PRIMARY CILIARY DISQUINESIA RELATED BRONCHIECTASIS. RELEVANCE OF PIGMENTARY RETINITIS AMONG CLINICAL DATA.

BRONQUIECTASIAS NO FQ DEL ADULTO RELACIONADAS CON DISQUINESIA CILIAR PRIMARIA. LA RELEVANCIA DE LA RETINITIS PIGMENTOSA ENTRE LOS DATOS CLÍNICOS.

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Non-CF bronchiectasis (NCBE) is a persistent disease and sometimes progressive, characterized by the permanent dilation and thickening of the bronchi.

It can be associated to a large and heterogeneous group of diseases, although its importance may range from an unexpected discovery in an image or in the histopathology to a severe disease that compromises the respiratory function and the overall status. [1, 2]

They are known to be, at present, as an entity that contributes to respiratory morbidity, particularly in countries that have worst health and socio-economic conditions.

The diagnosis, usually oriented by its clinical presentation, it is frequently different from the classic bronchorrhoeic form, may go unnoticed for years, both in children and adults, and in some cases, the disease will cause a significant, yet slow and progressive, deterioration of the respiratory function. [3]

Among the entities associated to the development of NCBE, the Primary Ciliary Dyskinesia is described. This disease's definite diagnosis can be very hard to reach. [2]

In cases like this, it is essential to recognize the clinical data that support the clinical suspicion so that to apply complementary diagnosis methodology that is expensive, debated, hardly available and of complex interpretation.

The right selection of patients, based on clinical data, and the improvement in the specificity of the complementary diagnosis methodology would allow the practitioner to identify the sick, with research goals and including data originating from more numerous groups, as the experts authors of a recently published article in the European Respiratory Journal expressed. [4]

As suggested by relevant clinical data: persistent productive cough, mediastinal abnormality in the rotation, persistent rhinosinusitis, chronic middle ear disease with or without defecting hearing, lower and top respiratory symptoms, admission into intensive care in children born full-term, wheezing, asthma, pneumonia, bronchiectasis and infertility. [4]

The absence of mediastinal abnormality in the rotation does not exclude a clinical suspicion.

Regarding this disease, a broad range of aspects affect the disease's presentation, including: numerous ultra-structural defects identified by ciliar electron microscopy, alteration of the ciliar motility, that ranges from still, to disorganized, rigid and hyperkinetic, the multiple known original genetic defects and the functional consequences.

The ciliar motility is altered as well in inflammatory or infectious processes and secondary ciliary dyskinesia. This should be taken into account when obtaining the tissue or studying the ciliar functions using nasal nitride oxide, for example. [5]

In recently published guidelines, one of the injuries that usually accompany the NCBE and that greatly impact the life quality of patients: the Retinitis Pigmentosa. It should be mentioned that it is mentioned in the 2009 consensus. [4, 6]

The combination of NCBE and Retinitis Pigmentosa is important because it does not have any other pathophysiologic link than the DCP. As a result, this combination provides valuable clinical data about an entity whose conclusion is complex. [7, 8]

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There are, basically, two groups of cilia, one whose functions depend on motility and other sensitive. The respiratory consequences are related to the dysfunction in the former and Retinitis Pigmentosa to the latter. [9]

The multiplicity of genetic defects that may affect the structure and/or the function of the different type of cilia could potentially generate numerous phenotypes, which identification is key to apply the delicate methods used to establish diagnosis.

The NCBE and Retinitis Pigmentosa phenotype, associated to defects in the mobile and sensitive cilia could provide therefor valuable data so it is important to acknowledge that.

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