STUDY OF MOLECULAR PATHOLOGIC MARKERS FOR HYPERTROPHIC CARDIOMYOPATHY

FINAL REPORT

Patient Name: NA	**Patient Birth Date**: NA
Medical reference: NA	**Patient Gender**: NA

Consultancy Referral Number: NA
Specimen Type: NA
Requisition Date: NA
Fulfillment Date: NA

Referring physician: NA,
Harvesting facility: NA
Referring facility: NA
Purpose: NA

Referral reason: NA

Sample Reference: NA

1 – Results

This genetic study shows 1 genetic variant in the gene *MYBPC3* that can be associated with hypertrophic cardiomyopathy.

1.1 – Genetic Information

The results, described according to HGVS nomenclature (http://www.hgvs.org), are presented in the following table.

Gene	Genetic variant references		Nucleotidic change	Aminoacidic change	Observation
	HGMD	Ensembl			
MYBPC3	CM981322	–	c.772G>A	p.Glu258Lys	Mutation in heterozygosity.

Hypertrophic cardiomyopathy is mainly caused by genetic alterations that deregulate the cardiac contraction mechanism comprising the dysfunction of the mechanical, biochemical and cell bioenergetics.

- ***MYBPC3*, CM981322**: The *MYBPC3* gene encodes for the cardiac myosin binding protein C, a sarcomeric motor protein important for cardiac muscle contraction. This mutation is pathogenic and associated with HCM. It abolishes the interaction between MYBP-C and myosin-S2 and disturbs muscle contraction kinetics [1]. It is associated with family history of HCM, early onset phenotype, moderate to severe hypertrophy, sudden cardiac death and cardiac transplant [2, 3, 4, 5, 6].

This information is supported by peer reviewed scientific papers indexed on PubMed (http://www.ncbi.nlm.nih.gov/pubmed) and also by The Human Gene Mutation Database (HGMD Professional ® version 2013.4) [7], accessed on 4^{th} November 2014.

Hypertrophic cardiomyopathy has a 50% chance of being transmitted to family relatives; therefore, we recommend a carrier testing for these genetic variants to direct members of the patient.

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Sample Reference: NA
Genetic Test: HCMMPMHG2

2 – Technical Information

2.1 – Methodology

1. A commercial kit was used to perform DNA extraction and purification from na. DNA concentration and quality were evaluated with MultiskanGo spectrophotometer (Thermo Scientific).
2. Genotyping was performed through molecular analysis of 218 genetic variants in 18 genes associated with hypertrophic cardiomyopathy.
3. Genotyping was achieved using a high-throughput DNA Microchip platform, the iPlex MassArray system from Agena. This array platform allows an optimal genetic analysis by combining the benefits of accurate primer extension chemistry with MALDI-TOF mass spectrometry. The different masses of each generated PCR product are then converted into genotype information.

2.2 – Test Accuracy

The technical accuracy of this test is estimated to be 99%.
HeartGenetics applies a rigorous quality control which may not exclude the possibility of error that might influence the test results. The results presented in this report are limited to the available scientific knowledge at the time this test was developed.

2.3 – HeartGenetics Panel

TCAP (NM_ 003673), ***ACTN2*** (NM_ 001103), ***TNNT2*** (NM_ 000364), ***MYH7*** (NM_ 000257), ***TNNC1*** (NM_ 003280), ***ACTC1*** (NM_ 005159), ***TNNI3*** (NM_ 000363), ***CRYAB*** (NM_ 001885), ***TPM1*** (NM_ 000366), ***LDB3*** (NM_ 001080116), ***FHL1*** (NG_ 015895.1), ***MYBPC3*** (NM_ 000256), ***BRAF*** (NM_ 004333), ***MYL2*** (NM_ 000432), ***CSRP3*** (NM_ 003476), ***FLNC*** (NM_ 001458), ***MYL3*** (NM_ 000258), and ***LAMP2*** (NM_ 002294).

Cantanhede, NA

Technical Direction

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3 – REFERENCES

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