associated congenital abnormalities. In 1977, Stocker et.al originally described the findings and classified CCAM$^3$. 

122
Some authors have observed predilection of the right lung over the left for this anomaly\(^4\) as observed in our case. Clinically, breath sounds may be diminished with mediastinal shift away from the lesion. CXR reveals a cystic mass, sometimes with mediastinal shift. Occasionally, an air fluid level suggests a lung abscess.

Antenatal intervention in affected infants is controversial but may include excision of the affected lobe for microcystic lesions, aspiration of macrocystic lesions and open fetal surgery. The definitive treatment of CCAM is surgery. In the postnatal period, surgery is indicated for all symptomatic patients, history of recurrent infection and risk of malignancies which have been rarely reported\(^5\).

The outcome of lobectomy/pneumonectomy is good in children. In lobectomy, the removing lung grows and expands well enough so that total lung volume and pulmonary function tests return to normal\(^6\). The response is most vigorous in the very young because new acini and alveoli form up to 5 years of age. Post resection majority of the patient have an excellent result. Younger patients have lower ratio of residual volume to TLC and higher maximum breathing capacity. This suggests that hyperplasia rather than overdistension occurs in the remaining lung\(^7\). The factors indicating poor prognosis include bilateral lung involvement, associated hydrops and presence of other congenital anomalies\(^8\). In our case, involvement was unilateral with features of down's syndrome.

References