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Bronchiectasis and Retinitis Pigmentosa: A Rare Association Suggestive Of Primary Ciliary Dyskinesia

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Abstract:
Primary ciliary dyskinesia (PCD) is a rare hereditary disease; it can rarely coexist with other rare disorders such as retinitis pigmentosa which is a hereditary cause of blindness by retinal ciliary dysfunction. This association is not obvious at first sight. We report the case of a patient with retinitis pigmentosa and bronchiectasis as the first apparent symptom of PCD

Key Words: *primary ciliary dyskinesia, bronchiectasis, retinitis pigmentosa.*

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I. Introduction:

Primary ciliary dyskinesia is a rare disease inheritance in most cases characterised by ciliary dysfunction, generally (but not always) associated to structural abnormalities of the cilia. The frequency is estimated at 1/12,500 live birth.

The association between PCD and retinitis pigmentosa (RP) is not obvious at first sight. We report the case of a patient suffering from retinitis pigmentosa and bronchiectasis as the first apparent symptom of PCD.

II. Case Report:

A 17 year old adolescent, with a history of neonatal respiratory distress and the onset of night blindness in childhood, he had Family history of retinitis pigmentosa in 2 uncles on the maternal side. The patient presented since the age of 2 years a cough with chronic bronchorrhoea with a notion of recurrent bronchitis associated with chronic rhinitis. The clinical examination found diffuse crackles at auscultation and a digital hippocratism. Chest X-ray (Figure 1) reveals Tram-track opacities and air-fluid levels;

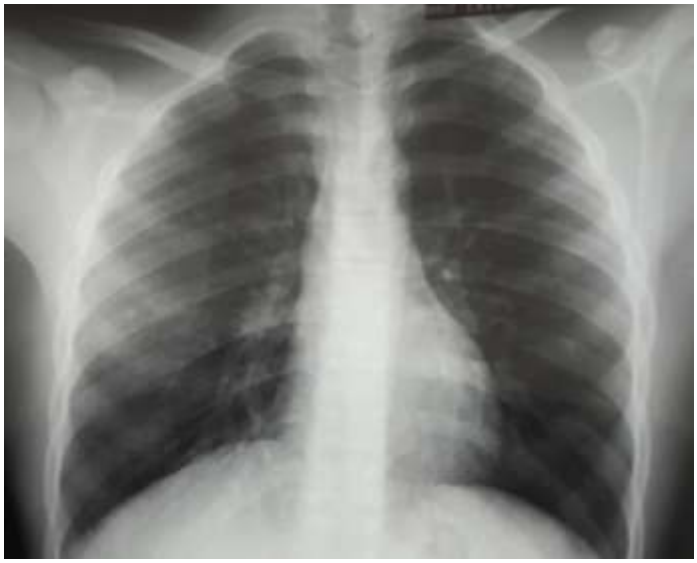

Figure 1: Chest X-ray revealing tram track opacities

Thoracic CT scan (Figure 2,3) reveals bilateral and diffuse bronchiectasis.

