

## CASE REPORT

**Congenital Cystic Adenomatoid Malformation of Lung-Rare Case Report***N. S. Kamakeri**Department of Pathology, Karnataka Institute of Medical Science,  
Hubli-580022 (Karnataka) India***Abstract:**

Congenital cystic adenomatoid malformation of lung associated with Cystic dysplasia of kidney, cystic disease of liver with mixed gonadal dysgenesis is rare and is not reported in literature so far. Hence an attempt is made to present this rarest entity.

**Keywords:** Cystic malformation, Cystic Dysplasia, Mixed Gonadal Dysgenesis

**Introduction:**

Congenital Cystic Adenomatoid Malformation (CCAM) of lung is rare and is most common cause of neonatal death and cause of respiratory tract infection and development of pulmonary blastomas and adenocarcinomas in older children and adults [1]. CCAM is associated with other congenital malformations like intestinal atresia, congenital heart abnormalities, cleft palate, diaphragmatic hernia, skeletal abnormalities, hydrocephalous, renal agenesis, cystic renal dysplasia, encephalocele [2].

**Case Report:**

A 24 year female presented with 9 months of amenorrhea with 3<sup>rd</sup> degree of consanguineous marriage. Her obstetric history reveal G<sub>2</sub>P<sub>1</sub>L<sub>1</sub>, First child is 5 year old girl. This pregnancy is uneventful and she presented to our institute in active labour with breech presentation. Her past history, family history and investigations are not

contributory and ultrasonography findings are not available. Assisted breech delivery was conducted and baby did not cry immediately after birth. Suspected anomalous male baby weighing 3.21kg with mass in abdomen was sent for autopsy.

A linear midline incision was taken from symphysis menti to symphysis pubis. External examination revealed male baby with unilateral testis in scrotal sac. All organs removed enblock. On dissection right lung was larger than left and was cystic on cut section with irregular cysts measuring 0.2-0.5cms in diameter. Both kidneys were enlarged and were cystic with loss of fetal lobulation on cut section. There were irregular cysts larger measuring 0.3cms in diameter filled with mucin fluid with intervening immature mesenchymal tissue. Pelvic mass reveal large uterus cervix with unilateral tube with bilateral attachment of both ureters with attached bladder. Cut section show endometrial cavity filled with mucoid material (Fig. 1 and 2).

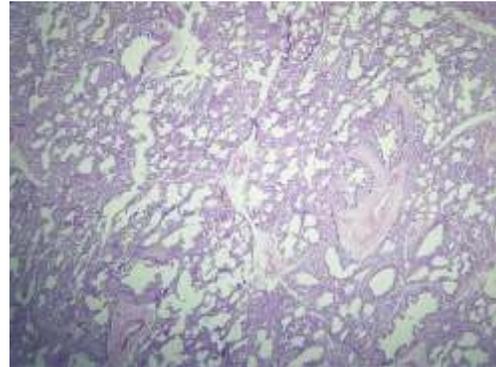
Microscopy revealed congenital cystic adenomatoid malformation of lung type II (Fig. 3), Cystic liver disease (Fig. 4) bilateral cystic renal dysplasia (Fig.5), Uterus with endometrial cavity (Fig. 6) Testis with epididymis with seminiferoustubules with areas of hemorrhage (Fig. 7)



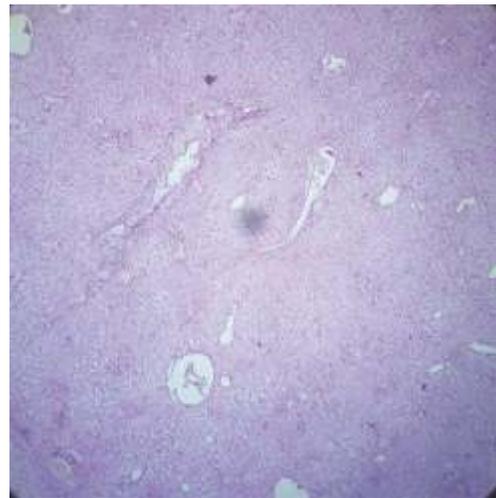
**Fig. 1: Specimen of Dissected Male Baby with Enblock Removal of Organs (I) External Genitalia of Male Type (ii) Enblock of All Organs (iii) Cut Section of Cystic Dysplasia of Kidney with Uterus**



