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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 alpha-thalassemia				

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

Results: **Detection of the mutation c.358C>T (p.Pro120Ser) in the *HBA1* gene in heterozygous constellation; α-thalassemia confirmed.**
No deletion/duplication was detected in the α-globin genes (*HBA1, HBA2*).

Methods: Genomic DNA was isolated and mutation analysis was done by PCR-amplification and sequencing of the α-globin genes *HBA1* and *HBA2* including the 5'- and 3'-flanking regions and the exon-intron boundaries (reference sequences ENSEMBL ENST00000320868 for *HBA1,* ENST00000251595 for *HBA2).* Analysis of deletions and duplications in the α-globin gene region was performed by Multiplex Ligation-dependent Probe Amplification-Method (MLPA; Kit-No.: P140-C1, reference sequence NG_00006.1).

Interpretation: The molecular genetic analysis revealed the mutation c.358C>T (p.Pro120Ser) in the *HBA1* gene in heterozygous constellation. This mutation is already described in the literature as *Hb Groene Hart*. In heterozygous constellation it is associated with mild anemia and microcytosis.

The molecular genetic analysis confirms the clinical suspicion of α-thalassemia in your patient.

There is an increased probability for α-thalassemia for the patient's children.

No deletion or duplication was detected in the α-globin genes *HBA1* and *HBA2.*

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