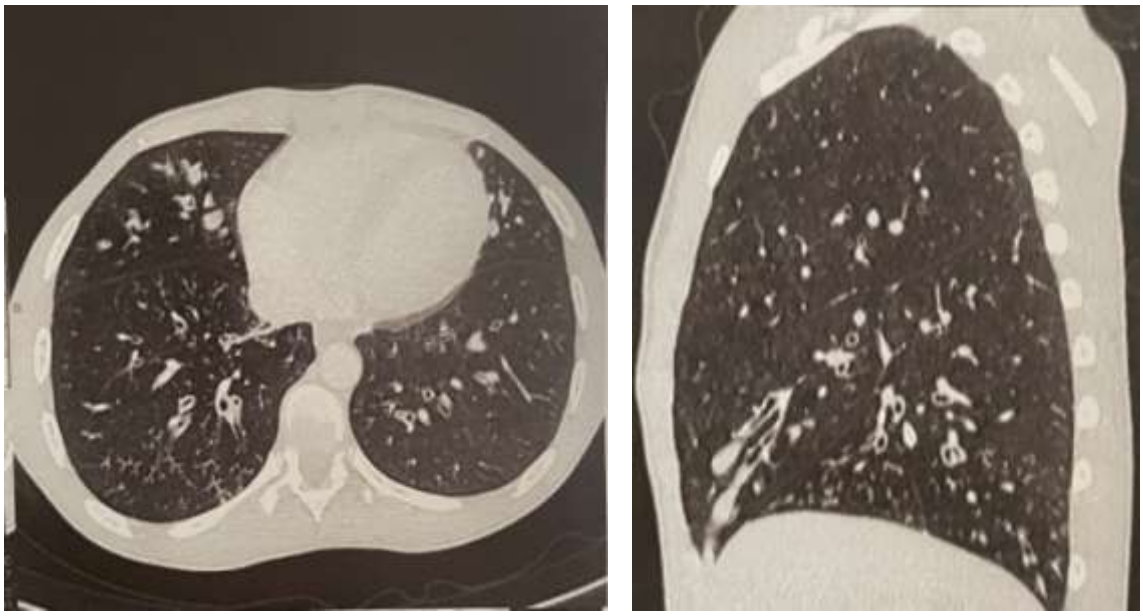

Figure 2, 3: Thoric CT scan revealing bilateral bronchiectis

An ophthalmological assessment was performed: The electroretinogram (ERG) showed the dysfunction of the rods and cones. The examination of the back of the eye (Figure 3) showed the presence of pigmented deposits (scotomas) thus confirming the diagnosis of retinitis pigmentosa.

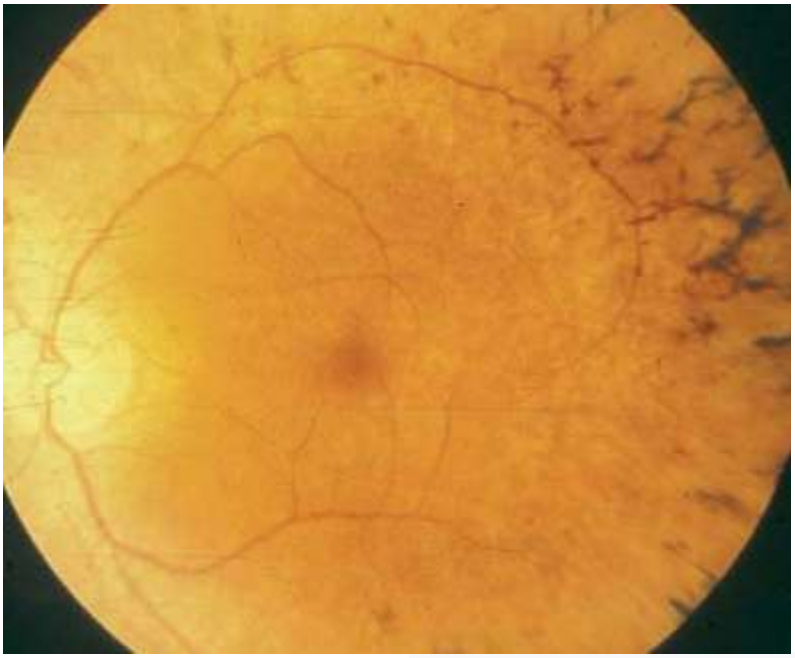

Figure 3: Fundus photographs showing pigment deposits in the mid periphery

The biological assessment was without particularities; protein electrophoresis finds a hypo albuminemia without alpha1-antitrypsin deficiency, the sweat test and the saccharin test returned negative. Plethysmography reveals a restrictive ventilatory disorder with a vital capacity at 1.58 (37%), FEV1.58 (42%), with a FEV/FVC ratio at 100%, total lung capacity at 3.32 (57%) RV at 122, the diffusion capacity was low at 56%. The diagnosis was retained after eliminating differential diagnoses such as cystic fibrosis or immune deficiencies and demonstrating the characteristic association of rhinitis, retinitis pigmentosa and bronchiectasis. Therapeutic management was multidiscidiciplinary aprouach between pulmonogist ophthalmologist otologist surgeons.

