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SCIENTIFIC LETTER

A Novel Homozygous Nonsense *HYDIN* Gene Mutation p.(Arg951*) in Primary Ciliary Dyskinesia

Antony Terance Benjamin[1] · Ram Ganesh[1] · Balan Louis Gaspar[1] · Jane Lucas[2] · Claire Jackson[2] · Marie Legendre[3] · Rahma Mani[3] · Estelle Escudier[3]

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To the Editor: Primary ciliary dyskinesia (PCD) is a rare autosomal recessive disorder that disrupts the structure and function of motile cilia thereby resulting in impaired muco-ciliary clearance. In PCD, cilia are immotile or dyskinetic. Clinical phenotype includes term neonate with respiratory distress, early onset persistent wet cough, purulent nasal discharge, otitis media and later, bronchiectasis and fertility issues. Around one-half of PCD patients have situs inversus. In a patient with typical phenotype, the diagnosis of PCD is made using a combination of tests [1]. We report for the first time a novel loss of function *HYDIN* gene mutation in two siblings with PCD.

An 18-mo girl and her elder sibling, a 5-y-old boy, born out of consanguineous marriage, presented with persistent productive cough and recurrent respiratory infections since infancy. Both of them were born at term and had neonatal respiratory distress. They were thriving well with no digital clubbing but had purulent nasal discharge. Chest examination revealed occasional crackles. There was no family history of atopy. Chest X-rays and immunoglobulin profiles were within normal limits.

Their PICADAR predictive score was 7 [2]. High speed video microscopy (HSVM) analysis of nasal brushings showed motile dyskinetic cilia with uncoordinated and stiff motility. An additional observation of rotational ciliary movement was observed in elder sibling. Targeted NGS of 38 genes known to be associated with PCD was perfomed and homozygous *HYDIN* c.2851C > T p.(Arg951*) nonsense mutation was identified in both siblings (Fig. 1) [3].

HYDIN (hydrocephalus-inducing protein homolog) gene encodes for axonemal central pair apparatus protein considered important for coordinated ciliary motility. Olbrich et al. identified a PCD associated locus on chromosome 16q21-q23 which includes the *HYDIN* gene and published on a series of PCD patients with *HYDIN* mutations [4]. Individuals with PCD harboring *HYDIN* mutations usually have normal situs and ciliary ultrastructure [5]. Our patients also had normal situs. We hereby document a novel homozygous

✉ Balan Louis Gaspar
louisbalan@gmail.com

[1] Department of Molecular Pathology, Primary Ciliary Dyskinesia Unit, G Kuppusamy Naidu Memorial Hospital, Coimbatore 641037, India

[2] Primary Ciliary Dyskinesia Centre, University Hospital Southampton NHS Foundation Trust, Southampton, UK

[3] U.F. de Génétique moléculaire, Hôpital Armand Trousseau, Paris, France