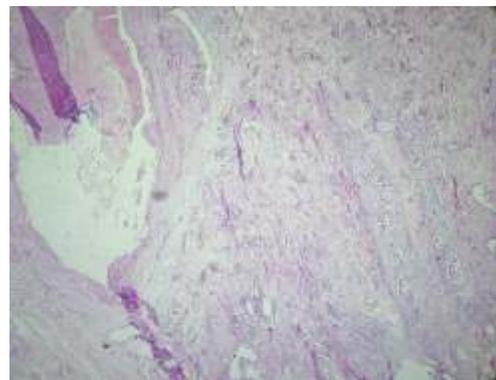
**Fig. 2: Specimen of Cystic Dysplasia of Kidney with Ureter and Uterus (Probe) (ii) Cut Section of Cystic Dysplasia of Kidney (iii) Cut Section of Congenital Cystic Adenomatoid Malformation of Lung**



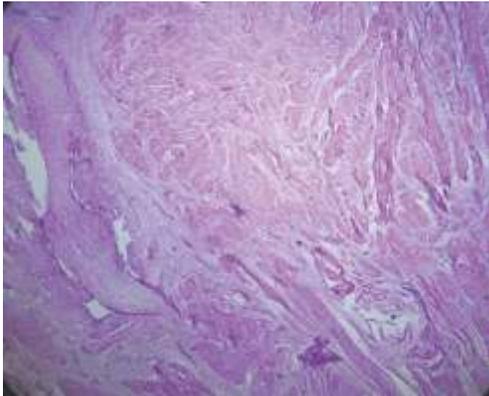
**Fig. 3: Section of Lung showing Congenital Cystic Adenomatoid Malformation of Lung Type-2**



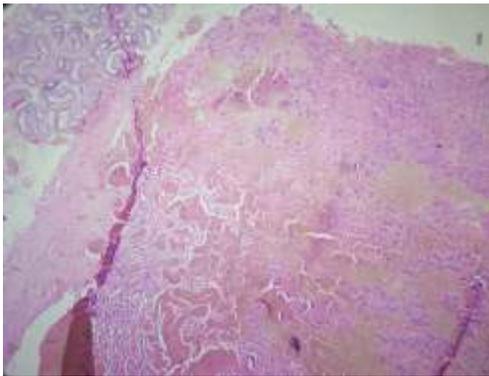
**Fig 4: Section of Liver showing Cystic Change**



**Fig. 5: Section of Kidney showing Cystic Renal Dyplasia with Areas of Immature Mesenchymal Tissue with Abortive Tubules**



**Fig. 6: Section of Uterus with Endometrial Cavity**



**Fig. 7: Section of Testes with Epididymis showing Seminiferous Tubules with Areas of Hemorrhage**

#### Discussion:

This entity is known since 1897 [3], but the term Congenital Cystic Adenomatoid Malformation (CCAM) was introduced in 1949 and occurs with equal frequency in both lungs with rare bilateral involvement [4]. In 1973 three varieties were recognised cystic, intermediate and solid [5]. Stocker recommended that these be termed types 1, 2 and 3 and later added types 0 and 4. This type 0 a condition previously described as acinar dysplasia [6-8], is described as bronchial type, type 1 as bronchial/ bronchiolar, type 2 as bronchiolar and type 3 as bronchiolar/ alveolar duct and type 4 as peripheral.

