



Specimen Number	Patient ID	Control Number	Account Number	Account Phone Number	Route
Patient Last Name		Both patients have identical genetic variant analysis results, therefore we are reporting a single result for both. <i>Please note: This would NEVER happen in real life!</i>			
Patient First Name	Patient Middle Name				
Patient SS#	Patient Phone	Total Volume	 		
Age (Y/M/D)	Date of Birth	Sex			
Patient Address			Additional Information Peripheral Blood and CSF Samples: Alexis – 12 y.o. Female & Noah – 12 y.o. Male		
Date and Time Collected	Date Entered	Date and Time Reported	Physician Name	NPI	Physician ID

Tests Ordered
CBC, Platelet Ct, and Diff ; Select Pterin/Neurotransmitter Basic Panel (Neopterin, BH4, 5HIAA, HVA)
General Comments

TEST RESULTS SHOWN ON BACK

PRELIMINARY INTERPRETATION:

Alexis 12 y.o. Female:
 A preliminary diagnosis of Sepiapterin Reductase Deficiency Syndrome can be made based on normal levels of Neopterin and very low levels of BH4 and both Serotonin (5HIAA) and Dopamine (HVA) metabolites.
 Dopa-responsive Dystonia, type 5 (due to a deficiency in GTP cyclohydrolase 1) is unlikely due to normal levels of Neopterin. Segawa Dystonia (due to a deficiency in Tyrosine hydroxylase) is unlikely due to the extremely low levels of both Dopamine (HVA) and Serotonin (5HIAA) pathway metabolites. Other dystonia-like syndromes (Juvenile Parkinson disease and Cerebral Palsy with global delays) can be ruled out due to the positive results of these lab findings.

Noah 12 y.o. Male:
 A preliminary diagnosis of Sepiapterin Reductase Deficiency Syndrome can be made based on normal levels of Neopterin and very low levels of BH4 and both Serotonin (5HIAA) and Dopamine (HVA) metabolites.
 Dopa-responsive Dystonia, type 5 (due to a deficiency in GTP cyclohydrolase 1) is unlikely due to normal levels of Neopterin. Segawa Dystonia (due to a deficiency in Tyrosine hydroxylase) is unlikely due to the extremely low levels of both Dopamine (HVA) and Serotonin (5HIAA) pathway metabolites. Other dystonia-like syndromes (Juvenile Parkinson disease and Cerebral Palsy with global delays) can be ruled out due to the positive results of these lab findings.

PLEASE NOTE: There are well established pathogenic genetic variants that are strongly correlated with the various Dystonia Syndromes, including Sepiapterin Reductase Deficiency. The presence of a hereditary disorder has consequences for family members. Genetic testing and counseling should be considered by the patient and family members.

Peripheral Blood TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
Alexis - 12 y.o. Female					Noah - 12 y.o. Male				
CBC, Platelet Ct, and Diff									
Hematocrit	30.6			30.9 - 37.0	34.8		%	30.9 - 37.0	07
Hemoglobin	10.1			10.3 - 12.4	11.7		g/dL	10.3 - 12.4	07
Red Blood Cell Count	4.70			4.10 - 5.00	4.85		x10E6/uL	4.10 - 5.00	07
White Blood Cell Count	14.3			6.2 - 14.5	12.2		x10E3/uL	6.2 - 14.5	07
RDW	nd			N/A	nd		%	N/A	07
MCV	71.2			70.5 - 81.2	73.7		fL	70.5 - 81.2	07
MCH	14.5			23.2 - 27.5	26.2		pg	23.2 - 27.5	07
MCHC	32.2			31.9 - 35.0	34.1		g/dL	31.9 - 35.0	07
Imm.Granulocytes (Absolute)	4.9			1.6 - 8.3	7.2		x10E3uL	1.6 - 8.3	07
Granulocytes (Percent)	29.4			21.3 - 66.7	45.3		%	21.3 - 66.7	07
Eosinophils (Absolute)	nd		x10E3uL	NA	nd		x10E3uL	NA	07
Eosinophils (Percent)	nd		%	0.0 - 3.3	nd		%	0.0 - 3.3	07
Basophil (Absolute)	nd		x10E3uL	NA	nd		x10E3uL	NA	07
Basophil (Percent)	nd		%	0 - 2	nd		%	0 - 2	07
Monocytes (Absolute)	nd		x10E3uL	N/A	nd		x10E3uL	N/A	07
Monocytes (Percent)	nd		%	5 - 11	nd		%	5 - 11	07
Lymphocytes (Absolute)	5.8		x10E3uL	1.9 - 6.8	4.7		x10E3uL	1.9 - 6.8	07
Lymphocytes (Percent)	61			20 - 64	52		%	20 - 64	07
Platelets (Absolute)	276			219 - 452	308		x10E3uL	219 - 452	07

CSF TESTS	RESULT	FLAG	UNITS	REFERENCE INTERVAL	RESULT	FLAG	UNITS	REFERENCE INTERVAL	LAB
Alexis - 6 y.o. Female					Noah - 6 m.o. Male				
Selected Pterin/Neuro Basic									
Neopterin	13		nmol/L	7-40	38		nmol/L	7-40	07
Tetrahydrobiopterin (BH4)	0.8	Very low	nmol/L	9-40	1.2	Very low	nmol/L	9-40	07
5HIAA (Serotonin metabolite)	6	Very low	nmol/L	88-278	8	Very low	nmol/L	88-278	07
HVA (Dopamine metabolite)	28	Very low	nmol/L	200-800	31	Very low	nmol/L	200-800	07