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| **Time of Appointment:** 1:00 – 1:30 PM | 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.
* **Relevant Family History:** Family history has not been formally collected

**Summary of recent encounters:**

* Plastic Surgery at age 9mo for tumor removal: pathology diagnosed as neurofibroma
* Ophthalmology: seen annually to look for lisch nodules
* Weekly visits with speech therapy
* Neurology: seen last week -Brain MRI (normal)
* Dermatology: seen last month, no neurofibromas requiring additional biopsy

**Prior studies:**

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| --- | --- | --- |
|  | **Study** | **Results/Findings** |
| **Disclosed** | Brain MRI | No significant findings |
| Dermatology Exam | >100 café-au-lait spots, several >15mm; axillary and inguinal freckling; multiple neurofibromas (dermal and subcutaneous) |
| **Undisclosed** | N/A | |

**Differential Diagnosis:**

* Neurofibromatosis 1 (NF1)

**Appointment plan:**

* Assess patient/family’s current understanding of the reason for referral
* Collect family and medical history
* Physical exam à Establish testing plan
* Educational Counseling:
  + Genes and chromosomes, some genetic differences cause health concerns, overview of NF1
* Informed consent process
  + Testing plan, benefits, and limitations
    - NF1-comprehensive analysis
  + Facilitate decision-making
* Psychosocial check-in