Title (en)

USE OF GENETIC MARKERS TO DIAGNOSE FAMILIAL DYSAUTONOMIA

Title (de)

VERWENDUNG VON GENETISCHEN MARKERN ZUR DIAGNOSTIK VON FAMILIÄRER DYSAUTONOMIAE

Title (fr)

UTILISATION DE MARQUEURS GENETIQUES POUR DIAGNOSTIQUER LA DYSAUTONOMIE FAMILIALE

Publication

EP 1274860 A4 20060208 (EN)

Application

EP 00916382 A 20000315

Priority

US 0006851 W 20000315

Abstract (en)

[origin: WO0168917A1] Familial Dysautonomia (FD), is an autosomal recessive disorder characterized by developmental arrest in the sensory and autonomic nervous systems and Ashkenazi Jewish ancestry. The familial dysautonomia disease gene (DYS) has previously been mapped to an 11cM segment of chromosome 9q31-33 flanked by D9s53 and D9S105. Using new polymorphic loci, the location of the gene is narrowed to less than 0.5 cM between the markers 43B1GAGT and 157A3. Two markers in this interval, 164D1 and D9S1677, show no recombination with the disease. Haplotype analysis confirmed this candidate region. The identification of these close flanking markers of the familial dysautonomia disease gene allows accurate genetic testing for both familial dysautonomia families and carriers.

IPC 1-7

C12Q 1/68; C07H 21/04

IPC 8 full level

C12Q 1/68 (2006.01); **C12Q 1/6883** (2018.01)

CPC (source: EP)

C12Q 1/6883 (2013.01); C12Q 2600/156 (2013.01); C12Q 2600/172 (2013.01)

Citation (search report)

- No further relevant documents disclosed
- See references of WO 0168917A1

Citation (examination)

WO 9324657 A2 19931209 - GEN HOSPITAL CORP [US]

Designated contracting state (EPC)

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DOCDB simple family (publication)

WO 0168917 A1 20010920; AU 3749200 A 20010924; CA 2403696 A1 20010920; EP 1274860 A1 20030115; EP 1274860 A4 20060208; IL 151769 A0 20030410

DOCDB simple family (application)

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