MGZ

Medizinisch Genetisches Zentrum

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Partnerschaft von Fachärztinnen für Humangenetik mbB, MVZ

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Dr. Franz K.
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GENETIC ASSESSMENT

Munich, 30.10.16/djp

Name	Mustermann, Erika	**Date of receipt / Start**	19.09.16 / 20.09.16
Date of Birth	15.05.85	**Sex**	female
ID / your Ref	MGZ 654321/	**Sample**	EDTA blood
Analysis	DN16/1234	**Test(s) requested**	*Chorea Huntington - HTT*
Indication	Asymptomatic, family history of Huntington's disease.		

RESULTS

NORMAL RESULT

		no expansion
Chorea Huntington - HTT	repeat analysis	allele: 17 / 22 repeats

INTERPRETATION

No pathogenic repeat expansion was detected in the HTT gene. Thus the clinical diagnosis of Huntington disease caused by a pathogenic repeat expansion in the gene HTT cannot be confirmed.

If, based on clinical symptoms, you would like to consider further genetic studies, we can offer an appropriate single gene analysis or analysis via one of our NGS multi-gene panels (see www.mgz-muenchen.com for available analyses).

METHOD

PCR and fragment length analysis to determine the number of CAG repeats in exon 1 of the HTT gene (OMIIM: 613004, RefSeq: NM_002111.6). Testing of two different independent PCR reactions.

Allele lengths: Normal: <26 repeats; intermediary: 27-35 repeats; reduced penetrance: 36-39 repeats; pathological: >39 repeats.

Should you have any questions or concerns, please do not hesitate to contact us.

Validation
C. Dineiger
M. Wendlandt
PD Dr. med. I. Diebold

Prof. Dr. med. E. Holinski-Feder / PD Dr. med. A. Abicht

Certified in accordance with DIN EN ISO 15189:2014. Genetic analyses have a high, however not 100% sensitivity and, as with all laboratory analyses, have an error rate due to pre-analytic, analytic, and post-analytic processes. This test result is based on the scientific data available at the time of reporting. The interpretation of familial test results is only valid provided that the familial relations given are correct. The results of genetic testing should be disclosed within the framework of genetic counseling, especially in cases of a positive result.

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DAkkS Deutsche Akkreditierungsstelle D-ML-13242-01-00

Die Akkreditierung gilt für den in der Urkundenanlage D-ML-13242-01-00 festgelegten Umfang.

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[5] in Weiterbildung zum Facharzt für Humangenetik

GENETIC ASSESSMENT

Name	Mustermann, Erika	**Sample ID**	654321
Date of Birth	15.05.85	**Analysis**	*Chorea Huntington - HTT*

GENERAL INFORMATION

Huntington's disease (HD) is an autosomal dominantly inherited disorder which leads to progressive degeneration of nerve cells in specific regions of the central nervous system and manifests clinically as a movement disorder with psychiatric disturbances. Nearly all cases of HD are associated with a CAG triplet repeat expansion in the gene HTT located on chromosome 4p16.

A normal genotype in healthy individuals reveals 10 to 26 CAG repeats.

- Individuals with 27 to 35 repeats have no risk of developing HD themselves. In rare cases, however, meiotic instability may result in a further expansion of this allele into the pathological range and thus to the development of HD in one's children (this is an example of the phenomenon of anticipation), especially when inherited via the male germline.
- Patients with 36 to 39 CAG triplet repeats carry a mutation in the gene HTT and are at risk for developing HD, especially at an advanced age.
- Patients with more than 39 repeats develop the symptoms of HD during the normal course of their lives. In principle, the larger the number of triplet repeats, the earlier the age of onset and more severe the progression. Nevertheless, a precise prediction as regards age of onset and progression of the disease cannot be made.

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