Trio Exome Diagnostics all genes

for patients with rare diseases

State of the art sequencing

- For patients with **complex, unspecific, and rare diseases**
- High probability of identifying the genetic cause of the patient's disease
- Comparison of patient and parents, analysis of additional family members (e.g. siblings) possible
- Target region covers all coding genes (including flanking intronic regions)
- Average coverage: >100x coverage of >30x is achieved for more than 95% of target regions
- In-house bioinformatics – filtering for de novo, compound heterozygous, x-linked, and homozygous (with heterozygote parents) variants
- Most up-to-date exome enrichment from Agilent (**Sure Select Human All Exon V7**)
- Paired-End sequencing with 2x100 bp reads
- Interpretation includes single nucleotide variants (SNVs), small insertions and deletions (INDELs), and copy number variants (CNVs) of single and/or multiple exons

Quality Made in Germany

- Comprehensive and easy to understand medical report with interpretation of the findings and recommendations
- Issued by an interdisciplinary team of scientists and medical doctors specialized in human genetics
- Fast results within 4 weeks
- Entire service performed in-house
- Neither samples nor data leave CeGaT
- Excellent price-performance-quality ratio

Case report

Patient: Six month old girl

Symptoms: Congenital bone marrow failure, infections, anemia, dystrophy, reduced number of neutrophils and thrombocytes

Familial history: Older brother with same symptoms passed away at the age of seven months

Initial planned therapy strategy: Bone marrow transplantation

Results of Trio Exome Analysis: Homozygous mutation in the gene TCN2 (codes for TC II, a vitamin B12 carrier protein)

Therapeutic consequences: No bone marrow transplantation, high-dose therapy with vitamin B12 → child is able to live a normal life

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CLIA CERTIFIED ID: 99D2130225

www.cegat.de/exome

Medical Report

The medical report contains the results from the genetic testing, as requested by the referring physician.

1 Patient information

In the header, we display summarized patient information which has been communicated to by the referring physician:

- Name, date of birth, sex of the patient, and external ID where applicable
- Suspected diagnosis or indication for molecular genetic testing
- Request
- Our internally assigned patient ID

2 Results

On the first page of our report we summarize the identified genetic changes, tabulated and sorted by their disease relevance. You will find a table that addresses the most likely causative variant(s) and provides information on zygosity, inheritance, allele frequency in the population, in silico prediction, and our classification.

Just below the table, we provide details to aid in the interpretation of the information in the table.

3 Interpretation

In this paragraph, we summarize the current state of scientific literature for the variants found and discuss them in relation to the patient. The more clinical information that is provided by the referring physician, the more accurate and precise our evaluation can be. This is of particular importance with findings from panel or exome sequencing, as variants may be identified which do not yet have a clear clinical association.

We then explain in a concluding summary sentence the strength of the evidence and associations with the clinical features of the patient.

4 Variants of unknown significance

In some cases, the second table may also be employed to indicate further variants which may have an unclear association with the indicated disease. Variants will be placed in one of the two tables based upon, but not limited to; published data, frequency in the normal population, and pathogenicity predictions given by several pathogenicity prediction algorithms. These data are used to make an assessment on the role of the gene in disease, and the potential changes to the function of the expressed protein.

5 Genetic relevance

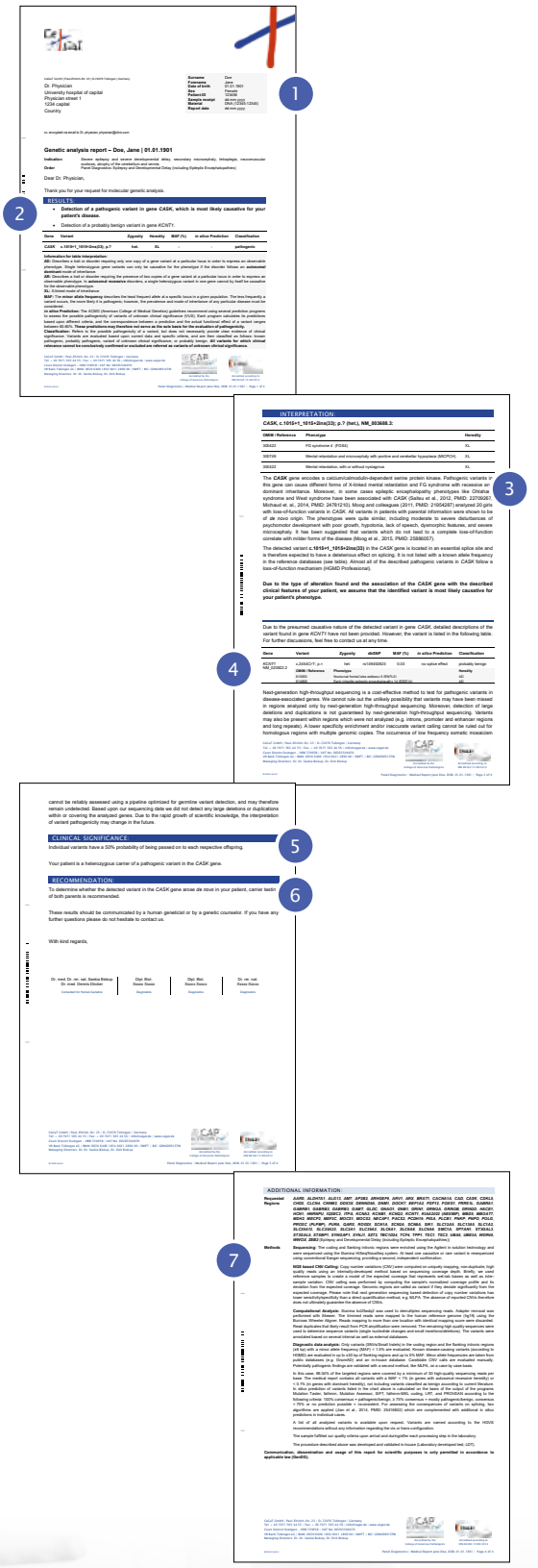
In this section, we explain the inheritance patterns; the possibility of other similarly affected family members, the risk of disease recurrence in the family, and the extent to which other family members may be unaffected carriers.

6 Recommendation

To address any remaining ambiguities regarding the causality of the variant, this section has space for additional recommendations. This could be further clinical investigations which may indicate the pathogenicity of the identified variants, or further molecular genetic tests for both affected and unaffected family members (e.g. segregation analysis).

7 Additional information

At the end of the report we present technical information. This includes information such as technology used and bioinformatic parameters.



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