Genetic Testing for Joubert Syndrome in the Jewish Population

Tech ID: 19946 / UC Case 2009-359-0

FULL DESCRIPTION

The "ciliopathies" are a newly emerging group of diseases due to defects in the function or structure of cellular primary cilia, which are small cellular appendages previously of unknown function. UC San Diego researchers and colleagues have identified five genes for Joubert Syndrome (JS), which is a ciliopathy that is characterized by cerebellar ataxia, blindness, renal failure, and mental retardation. Most of these mutations occur randomly throughout the gene, which makes genetic diagnosis very laborious.

Researchers found that all Jewish patients with JS share a common mutation in a newly identified gene. Of the Jewish families tested thus far, 100 percent of the patients were homozygous for the point mutation. It is anticipated that this discovery will make it possible to perform genetic testing in this population easily.

PATENT STATUS

Country	Type	Number	Dated	Case
United States Of America	Issued Patent	9,879,322	01/30/2018	2009-359
United States Of America	Issued Patent	8,614,094	12/24/2013	2009-359
United States Of America	Published Application	20180195126	07/12/2018	2009-359

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OTHER INFORMATION

CATEGORIZED AS

- Medical
  - Diagnostics
  - Disease: Genetic Diseases and Dysmorphic Syndromes
  - Gene Therapy

RELATED CASES

2009-359-0