Pulmonary management was mainly based on respiratory physiotherapy the clearance of mucus, prevention of respiratory infections, and vigorous treatment of bacterial infections. Annual influenza and pneumococcal vaccinations have been prescribed

III. Discussion:

Primary ciliary dyskinesia (PCD) is a genetically heterogeneous, rare lung disease causing chronic oto-sino-pulmonary disease and irreversible lung damage that may progress to respiratory failure. The estimated incidence of PCD is approximately one in 10,000–40,000 live births(1)

PCD results from a functional and structural defect of cilia (2), leading to impaired mucociliary clearance responsible for recurrent respiratory infections, mainly bronchiectasis and chronic sinusitis with nasal polyposis and serous otitis.

PCD can rarely co-exist with other rare disorders including Retinitis Pigmentosa wich is an inherited cause of blindness from retinal ciliary dysfunction. they are X-linked disorders involving ciliary genes, RPGR (3). Although these account for a very small minority of PCD cases, there may be further overlap of retinal and respiratory cilia. Thus, retinal examination is recommended in individuals with PCD due to gene mutations in RPGR, clinical visual disturbances, or a family history of Retinitis Pigmentosa (our patient had Family history of retinitis pigmentosa in 2 uncles on the maternal side, a night blindness and the ophthalmological examination had confirmed the diagnosis (4)

The clinical presentation of PCD is aspecific and overlaps with other common chronic respiratory diseases. However, there are some phenotypic clues that may lead physicians to early diagnosis of PCD and eventually early management and better prognosis (5).

Most PCD patients (73%–91%) present in the neonatal period with neonatal respiratory distress, which is commonly attributed to congenital pneumonia and transient respiratory distress (transient tachypnea of the newborn) (6). Chronic nasal congestion is another aspecific presentation that is considered one of the hallmark features of PCD, especially if present at birth

In early childhood, chronic upper and lower respiratory tract infections are the common presentation (wich is the case of our patient)

Most men with PCD have infertility secondary to sperm immotility as a result of defective sperm-flagella movement (7)

The European Respiratory Society guidelines for the diagnosis of PCD recommend an evaluation for PCD if several of the following clinical features exist: persistent wet cough, situs anomalies, congenital cardiac defects, persistent rhinitis, chronic middle-ear disease with or without hearing loss, history in term infants of neonatal upper and lower respiratory symptoms, or neonatal intensive care admittance (8).

(Our patient had a neonatal respiratory distress, night blindness a persistent wet cough persistent rhinitis and a chronic respiratory tract infection)

The present association of PCD and RP in our case is not classic at all and has only been described in some few cases The clinical and genetic heterogeneity of PCD contribute to the challenges of diagnosis. The diagnosis of PCD is still difficult, despite the availability of sophisticated diagnostic tests. Currently, diagnosis incorporates multiple complex and expensive technologies, including nasal nitric oxide (nNO), high-speed video microscopy analysis and transmission electron microscopy, genetic testing, and IF of ciliary proteins. The diagnosis of PCD can be confirmed by genetic testing (5).

There is no "gold standard" reference test (8), The combination of bronchiectasis, rhinitis and Retinitis Pigmentosa is important because it does not have any other pathophysiologic link than the primary ciliary dyskinesia (as in our case). As a result, this combination provides valuable clinical data about an entity whose conclusion is complex

The aim of PCD treatment is to maintain or recover lung function by early detection and aggressive management of complications. The main objectives are the clearance of mucus, prevention of respiratory infections, and vigorous treatment of bacterial infections.

Prophylactic courses of antibiotics are used in some centers, but this is not routinely recommended. Azithromycin is known to have antibacterial, anti-inflammatory, and anti-quorum-sensing properties and is commonly used in chronic respiratory diseases chronic macrolide therapy in PCD demonstrate some benefits(9)

PCD patients should receive recommended vaccinations . Annual influenza and pneumococcal vaccinations are recommended(11).

Bronchiectasis is a prominent feature in PCD, and surgical resection with lobectomy or segmentectomy in patients with bronchiectasis is thought to decrease the risk of infection progressing into healthier lung tissue.

IV. Conclusion:

PCD is a genetically heterogeneous recessive condition with defective ciliary motility, it's co existing with retinis pigmentosa is extremely rare. Diagnosis is based on the identification of abnormalities in ciliary structure and function through highly specialized examinations leading to therapeutic management. Due to its rarity and lack of clinical trials, the management of PCD remains challenging and mainly supportive.

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