MAYO CLINIC LABORATORIES

MDYSP

Inherited Muscular Dystrophy Gene Panel, Varies

Patient ID	Patient Name		Birth Date	Sex	Age
1006232455	BABEKOV, EDWARD		1974-10-12	M	50
Order Number	**Client Order Number**	**Ordering Physician**	**Report Notes**		
M198147368	62508601356	MAGDA,PAUL			
Account Information		**Collected**			
C7029975 NewYork-Presbyterian/Queens Hospital		27 Mar 2025 08:11			

Muscular Dystrophy Gene Panel

Interpretation MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Result Summary MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Result MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Test Description MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Specimen MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Source MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Additional Results MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Resources MCR

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Additional Information MCR

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Method MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Genes Analyzed MCR

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Disclaimer MCR

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Released By MCR

Muscular Dystrophy Gene Panel was cancelled on 04/16/2025 at 10:07; Incorrect test was ordered.

Received: 29 Mar 2025 08:42 **Reported:** 16 Apr 2025 10:07

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	Nikola Baumann Ph.D.	24D0404292

Athena Diagnostics

OPMD Repeat Expansion Test

athena diagnostics®

Testing that Makes a Difference.

Patient	Requesting Physician	Accession Number
BABEKOV, EDWARD	PAUL MAGDA	25029660
Date of Birth / Sex: 10/12/1974 / M	Report to: MAYO CLINIC LABORATORIES	Family Number/Kindred Number:
Specimen Type: DNA	Address: P.O. BOX 4100	Patient Number: L027059285
Test Category: Diagnostic (Symptomatic)	ROCHESTER, MN 55903-4100	Specimen Collection Date: 03/27/2025
Test Requested: OPMD Repeat Expansion Test		Date Received: 04/17/2025
	Additional Reports to:	Report Date: 04/25/2025

OPMD Repeat Expansion Test

POSITIVE

This test identified a pathogenic repeat expansion in the *PABPN1* gene.

INTERPRETIVE RESULTS TABLE

	Gene/Test	Technical Result	Variant Type	Inheritance	Clinical Relevance	Pub Med ID
Positive	**PABPN1**	**13 and 10 repeats**	**Repeat Expansion**	**Autosomal Dominant**	**Pathogenic**	

OPMD Repeat Range: Normal (<=10), Carrier (11), Positive (>=12)

Comments: This test result is consistent with a diagnosis of, or a predisposition to develop, oculopharyngeal muscular dystrophy associated with *PABPN1* repeat expansions.

Recommendations: This individual's family members are at risk for possessing or inheriting this variant. Careful reconciliation of this molecular data with this individual's clinical presentation, family history, and other laboratory results, in conjunction with genetic counseling, is highly recommended.

Health care providers, please contact the Athena Diagnostics Client Services Department at 1-800-394-4493 if you wish to consult with a Laboratory Director or a Genetic Counselor regarding this test result.

Background Information: Oculopharyngeal muscular dystrophy (OPMD) is an autosomal dominant, and more rarely autosomal recessive, primary muscle disorder characterized by late-onset involvement of small distinct muscles of the eyelid and muscles of the pharynx which, respectively, cause ptosis and dysphagia. Symptoms usually manifest around the fifth decade of life with slow disease progression. In rare instances involvement of the peripheral nervous system has been described. Reported prevalence of the disorder ranges from 1:600 to 1:100,000 based on geographical locations of populations studied. Autosomal recessive inheritance is rare and, compared to dominantly inherited OPMD, occurs with a later onset and milder progression.

The *PABPN1* gene encodes an abundant nuclear protein, PABP2, which binds with high affinity to mRNA poly(A) tails to regulate gene expression. The protein plays a direct role in the export of mRNAs from the nucleus to the cytoplasm. Triplet repeat expansion of a polyalanine domain within *PABPN1* is thought to lead to inefficient degradation of PABP2 protein with concomitant abnormal protein aggregation. Subsequent interference with normal cellular processes results in OPMD clinical symptoms.

PABPN1 gene information: MIM ID: *602279; Chromosome Location: 14q11.2 ; NCBI Reference Sequence: NM_004643.3; In the cDNA, the initiator codon, ATG (methionine), is designated as codon number 1 and the "A" is designated as nucleotide +1. The initiator codon is located in exon 1 and all subsequent numbering is sequential.

Phenotype information: MIM ID: #164300 for Oculopharyngeal muscular dystrophy.

Methods:
Direct testing for the common OPMD repeat expansion (an expansion of the GCG repeat in the *PABPN1* gene) was performed by PCR amplification of the repeat region from genomic DNA extracted from the individual's whole blood specimen, followed by high-resolution electrophoresis to determine the number of tandem repeats in each allele. This methodology approaches 100% accuracy for the detection of repeat expansions in this gene. Clinical studies report that this diagnostic technology has greater than 99% sensitivity and specificity for detection of OPMD repeat expansions.

Name: BABEKOV, EDWARD **DOB: 10/12/1974** **Accession: 25029660**

OPMD Repeat Expansion Test

athena diagnostics

OPMD Repeat Expansion Test

Limitations of analysis: The accuracy of repeat number determination is within ± 1 for repeat expansions in this gene. Although rare, false negative results may occur due to allele dropout. All results should be interpreted in the context of clinical findings, relevant history, and other laboratory data.

Background References

1. Brunet, G, et al. (1997) Neuromuscul Disord 7 Suppl 1: S34-7. (PMID: 9392013)
2. Blumen, SC, et al. (1997) Neuromuscul Disord 7 Suppl 1: S38-40. (PMID: 9392014)
3. Brais, B. (2003) Cytogenet Genome Res 100: 252-60. (PMID: 14526187)
4. Dubbioso, R, et al. (2012) J Neurol 259: 833-7. (PMID: 21956377)
5. Schober, R, et al. (2001) Neuropathology 21: 45-52. (PMID: 11304042)
6. Brais, B, et al. (1998) Nat Genet 18: 164-7. (PMID: 9462747)
7. Calado, A, et al. (2000) Hum Mol Genet 9: 2321-8. (PMID: 11001936)

This test was developed and its analytical performance characteristics have been determined by Athena Diagnostics. It has not been cleared or approved by the U.S. Food and Drug Administration. This assay has been validated pursuant to the CLIA regulations and is used for clinical purposes.

Laboratory results and submitted clinical information reviewed by,

Zhenyuan Wang, PhD, FACMG

Senior Director, Genetics

Laboratory oversight provided by Vivekananda Datta, M.D., Ph.D., CLIA license holder, Athena Diagnostics (CLIA# 22D0069726)