

Muscle Disorders Panel – Roadmap2Rare Diagnostics Program



As part of Sanofi Canada's sponsored Roadmap2Rare Diagnostic Program, an expanded **122-gene** muscle disorders (MD) panel is available at **no charge** to facilitate differential diagnosis of various muscular and neuromuscular diseases, including Pompe disease. This testing is made available in collaboration with Revvity Omics (formerly PerkinElmer Genomics).



Roadmap *2 Rare*



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 Revvity Omics mail kits directly to patients for self-collection.

Genetic testing criteria

To be eligible for testing on the panel, patients need to show laboratory evidence suggestive of a muscle pathology (i.e. EMG, CK, muscle biopsy, MRI), in addition to at least one of the following:

- Muscle weakness, or
- Unexplained respiratory insufficiency, or
- Other symptom(s) suggesting muscle involvement (i.e. exercise intolerance, rhabdomyolysis, myalgia)



The Muscle Disorders Panel includes the following groups of disorders:

- Muscular dystrophies with predominant limb-girdle weakness patterns,
- Congenital muscular dystrophies,
- Rigid spine syndrome,
- Congenital myopathies,
- Inclusion myopathies,
- Congenital myasthenic syndromes,
- Scapuloperoneal syndromes,
- Metabolic myopathies

For more information about the Roadmap2Rare Diagnostic Program services, and to access the MD panel, please visit the Roadmap2Rare landing page on the Revvity Omics website. To be directed to this landing page, visit www.roadmap2rare.ca/laboratory.

Patient and physician identifying information provided to Revvity Omics remains confidential and is not shared with Sanofi.

Limb-girdle muscular dystrophy (LGMD) is a heterogeneous group of genetic disorders characterized by progressive muscle weakness involving the shoulder and pelvic girdles.¹

Pompe disease is a life-threatening, progressive neuromuscular disorder that causes progressive, irreversible muscle damage.²

From a similar Canadian panel, of all diagnosed patients, more than 36% of patients were diagnosed with an LGMD.³



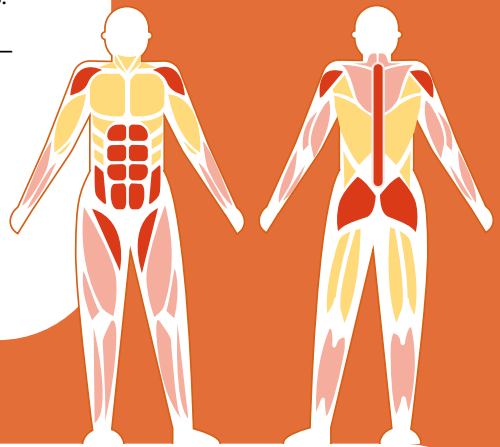
For information about Sanofi Canada's Roadmap2Rare initiatives, visit roadmap2rare.ca.

Proximal muscle weakness

Progressive proximal muscle weakness: the hallmark symptom of Pompe disease⁴

Progressive proximal muscle weakness affects the majority of late-onset Pompe patients.⁵

1. Progressive proximal muscle weakness – especially in lower limbs.^{4,5}
2. Respiratory insufficiency because of diaphragm weakness.⁶
3. Mild-to-moderate hyperckemia levels.⁶



● >80% of patients ● 50-80% of patients ● <50% of patients

Distribution of skeletal muscle weakness in 94 adults with Pompe disease. Adapted from van der Beek.⁵

Though symptoms vary, progressive proximal muscle weakness, with or without respiratory insufficiency or hyperckemia, should raise suspicion of Pompe disease.⁶

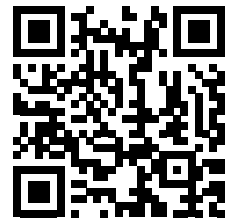
Genes included in the Muscle Disorders Panel

GENE	GENE	GENE	GENE
ACTA1	DES	LMOD3	SCN4A
ACTN2	DMD	LRP4	SELENON
AGRN	DNAJB6	MEGF10	SGCA
ANO5	DNM2	MTM1	SGCB
ATP2A1	DOK7	MUSK	SGCD
B3GALNT2	DPAGT1	MYBPC1	SGCG
B4GAT1	DPM3	MYF6	SLC18A3
BAG3	DYSF	MYH2	SLC25A1
BIN1	EMD	MYH7	SLC5A7
BVES	FHL1	MYO18B	SPEG
CACNA1S	FKBP14	MYOT	STAC3
CAPN3	FKRP	MYPN	STIM1
CASQ1	FKTN	NEB	SYNE1
CAV3	FLNC	ORA11	SYNE2
CCDC78	GAA	PABPN1	SYT2
CFL2	GFPT1	PAX7	TCAP
CHAT	GMPPB	PFKM	TMEM43
CHKB	GNE	PHKA1	TNNT1
CHRNA1	HNRNPDL	PLEC	TNPO3
CHRNB1	INPP5K	PNPLA2	TOR1AIP1
CHRND	ISPD/CRPPA	POGLUT1	TPM2
CHRNE	ITGA7	POMGNT1	TPM3
COL12A1	KBTBD13	POMGNT2	TRAPPC11
COL13A1	KLHL40	POMK	TRIM32
COL6A1	KLHL41	POMT1	TRIP4
COL6A2	KY	POMT2	TTN
COL6A3	LAMA2	PYGM	VAMPI
COLQ	LAMP2	RAPSN	VCP
CPT2	LARGE1	RBCK1	VMA21
CRYAB	LDB3	RXYLT1/TMEM5	
DAG1	LMNA	RYR1	



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.** Limb-Girdle Muscular Dystrophy. Muscular Dystrophy Canada website. <https://muscle.ca/discovermd/types-of-neuromuscular-disorders>. Accessed Jan 20, 2020. **2.** Hagemans MLC et al. *Neurology*. 2005;64(12):2139-2141. **3.** Thuriot et al. *Neurol Genet*. 2020;6(2):e408. **4.** Hirschhorn R et al. Glycogen storage disease type II: acid alpha-glucosidase (acid maltase) deficiency. In: Scriver CR et al, eds. *The Metabolic & Molecular Bases of Inherited Disease*. 8th ed. New York, NY: McGraw-Hill; 2001:3389-3420. **5.** van der Beek N et al. *Orphanet J Rare Dis*. 2012;7:88. **6.** American Association of Neuromuscular & Electrodiagnostic Medicine. *Muscle Nerve*. 2009;40(1):149-160.