

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 (<i>DYS</i>) has previously been mapped to an 11cM segment of chromosome 9q31-33 flanked by <i>D9s53</i> and <i>D9S105</i>. 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 <i>D9S1677</i>, 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]

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DOCDB simple family (publication)  
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