Identification of an Aberrant Karyotype in Bronchiectasis with Congenital Anomalies - A Case Report

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1. Introduction

Bronchiectasis is defined by permanent and abnormal widening of the bronchi [1]. Bronchiectasis, as a clinical syndrome, has the classical "symptoms triad" of cough, excess sputum production and repeated infection [2]. Polydactyly is one of the most common congenital deformities of the hands. It can occur as an isolated disorder, in association with other malformations of the hands or feet, or as part of a syndrome. It can occur sporadically but it can also be inherited with a mainly autosomal dominant inheritance [3]. A common and conspicuous congenital hand anomaly, polydactyly commonly involves only the hand or the foot. Polydactyly involving both hands and feet is rare [4]. Genetic defects in cilia, motile and sensory organelles with important roles in human development, also has been found to produce a host of disease symptoms, including polycystic kidney disease, hydrocephalus, retinal degeneration, chronic bronchiectasis, infertility, and polydactyly [5].

2. Case Report

We present a case of 33 yr old female, who presented with history of cough and breathlessness of one month duration. The cough was productive, with mucoid sputum. The patient had no complaints of chest pain and had no significant past history of any heart disease. On examination, patient was conscious, oriented, and afebrile. Patient had no pallor/ jaundice/ cyanosis/ clubbing/ lymphadenopathy/ pedal edema or goitre. She was short statured, with height of 142cm, and was moderately nourished. She had polydactyly in all four limbs. The limbs exhibited short broad digits with dysplastic nails. The examination of the respiratory system had features of bilateral lower lobe bronchiectasis with above congenital abnormalities. Other system examinations were normal.

Chromosomal aberration analysis was carried out by the following standard procedure (Hoyos et al., 1996). Briefly, 0.5 ml of whole blood was added to 5.0 ml RPMI 1640 medium (Hyclone), supplemented with 20% fetal bovine serum (PAA Laboratories), 2mM L-glutamine (Himedia), 1% streptomycin-penicillin antibiotic (Himedia) and 0.2 ml of phytohemagglutinin (Gibco). The mixture was incubated at 37°C for 72 hours. After 71 hours, the cells were treated with 0.01% Colchicine (Hi media) to arrest cells in mitosis. Lymphocytes were harvested upon the completion of 72 hours by centrifuging the cells at 1800 rpm for 7 minutes. About 6mL of pre-warmed (37oC) hypotonic solution (Kd 0.075 M) was added and left aside for 20 minutes at room temperature.

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After removing the hypotonic solution by centrifugation, the cells were fixed in Carnoy’s fixative. Slides were prepared and stained in 2% Giemsa stain. For the chromosomal aberration analysis, 100 well spread metaphase plates were examined per subject under a microscope (100X) to identify numerical and structural chromosomal aberrations [6].

### 3.2. Clinical Investigations

Chest X-Ray showed ectatic changes in both lower lung fields. Blood complete hemogram showed mild anaemia of 10.2 gm%. Other parameters were normal. Basic biochemical investigations like renal function tests, liver function tests were normal. Urine analysis was normal. Sputum microscopic examination for Acid Fast Bacilli was negative. Sputum Culture had NO growth. Further patient was subjected for karyotypic study, which revealed the following Karyotype (GTG banding technique at 450 -550 band resolution) – 46, XX 15q-.

### 3.3. Clinical Diagnosis

Congenital bilateral lower lobe bronchiectasis with short stature and Polydactyly

### 4. Discussion and conclusion

Bronchiectasis is a structural abnormality characterized by abnormal dilation and distortion of the bronchial tree, resulting in chronic obstructive lung disease. There is good evidence for believing that the disease is usually of acquired origin, but in a few cases the presence of other developmental anomalies favours the view that there may also be a congenital factor. Here the additional anomalies like polydactyly support a congenital development of the condition. The proper formation of alveoli does not occur in portions of at ectatic lung, so that the evacuating mechanism of an expulsive blast of air through the bronchi on expiration or coughing is not established. Though secreted mucus may become infected if not removed adequately, here sputum was not infected though bronchial weakening and dilatation was found. The presence of an abnormal karyotype also points towards a genetic defect but it needs a larger study for confirmation.

Primary ciliary dyskinesia (PCD), previously known as immotile cilia syndrome, is an autosomal recessive hereditary disease that includes various patterns of ciliary ultrastructural defects. The literature shows that sinusitis, bronchiectasis, and digital clubbing are late complications of PCD that can progress to chronic cor pulmonale and its consequences. There have been reports in the literature that other malformations, such as hydrocephalus, cleft palate, cardiac malformations, polydactyly, hypospadia, can accompany PCD [7]. Hence this case points to such defects but the clinical diagnosis needs confirmation through transmission electron microscopy.

Altogether this case report presents one of the rare conditions of congenital bilateral lower lobe bronchiectasis along with other congenital anomalies in form of polydactyly and short stature along with a previous unreported chromosomal abnormality - 46, XX 15q-.

### 5. Acknowledgement

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![Figure 1. Short stature of the patient](image1.png)

![Figure 2. Polydactyly condition in Feet](image2.png)
Figure 3. Polydactyly condition in Hand

Figure 4. Dysplastic Nail Conditions

6. References


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