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| **Time of Appointment:** 10:00 – 10:30 AM | New Patient Established Patient |

**Patient Details**

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| **Name:** Hawkins, Christine | **Age:** 16 y/o | **Biological Sex:** Female | **Gender:** Female |

**Referral/comments:**

* **Indication Summary:** Patient with >100 café-au-lait spots, skin-fold freckling, multiple dermal and subcutaneous neurofibromas scattered along the body. Noted to have tumor in nostril at 9 mo., since removed. Previously diagnosed with a speech delay and intellectual disability. A clinical diagnosis of neurofibromatosis type 1 was given at age 4, but the family would like to obtain genetic confirmation during today’s visit.. NF1 testing ordered, results available.
* **Relevant Family History:** No family history suggestive of NF1. Patient’s mother is in good health. Father previously had kidney stones. Maternal grandmother has high blood pressure. Paternal grandmother reported to have arthritis. Family history unremarkable for developmental delays, stillbirths, multiple miscarriages, sudden deaths, birth defects, or known genetic conditions. No consanguinity reported.

**Prior studies:**

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|  | **Study** | **Results/Findings** |
| **Undisclosed** | UAB Comprehensive Analysis Neurofibromatosis Type 1 | Pathogenic variant-Type 1 NF1 microdeletion |

**Appointment plan:**

* Review the test result
* Educational Counseling:
  + NF1 natural history, management plan, recurrence risk
* Psychosocial Counseling
* Discuss/provide resources for NF1 education, support, and community involvement