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Freiburg Medical Laboratory
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P.O. Box 3068
Dubai, UAE

Munich, 28.04.2016

GENETIC REPORT

Patient:	**Klara Testfrau**				
DOB:	**11.11.1975**	Sample:	EDTA-Blut	Sampling date:	22.03.2016
Gender:	female	Sample No.:	**16/113300**	Sample receipt:	23.03.2016
Your reference:	123456			Order receipt:	23.03.2016
Indication:	suspicious for beta-thalassemia				

Dear Colleagues,
thank you for sending the sample for genetic testing.

Result: **The mutation c.-31C>T was detected in the *HBB* gene in heterozygous constellation; ß$^+$ -Thalassemia confirmed**

Methods: Genomic DNA was isolated and the *HBB* gene including the 5' and 3' regions was amplified and directly sequenced (reference sequence: ENSEMBL ENST00000335295).

Interpretation: The sequencing analysis revealed the mutation c.-31C>T in the *HBB* gene in heterozygous constellation. The result of the analysis is consistent with the clinical phenotype of a Thalassaemia minor.

Based on the autosomal dominant inheritance, the probability of beta thalassemia due to the familial mutation in children, siblings and parents of the parent is at maximum 50%.

Genetic counseling is recommended.

If you have any further questions please do not hesitate to contact us.

Yours sincerely,

Dr. med. Dr. rer. nat. Claudia Nevinny-Stickel-Hinzpeter
Human Geneticist

Dr. rer. nat. Florian Rieß
Certified Biologist

Jeannine Bek
MTLA