Sample ID 5555

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Patient´s Jane Doe (female, dob 13.05.1991) DNA analysis for mutations in the **ABCA4 gene** associated with **Stargardt disease (STGD), CRD (cone-rod dystrophy) and RP19 (retinitis pigmentosa)** was performed.

Reason for referral: Confirmation of clinical diagnosis and mutation screening.
Patient´s clinical diagnosis and findings: Stargardt disease *in suspectu*. Patient has central vision loss from the age of 14, colour vision deficiency (red and green colours), general sight deterioration. Family history for Stargardt disease presence is negative.

Date of sample collection: 10.02.1010
Date of sample arrival: 17.02.2010
Date of test report: 23.02.2010
Specimen type: DNA sample extracted from periperal blood leucocytes
Analysis method: PCR and APEX (arrayed primer extension) method, mutation confirmation by sequencing
Tested gene: ABCA4
Number of mutations/SNPs tested: 558
Comments: sequencing was performed by Estonian Biocentre and Tartu University´s Institute of Molecular and Cell Biology, Riia 23b-304, Tartu 51010, Estonia

Result of the analysis:
Patient Jane Doe (female, dob 13.05.1991) has germline mutations p.G1961E, p.A1038V and p.L541P in ABCA4 gene in heterozygosis by APEX and sequencing analyses.

Biological interpretation:
The mutations p.G1961E and complex allele p.A1038V/p.L541P are missense mutations causing amino acid changes Glucine to Glutamic acid, Alanine to Valine and Leucine to Proline respectively in ABCR protein. Previously two patients with same phenotype are described by *Hartigai et al, 2005* in Hungarian STGD patients classified into phenotype group II according to Fishman et al, 1999. The patients presented with markedly thinned retina in the foveola and decreased macular volume and severe reduction of visual acuity was observed in relation with the duration of disease.
All detected mutations are reported to be present at high frequency in some populations: German population (*Rivera et al, 2000*), Hungarian population (*Hartigai et al, 20*05).

Clinical interpretation:
The molecular analysis of ABCA4 gene confirms the patient´s diagnosis of Stargardt disease. Jane Doe is compound heterozygote for p.G1961E, p.A1038V and p.L541P mutations.

The genetic councelling of the patient and her at-risk relatives is highly recommended.

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