Congenital cystic adenomatoid malformation of lung is rare, the reported incidence being between 1:25,000 and 1:35,000 [9]. The Pathogenesis is attributed to maturation defect [2, 10], but recently revealed bronchial obstruction (atresia) with downstream developmental consequences. They are mainly diagnosed in stillborn fetuses or newborn infants but some are not detected until adult life [11]. Rare cases show malignant transformation usually to broncho alveolar carcinoma [12]. It is a proliferative lesion that is usually symptomatic in the first days of life [8]. Adults with CCAM have been reported [13, 14]. Some patients with CCAM may present with Pneumothorax [15], repeated respiratory tract infection with pneumatocele with pleural effusion [16]. These patients showed clinical and pathological evidence of chronic inflammation which is not seen in neonatal cases.

An expanded concept of CCAM has been presented by stocker [17] who has divided CCAM into five different categories based on the proposed site of the defect in the trachea bronchial tree.

Type 0 described by Rutledge and Jensen [8] as acinar dysplasia is incompatible with life and is associated with cardiovascular anomalies and dermal hypoplasia. The lungs are firm and small.

Type 1 lesion is characterized by multiple large cysts or occasionally a single dominant cyst with smaller cysts surrounding larger structure. The cysts communicate with the bronchial tree of affected lobe. A smooth glistening membrane lines the cysts. On microscopic examination large cysts are lined by ruffled layer of ciliated columnar to pseudostratified tall columnar epithelium overlying a thin to moderately thick fibromuscular layer. Elastic tissue is greater than normal. Large clusters of mucogenic cells along the walls of the

larger cysts or in smaller cysts or alveoli in the adjacent parenchyma were seen in this type but rarely seen in other types. Cartilage may be seen.

Type 2 lesion is composed of multiple evenly spaced cysts that rarely exceed 1.2cms in diameter. The cysts communicate with the bronchial tree and contain air if infant has breathed. The cysts are lined by a smooth or wrinkled glistening membrane or cysts may be lined by ciliated cuboidal to columnar epithelium overlying a fibromuscular layer. Cartilage is present in 5-10% of cases.

Type 3 lesion is bulky, firm mass that causes mediastinal shift in all cases, more commonly smaller cysts resembling evenly spaced bronchi can be seen distributed throughout the lesion. On the microscropy cysts are lined by simple to intricately folded ciliated cuboidal epithelium that extends onto adjacent alveoli.

Type 4 lesion is the peripheral type with presumed distal acinar origin. It is seen with equal frequency in both sexes with an age range of 0-4 years. It may present with sudden respiratory distress from tension pneumothorax. Grossly, the cysts are located at the periphery of the lobe. Microscopically they are lined by flattened epithelial cells over most of the wall, with occasional cuboidal epithelium. The wall is

composed of loose mesenchymal tissue with prominent arteries and arterioles. Congenital cystic adenomatoid malformation of lung may be associated with other congenital lesions such as cardiac abnormalities such as atrioventricular defects, intestinal atresia, renal agenesis, encephalocele, hydrocephalous, cleft palate, diaphragmatic hernia, cystic renal dysplasia and skeletal abnormalities. In the present case there is bilateral congenital cystic adenomatoid malformation of lung associated with cystic disease of liver, bilateral cystic renal dysplasia with mixed gonadal digenesis (ambiguous genitalia). CCAM of lung with cystic renal dyplasia with mixed gonadal dysgenesis is not reported in the literarture so far and present case is unique entity.

#### **Conclusion:**

Cystic adenomatoid malformation of lung is rare and its association with other congenital abnormalities is still rare. CCAM of lung associated with cystic renal dysplasia with mixed gonadal dysgenesis is rarest and is not reported in literature so far.

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**Author for Correspondence:** Dr. N. S. Kamakeri, Department of Pathology, Karnataka Institute of Medical Science, Hubli-580022 India Email: drkamakerins2013@gmail.com Cell: 9448236242