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Dubai, UAE

Munich, 27.04.2016

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 alpha-thalassemia				

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

Results: **No deletion/duplication was detected in the α-globin-gene-region.**
No pathogenic point mutation was detected in the *HBA1*- and *HBA2*-gene.

Methods: After extraction of genomic DNA, screening for deletions/duplications was done by Multiplex Ligation-dependent Probe Amplification-Methode (MLPA-Methode Kit-Number: P140-B4, Reference sequence NG_00006.1). Analysis for point mutations was performed by PCR and sequencing of both α-globin-genes (*HBA1, HBA2*) including exon-intron-boundaries and 5´ and 3´untranslated regions (Reference sequence: ENSEMBL ENST00000320868 for *HBA1* and ENST00000251595 for *HBA2*).

Interpretation: No pathogeneic deletion/duplication and no pathogenic point mutation were detected in the α-globin genes *HBA1* and *HBA2*.

The clinical suspicion of α-Thalassemia in the patient is not supported by the molecular genetic analysis. Due to the complexity of the *HBA*-locus rare structural rearrangements cannot be excluded with ultimate certainty.

As a differential diagnosis a large deletion in the *HBB* gene region (γδβ-thalassemia) could also be considered. Please inform us if you wish further analysis to be performed.

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