KU Leuven

Report ID. 19020120001
Referal MD Michel
Date Friday, 19 Ap

Patient Information

Patient Name: **FN5 LN5**

Gender: **1**

Date of Birth: **06/03/2017 00:00:00**

Sample Information

Sequencing kit: **SureSelect Human All Exon V6 UTR**

Sample Type: **whole genome**

Sequencing Date: **07/08/2018 00:00:00**

Read Lenght: **150**

Coverage: **30**

Phenotype

Abnormal eye morphology
Abnormality of digit
Phenotypic abnormality
Abnormality of the eye
Abnormal retinal morphology
Abnormality of toe
Abnormality of limb bone morphology
Abnormality of the foot
Yellow/white lesions of the retina
Abnormal morphology of the posterior segment of th
Broad toe
Abnormality of limb bone
Abnormal fundus morphology
Abnormality of limbs

Abnormality of the nervous system
Abnormality of the skeletal system

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Classification	Gene	Inheritance	p-notation	c-notation	Zygosity
Uncertain significance	CLDN1	Autosomal recessive|	p.Gly49Gly	c.147G>A	heterozygous
Likely pathogenic	CYP21A2	Autosomal recessive|	p.Arg357Trp	c.1069C>T	heterozygous
Uncertain significance	CYP21A2	Autosomal recessive|	p.Arg357Trp	c.1069C>T	heterozygous
Uncertain significance	CYP21A2	null|	p.Arg357Trp	c.1069C>T	heterozygous
Likely benign	GBA	Autosomal dominant|	p.Asp448His	c.1342G>C	heterozygous
Likely pathogenic	GBA	Autosomal recessive|	p.Asp448His	c.1342G>C	heterozygous
Uncertain significance	GBA	Autosomal recessive|	p.Asp448His	c.1342G>C	heterozygous
Uncertain significance	GBA	Isolated cases|Multifact	p.Asp448His	c.1342G>C	heterozygous
Uncertain significance	GBA	null|	p.Asp448His	c.1342G>C	heterozygous
Likely benign	GDF5	Autosomal dominant|	p.Pro166His	c.497C>A	heterozygous
Likely benign	GDF5	Autosomal recessive|	p.Pro166His	c.497C>A	heterozygous

Uncertain significance	GDF5	Autosomal recessive|	p.Pro166His	c.497C>A	heterozygous
Likely benign	GDF5	Autosomal recessive|Au	p.Pro166His	c.497C>A	heterozygous
Uncertain significance	GDF5	null|	p.Pro166His	c.497C>A	heterozygous