DNAI1 mutations explain only 2% of primary ciliary dykinesia

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Abstract

BACKGROUND: Primary ciliary dyskinesia (PCD) is a rare recessive hereditary disorder characterized by dysmotility to immotility of ciliated and flagellated structures. Its main symptoms are respiratory, caused by defective ciliary beating in the epithelium of the upper airways (nose, bronchi and paranasal sinuses). Impairing the drainage of inhaled microorganisms and particles leads to recurrent infections and pulmonary complications. To date, 5 genes encoding 3 dynein protein arm subunits (DNAI1, DNAH5 and DNAH11), the kinase TXNDC3 and the X-linked RPGR have been found to be mutated in PCD.

OBJECTIVES: We proposed to determine the impact of the DNAI1 gene on a cohort of unrelated PCD patients (n = 104) recruited without any phenotypic preselection.

METHODS: We used denaturing high-performance liquid chromatography and sequencing to screen for mutations in the coding and splicing site sequences of the gene DNAI1.

RESULTS: Three mutations were identified: a novel missense variant (p.Glu174Lys) was found in 1 patient and 2 previously reported variants were identified (p.Trp568Ser in 1 patient and IVS1+2_3insT in 3 ...

Reference

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Figure E1: Distribution of mutations in the DNAI1 gene

Numbered exons and introns are shown according to their respective position to genomic structure (hatched lines depict intron length $> 2500$ bp) are drawn into scale in genomic structure of gene DNAI1. Black boxes represent 5’- and 3-untranslated regions. Mutations are identified by vertical annotation above their location in the gene. Known mutations are depicted as well as SNPs that are in underlined italics ($p$.Ala8Ser, $p$.Val335Ile) and novel mutation ($p$.Glu174Lys) is depicted in bold.