ClinGenLab
Clinical Testing Laboratory, Inc.

Clinical Testing Lab of Washington
2150 Pennsylvania Avenue NW
Washington, DC 20037
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Specimen Number	Specimen Type	Control Number	Account Number	Account Phone Number	Route
	Peripheral Blood				

Patient Last Name	Patient First Name	Patient Middle Name
	Alexis & Noah	

Patient SS#	Patient Phone	Total Volume

Age (Y/M/D)	Date of Birth	Sex	Fasting
12 y.o & 12 y.o		Female & Male	

Patient Address

Both patients have identical genetic variant analysis results, therefore we are reporting a single result for both.

Please note: This would NEVER happen in real life!

Patient Barcode

Indication: Sagawa (TH Deficiency) or SPR Deficiency
Family History: No known family history
Ethnicity: Western European Caucasian

Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID
			Jane Ferreiro, MD		

Dystonia Comprehensive Panel — Tests Ordered

General Comments
Please send a copy of the final report to the Molecular Science/M1 Training office via Fax at (202) 555-1212

Clinical test results for Dystonia Comprehensive Panel

GENE	RESULTS	EXPLANATION
SPR (2p13.2)	Pathogenic variant detected: Arg150 Arg150Gly	**This result supports the diagnosis of Sepiapterin reductase deficiency**. This result should be interpreted in the context of clinical presentation and results of other laboratory tests (e.g., 5HIAA, 5HVA, BH4, Neopterin, etc.). A sequencing study with PCR validation has identified one copy of this reported pathogenic variation: Arg150Gly (SPR: g.6075A>G, c.448A>G, p.Arg150Gly) variation The Arg150Gly variation is an A to G change at nucleotide position 6075 in the SPR gene. This encodes an alternate residue at position 150 from a large, positively-charged, polar amino acid to one with a small, neutral side chain.
SPR (2p13.2)	Pathogenic variants detected: Lys251 Lys251Ter	**This result supports the diagnosis of Sepiapterin reductase deficiency**. This result should be interpreted in the context of clinical presentation and results of other laboratory tests (e.g., 5HIAA, 5HVA, BH4, Neopterin, etc.). A sequencing study with PCR validation has identified one copy of this reported pathogenic variation: Lys251Ter (SPR: g.9120A>T, c.751A>T, p.Lys251Ter) variation. The Lys251Ter variation is an A to T change at nucleotide position 9120 in the SPR gene. This forms a premature stop codon at amino acid position 251 resulting in an abnormally short or truncated protein.

No genetic variants were detected in:			
	ANO3 (11p14.3-14.2)	HPCA (1p35.1)	SLC2A1 (1p34.2)
	ATP1A3 (19q13.2)	KCTD17 (22q12.3)	SLC6A3 (5p15.33)
	CIZ1 (9q34.11)	PNKD (2q35)	TH (11p15.5)
	DRD2 (11q23.2)	PRKRA (2q31.2)	THAP1 (8p11.21)
	GCH1 (14q22.2)	PRRT2 (16p11.2)	TOR1A (9q34.11)
	GNAL (18p11.21)	SGCE (7q21.3)	TOR1AIP1 (1q25.2)
			TUBB4A (19p13.3)

DISCLAIMER:
Test results should be interpreted in context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete. Rare polymorphisms exist that could lead to false negative or positive results. If results obtained do not match the clinical findings, additional testing should be considered.

CLINICAL DESCRIPTION

Dystonia is a movement disorder characterized by sustained or intermittent muscle contractions causing abnormal, often repetitive movements and/or postures. Dystonic movements are typically patterned and twisting, and may be associated with tremor. Dystonia is often initiated or worsened by voluntary action and associated with overflow muscle activation. Dystonia can be classified clinically according to age of onset, body distribution, temporal pattern, and associated features (i.e., isolated dystonia – in which it is the only motor feature except tremor; combined dystonia – in which another movement disorder is present; or complex dystonia – in which other neurologic or systemic manifestations are present).

Conditions tested:

CONDITION(S)/PHENOTYPE(S)	ALSO KNOWN AS	GENE(S) TESTED
Dystonia		All listed below and: HPCA (1p35.1) KCTD17 (22q12.3) PARK2 (6q26) PNKD (2q35) SLC2A1 (1p34.2) TOR1AIP1 (1q25.2)
Autosomal dominant torsion dystonia 4		TUBB4A (19p13.3)
Dystonia 1	Dystonia 1, modifier of Early-Onset Primary Dystonia (DYT1)	TOR1A (9q34.11)
Dystonia 10	EPISODIC KINESIGENIC DYSKINESIA 1, Familial Paroxysmal Kinesigenic Dyskinesia	PRRT2 (16p11.2)
Dystonia 12	Rapid-Onset Dystonia-Parkinsonism	ATP1A3 (19q13.2)
Dystonia 16		PRKRA (2q31.2)
Dystonia 23		CIZ1 (9q34.11)
Dystonia 24		ANO3 (11p14.3-14.2)
Dystonia 25		GNAL (18p11.21)
Dystonia 5, Dopa-responsive type	DYSTONIA, DOPA-RESPONSIVE, GTP Cyclohydrolase 1-Deficient Dopa-Responsive Dystonia	GCH1 (14q22.2)
Dystonia 6, torsion		THAP1 (8p11.21)
Infantile Parkinsonism-dystonia	DOPAMINE TRANSPORTER DEFICIENCY SYNDROME	SLC6A3 (5p15.33)
Myoclonic dystonia	DYSTONIA 11, MYOCLONIC	DRD2 (11q23.2) SGCE (7q21.3)
Segawa syndrome, autosomal recessive	Tyrosine Hydroxylase Deficiency, Tyrosine Hydroxylase-Deficient Dopa-Responsive Dystonia	TH (11p15.5)
Sepiapterin reductase deficiency	Dopa-Responsive Dystonia Due to Sepiapterin Reductase Deficiency	SPR (2p13.2)

METHODOLOGY

Full gene sequencing and deletion/duplication analysis of targeted gene coding regions are performed using Next-Generation (NGS)/Massively Parallel Sequencing (MPS). All pathogenic variants and deletions/duplications are confirmed using orthogonal technologies.

PERFORMANCE

Our analytic validation study has demonstrated >99.9% sensitivity and specificity for tested mutations.