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| **Time of Appointment:** 3:30 – 4:30 PM | New Patient Established Patient |

**Patient Details**

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

**Referral/comments:**

* **Indication Summary:** Patient underwent whole exome sequencing at last genetics visit. Patient and family return today to discuss results. Results provided via phone, family returns to learn more about the results
* **Relevant Family Hx:** No significant changes in family or medical history.

**Current studies:**

* Whole exome sequencing
  + Heterozygous pathogenic variant in *ASXL3* gene c.3106C>T, p.Arg1036Ter
    - Consistent with diagnosis of Bainbridge-Ropers syndrome
    - Finding was de novo in patient

**Appointment plan:**

* Results disclosure
* Educational Counseling:
  + Natural history of Bainbridge-Ropers Syndrome, management plan, recurrence risk
* Psychosocial Counseling
* Discuss/provide resources for Bainbridge-Ropers Syndrome education, support, and community involvement