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01.01.2026

Dept of Clinical Genetics
University Hopital
Street
postal code, City
Country

Name: **Doe, Joanne**
Date of birth: **01.01.2010** Gender: **F**
Order number: **000000000**

Reception date:	01.01.2026	Order date:	01.01.2026
Collection date:	01.01.2026	External No.:	XXX
		Reference No.:	XXX

Sample material: EDTA blood

Dear Sir or Madam,

Thank you for your order from 01.01.2026.

Order: molecular genetic analysis of the long-QT-syndrome panel
Diagnosis/ indication: suspicion of Long QT syndrome (LQTS),ventricular tachycardia, prolonged QTc interval (465-485 ms)

Result:

No evidence of a variant with clinical relevance.

Interpretation:

In the panel analysed here, no genetic cause for the clinical symptoms was detected. This result does not rule out a genetic cause for the disease. Possible limitations arise from both the technical and diagnostic sensitivity of molecular genetic testing.

Clinical management and further information:

Genetic counselling is recommended.
Molecular genetic reanalysis or expanded diagnostic testing may be considered should the patient develop new clinical symptoms.

Kind regards,



Scientific validation / inquiries
Dr. rer. nat. Alexandra Musterfrau
01.01.2026

Medical validation
Dr. med. Erika Mustermann
01.01.2026

Analysed genes:

ALG10B (NM_001013620.4), CACNA1C (NM_000719.7), CALM1 (NM_006888.6), CALM2 (NM_001743.6), CALM3 (NM_005184.4), KCNE1 (NM_000219.6), KCNE2 (NM_172201.2), KCNH2 (NM_000238.4), KCNJ2 (NM_000891.3), KCNQ1 (NM_000218.3), RYR2 (NM_001035.3), SCN5A (NM_000335.5), TECRL (NM_001010874.5), TRDN (NM_006073.4)

Geschäftsführer
Dr. med. Hartmut Campe, Dr. med. Konstanze Hörtnagel
Dr. med. Hanns-Georg Klein, Dr. rer. nat. Stefan Mehrle

Ärztliche Leitung
Dr. med. Konstanze Hörtnagel

Amtsgericht München
HRB 241161
USt - IdNr. DE320949669

Deutsche Bank AG
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Name:	**Doe, Joanne**	DOB:	**01.01.2026**	Gender:	**F**
Order number:	**000000000**	Collection date:	**01.01.2026**	Report date:	**01.01.2026**

Additional information:

Method			
Sequencing	**Enrichment**	**SNV and CNV analyses**	**Data evaluation**
Next Generation Sequencing (Illumina)	Twist Exome 2.0 plus Comprehensive + Custom Spike-in (54 Mb target sequence)	Illumina Dragen Bio-IT Platform	Franklin by QIAGEN
Quality criteria	**Reference genome**	**Databases**	**In silico algorithms**
Quality Score >30 (accuracy >99,9%) for at least 75% of bases; sensitivity of SNV detection 99.92%; sensitivity of CNV detection 91%	hg38, NCBI GRCh38 (transcript variant from HGMD is usually used)	HGMD Professional release, ClinVar, LOVD, gnomAD (data of the population with the highest allele frequency are usually considered (prerequisite: sufficient number of alleles examined)	REVEL, Splice AI

Limitations

Reduced sensitivity for mosaics (<20%), deletions/insertions (>20 bp), repeat expansions, homopolymers and pseudogenes. No reporting of benign/likely benign variants. Variants (or variant constellations) of uncertain significance are only reported if, in case of autosomal dominant or X-linked inheritance, the posterior probability of pathogenicity for SNVs is ≥50% (Tavtigian et al. 2018, Genet Med 20:1054), or in the case of autosomal recessive inheritance, if at least one of two variants in a gene has a posterior probability of pathogenicity ≥67.5%, or if the score for CNVs is ≥0.5 each (Riggs et al. 2020, Genet Med 22:245)

Variant classification

Variants within +/- 10 bp of coding exons are classified according to Richards et al. 2015, Genet Med 17:405, with modifications based on the ClinGen SVI (Sequence Variant Interpretation) general recommendations and the ClinGen expert panel specifications to the ACMG/AMP variant interpretation guidelines. CNVs are classified according to Riggs et al. 2020, Genet Med 22:245; In silico algorithms used: REVEL and SpliceAI; Databases consulted: HGMD® Professional release, LOVD, ClinVar, and gnomAD (considering data from the population with the highest allele frequency, provided a sufficient number of alleles are examined)

Coverage

20-fold coverage of target region: 99.59%

Note

According to § 10 GenDG, genetic counselling should be offered as part of genetic testing. In the case of a non-treatable disease, the responsible medical person must offer genetic counseling. In the case of a predictive test, genetic counseling must be offered before the test and after the result is available. This report may only be forwarded in full.

Geschäftsführer
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