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Final Report

Date: 21.05.2014

Patient name:			
Patient no.:		Order no:	**62210171**
DOB: (dd.mm.yyyy)	**14.01.1980**	Order received: (dd.mm.yyyy)	**04.04.2014**
Sex:	**male**	Sample type:	**blood, EDTA**
Your ref.:	**PatID 10636726, SampleID 714001053**	Reviewer no.:	**8**

Request for: ATXN7 (OMIM: 607640) gene analysis - Spinocerebellar ataxia 7 (OMIM: 164500) – inheritance: autosomal dominant

Clinical information: no clinical information.

Result Details:

ATXN7 **no expanded allele**

A genetic diagnosis of spinocerebellar ataxia type 7 for the patient can be very likely excluded.

Comment:

We detected no expanded allele in the ATXN7 gene. We only found a homozygous allele with 11 repeats which is definitely within the normal range. Genetic counselling is recommended.

Best regards,

Prof. Arndt Rolfs, MD
Chief Medical Director

Nahid Nahavandi, PhD
Director Genetic Reporting

CLIA registration 99D2049715; CAP registration 8005167; ZLG-P-322.09.13 registration

Please note: scientific use of these results requires permission by the investigators. Note: if you would like to download your reports from our web portal, please contact us to receive your login and password. More information is available at www.centogene.com or support@centogene.com.

Additional information:
This test was developed and its performance validated by Centogene AG. The US Food and Drug Administration (FDA) has determined that clearance or approval of this method is not necessary and thus neither have been obtained. This test has been developed for clinical purposes. All test results are reviewed, interpreted and reported by our scientific and medical experts.

ZLG-P-322.09.13

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CENTOGENE
THE RARE DISEASE COMPANY

Patient name:	**A**		
Your ref.:	**PatID 10636726, SampleID 714001053**		
DOB (dd.mm.yyyy):	**14.01.1980**	Patient no.: **1049111**	Order no.: **62210171**

Methods:

The **ATXN7** gene(s) was analysed by PCR and capillary electrophoresis to screen the trinucleotide repeat region. In addition a repeat-primed assay (RPA) was performed to screen for a large expansion in the gene.

ATXN7 (Spinocerebellar ataxia 7, SCA7 -OMIM 164500)

Result of the fragment analysis:
allele 1 = 11 ± 1 repeats
allele 2 = 11 ± 1 repeats

CAG trinucleotide repeats in the ATXN7 gene:

non-pathogenic	mutable non-pathogenic	pathogenic with reduced penetrance	pathogenic with full penetrance
≤19	28-33	34-36	>36

From: *Bird TD, Pagon RA, La Spada AR (Updated September 6, 2007). Spinocerebellar Ataxia Type 7. In: GeneReviews at GeneTests: Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2011. Available at http://www.genetests.org.*