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Freiburg Medical Laboratory
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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: **No disease associated point mutation or deletion/duplication was detected in the *HBB* gene; beta-thalassemia not confirmed.**

Methods: DNA was isolated from peripheral blood and analysed by PCR and sequencing of the entire β-globin gene (*HBB*) and 5´and 3´untranslated regions (reference sequence: ENSEMBL ENST00000335295). Deletions of the β-, γ-, and δ-globin gene regions were analysed by the Multiplex Ligation-dependent Probe Amplification method (MLPA; Kit-No.: P102-C1, reference sequence NG_000007.3).

Interpretation: The molecular genetic analysis revealed no pathogenic point mutation or deletion/duplication in the *HBB* gene region. Furthermore deletions/duplications in the regulatory region upstream of the *HBB* gene were ruled out.

The clinical suspicion of beta-thalassemia could not be confirmed by the molecular genetic analysis.

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