



Patient Information	
Patient Name ALEXIS & NOAH	Patient Barcode Sticker 
DOB, Medical Record Number (MRN) <div style="background-color: #ccc; width: 100px; height: 15px; margin-bottom: 5px;"></div> <div style="background-color: #ccc; width: 100px; height: 15px;"></div>	
Requesting Provider	
Assigned Provider/Practice Name: Jane FERREIRO, MD / MyClinicalService	Specialty/Department: Pediatrics/Neurology
Address: 900 23rd St NW Washington, DC 20037	Phone: (202) 555-1212 Facsimile #: (202) 555-1212
Consultant Provider	
Provider's Name: to be assigned	Specialty/Department: Molecular Science/M1 Training
Address: 2300 I St NW, Suite 201 Washington, DC 20052	Phone: (202) 555-1212 Facsimile #: (202) 555-1212
Referral Information	
Authorization No:	Authorization Type:
Reason for Referral: Evaluation of Sepiapterin Reductase Deficiency	
Diagnosis: G24.1 – Sepiapterin Reductase Deficiency	
<p>Clinical Notes: 12 year old twin boy and girl, initially diagnosed at 24 months with Cerebral Palsy due to chief complaints of hypotonia, dystonia and spasticity and the results of a brain imaging study on the male, then diagnosed at 5 years old with Dopa-responsive Dystonia (Sagawa Disease) and treated fairly successfully with L-Dopa. However, symptoms (sleep disturbances, mood disorders, drooling) were still present and at 11 years old, Alexis developed serious respiratory complications. A preliminary diagnosis of Sepiapterin Reductase Deficiency Syndrome was made based on normal levels of Neopterin and very low levels of BH4 and both Serotonin (5HIAA) and Dopamine (HVA) metabolites.</p> <p>Whole genome sequencing was performed at the request of the family and will be analyzed for pathogenic variants that exist in both twins. The final report will be faxed to the Molecular Science/M1 Training program for evaluation.</p> <p>Please consult with the family and send a copy of the final report back to this office. Thanks.</p>	
Procedures: Variant Interpretation – Molecular Impact Characterization	
Visits Allowed: 3	
Unit Type: V (VISIT)	
Referral is Valid Until: 09/30/2018	
<p>Notes: Patient must arrive 30 minutes early, with a picture ID, Insurance card and have a copy of this referral. If the referred patient is a minor and anyone other than the child's parents are escorting the child to the appointment, a letter of consent by the parent is needed. Please bring a list of medications the patient is taking with you to this appointment (including over the counter).</p>	
Please send the final report by Fax to: (202) 555-1212	
Signature: 	
Ferreiro, Jane, MD on 08/29/2018 at 8:26 AM EDT	