CENTOGENE
THE RARE DISEASE COMPANY

Patient diagnosed with juvenile Parkinson disease after detecting the bi-allelic and compound heterozygous mutations in the PARK2 gene

Oliver Brandau, MD[1] • Aida Bertoli-Avella, MD[1] • Maria Farag, PhD[1] • Slobodanka Orolicki, MD[1]

[1] CENTOGENE AG, Rostock, Germany

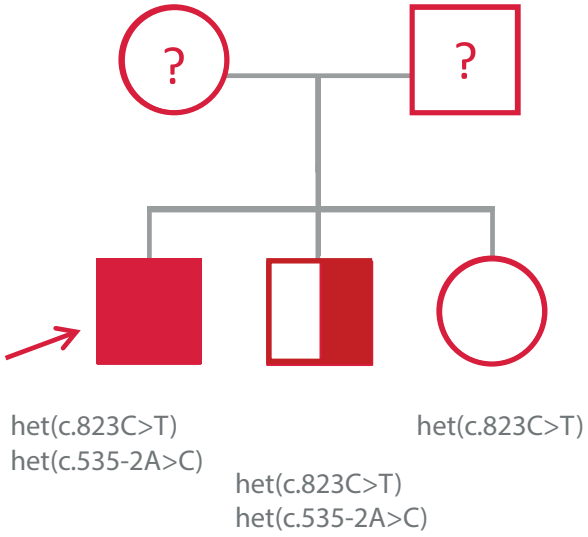
Georgraphic region

Argentina, South America

Clinical information

The patient is a young man (40 years) with a 3-year history of intentional tremor, spreading from one arm to the other. Symptoms are present on posture and rest.

Parents are unrelated, without history of toxic or drugs exposure. His younger brother has neuropathy of the lower limbs and slight intention and postural tremor.



Diagnostic procedure

A large number of genes could have been responsible for the patient's symptoms. Absence of family history did not allow for a focus on a reduced number of possible candidate genes. CentoXome® Trio Advanced was recommended for this patient.

End-to-end bioinformatics analysis and variant priorization found two heterozygous pathogenic variants in the **PARK2 gene** in this patient and his affected brother; c.823C>T and c.535-2A>C. The sister carries only the c.823C>T variant. Detected variants are in a compound heterozygous state (trans configuration) and thus, genetic diagnosis of juvenile Parkinson disease type 2 is confirmed.

Bi-allelic and compound heterozygous mutations in the PARK2 gene lead to Juvenile Parkinson disease type 2. Presentations consisting of rigidity, painful cramps followed by tremor, bradykinesia, dystonia, gait complaints and falls, and other non-motor symptoms, with age of onset between <20-40 years. Age of onset for both, the patient and the affected brother was in their mid-thirties (30-40 years of age).

CentoXome® Trio Advanced analysis helped identify the cause of the disease, select the most optimal treatment, and further help with genetic counselling for the family.

› Customer Service
Phone: +49 (0)381 203 652 - 222
Fax: +49 (0)381 203 652 - 119
E-mail: crd@centogene.com
www.centogene.com

CLIA #99D2049715

cap ACCREDITED™
15189SM cap ACCREDITED ISO 15189

V1.1_May2016