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| **Time of Appointment:** 3:00 – 3:30 PM | New Patient Established Patient |

**Patient Details**

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| **Name:** Hayes, Simon | **Age:** 7 y/o | **Biological Sex:** Male | **Gender:** Male |

**Indication Summary:**  7yo old follow-up patient who originally presented at 3 years of age with gross motor delays, speech delays, and a history of FTT/SGA. During the original visit he was noted to have low-set ears, facial hypotnia, and a recent CT scan noted ventriculomegaly secondary to craniosynostosis. Recent, around age 6, he was noted to have his first seizure. A recurrence took place 6 months later, and the seizures have since been controlled through medication. Neurology referred patient back to genetics due to the recent onset of new symptoms.

* **Relevant Family Hx:** No family history of craniosynostosis. Patient has 2 brothers who are both in good health. Patient’s mother has severe allergies and asthma. Patient’s father has a history of high cholesterol. Paternal grandfather diagnosed with prostate cancer at 71 y/o. Family history otherwise unremarkable for developmental delay, intellectual disabilities, stillbirth, multiple miscarriages, sudden death, birth defects, or known genetic conditions.  No consanguinity reported.

**Summary of recent encounters and results:**

* Head CT Scan – 10/03/2019
  + Mild ventriculomegaly
  + Poor ossification of metopic sutures
* Previously met with Pediatric Genetics (2019)
  + Chromosomal microarray-normal results
  + Fragile X: 30 repeats (within normal range)
  + Karyotype: normal results
* Neurology
  + Followed annually for ventriculomegaly
  + No significant change in cranosynostosis
  + Seizures controlled
* Genetics
  + Seen every two years
  + Patient was stable in presentation at age 5
  + Family elected to manage current symptoms and hold on further testing

**Differential Diagnosis:**

* Single gene condition

Intellectual Disability-related disorders

X-linked disorders

Disorders with Intellectual disability and seizures

**Appointment plan:**

* Assess how patient and family have been doing since last visit
* Collect any updates to the family and medical history
* Physical exam 🡪 Establish testing plan
* Informed consent process
  + Testing plan, benefits, and limitations
  + Explain how this test is different than microarray
  + Facilitate decision-making
* Psychosocial check-in