Roadmap 2 Rare

Hypertrophic Cardiomyopathy Panel – Roadmap2Rare Diagnostics Program

As part of Sanofi Canada's sponsored Roadmap2Rare Diagnostic Program, a new **60-gene** hypertrophic cardiomyopathy (HCM) panel is available at **no charge** to facilitate differential diagnosis of various hypertrophic cardiomyopathies and their underlying disorders. This testing is made available in collaboration with PerkinElmer Genomics.

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This 60-gene panel will be available to patients with either:

- a clinical diagnosis of cardiomyopathy or cardiac arrythmia of unknown etiology, or
- a confirmed family history of hypertrophic cardiomyopathy of unknown etiology

What is hypertrophic cardiomyopathy (HCM)?

Hypertrophic cardiomyopathy (HCM) is defined by the presence of unexplained left ventricular hypertrophy (LVH) in the absence of other cardiac or systemic causes. It is frequently diagnosed by echocardiography and/or cardiac MRI.[1]

Differential diagnoses for HCM include secondary LVH and syndromic HCM, as seen in Fabry disease[1] and the infantile-onset form of Pompe disease,[2] where HCM is part of a constellation of phenotypic features.

For individuals with HCM and no other systemic involvement (non-syndromic HCM), pathogenic genetic mutations account for 20-30% of probands without family history of HCM, and 50-60% of probands with family history.[1]

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Genetic testing is recommended for individuals who meet the diagnostic criteria for HCM and to confirm diagnosis in individuals with clinical evidence suggestive of HCM.[1]

This next-generation sequencing (NGS) testing is offered at no-charge and with complimentary genetic consultation/ results interpretation for clinicians; genetic counselling for patients is also available. Clinicians can collect specimens with complimentary saliva swab kits or have PerkinElmer mail kits directly to patients for self-collection.

For more information about the Roadmap2Rare Diagnostic Program services, and to access the HCM panel, please visit PerkinElmer Genomics (www.perkinelmergenomics.com/roadmap2rare).

Patient and physician identifying information provided to PerkinElmer Genomics remains confidential and is not shared with Sanofi.

The HCM panel includes the following groups of disorders:

- Hypertrophic cardiomyopathy,
- Dilated cardiomyopathy,
- Myopathy,
- Myofibrillar myopathy,
- Noonan/CFC/Costello syndromes,
- Arrhythmogenic right ventricular dysplasia/cardiomyopathy,
- Restrictive cardiomyopathy

For information about Sanofi Canada's Roadmap2Rare initiatives, visit roadmap2rare.ca.

Genes included in the HCM Panel

GENE	GENE	GENE	GENE
A2ML1	ELAC2	MYH6	RAF1
ACADVL	FHL1	MYH7	RASA1
ACTC1	FLNC	MYL2	RIT1
ACTN2	GAA	MYL3	RRAS
AGL	GATA4	MYLK2	SHOC2
ANKRD1	GLA	MYOM1	SOS1
BAG3	HRAS	MYOZ2	SOS2
BRAF	JPH2	MYPN	SPRED1
CACNA1C	KRAS	NEXN	TCAP
CALR3	LAMP2	NF1	TNNC1
CAV3	LDB3	NRAS	TNNI3
CBL	MAP2K1	PDLIM3	TNNT2
CPT2	MAP2K2	PLN	TPM1
CSRP3	MTO1	PRKAG2	TTR
DES	MYBPC3	PTPN11	VCL

For more information about Sanofi Canada's Roadmap2Rare initiatives, and for the full list of genes and associated disorders, visit roadmap2rare.ca.

Or scan the QR code on the right.

References: 1. https://www.ncbi.nlm.nih.gov/books/NBK1768/.
2. van der Ploeg AT, Reuser AJ. Pompe's disease. ***Lancet.*** 2008 Oct 11;372(9646):1342-53. doi: 10.1016/S0140-6736(08)61555-X. PMID: 18929906."

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