

MEDICAL GENOMICS LABORATORY: NF2 & SCHWANNOMATOSIS PHENOTYPIC CHECKLIST FORM



Patient ID: _____

Referring Physician: _____ Date of Exam ___/___/___

DEMOGRAPHIC INFORMATION

Gender: Male Female Date of Birth: ___/___/___

Ethnicity: Mother: White Black Native American Hispanic Asian Other:
Father: White Black Native American Hispanic Asian Other:

DIAGNOSIS

Does the patient have a clinical diagnosis of NF2? Yes No If Yes, age at diagnosis: _____

Family history: Sporadic Familial Unknown

SIGNS AND SYMPTOMS

Ear: Absent Unknown Present: Bilateral Deafness Unilateral Deafness Age of symptoms: _____
 Balance Dysfunction Tinnitus
 Audiometric Abnormality, please describe: _____
 Other, please describe: _____

Eyes Absent Unknown Age of Symptoms: _____
 Present: Blindness
 Lenticular opacities
 Lisch nodules
 Left Right
 Bilateral Unknown

Schwannomas

Vestibular schwannomas Age of Symptoms: _____
 Bilateral
 Unilateral
 No evidence of vestibular nerve tumor by MRI (Age: ___ yrs)
 Lack of symptoms of vestibular nerve tumor, but no MRI done (Age: ___ yrs)
 Unknown

Spinal schwannomas Age of Symptoms: _____
 Present
 No evidence by MRI (Age: ___ yrs)
 Lack of symptoms, but no MRI done (Age: ___ yrs)
 Unknown
Provide location of spinal tumors: C ___ to C ___, T ___ to T ___, L ___ to L ___

Other schwannomas Age of Symptoms: _____
 Absent Unknown
 Head Neck Trunk L Arm L Hand L Leg L Foot
 Abdomen Pelvis Genital area R Arm R Hand R Leg R Foot

Present only in an anatomically limited distribution(single limb or segment of the spine): yes or no

Result *SMARCB1*-staining on the tumor specimen: Abnormal (no *SMARCB1*-staining)
 Normal (*SMARCB1* staining)
 Not performed



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Meningiomas

- Present, Location: _____
- No evidence by MRI (Age: ___yrs)
- Unknown

Age of Symptoms: _____

Other spinal tumors

- Absent by MRI
 - Present, asymptomatic
 - Present, symptomatic
 - Unknown
- Pathology Known: Yes, please specify: _____ No

Age of Symptoms: _____

Provide location of spinal tumors: C ___ to C ___, T ___ to T ___, L ___ to L ___

Cranial nerve involvement

- Present, Location: _____
- No evidence by MRI (Age: ___yrs)
- Unknown

Age of Symptoms: _____

Skin

- CAL spots**
- 1
 - 2-3
 - 4-5
 - >5-10
 - >10

Age of Symptoms: _____

- Neurofibromas**
- 1-5
 - ≥6-99
 - ≥100

Age of Symptoms: _____

- Skin fold freckling**
- | | | |
|------------|--------------------------|--------------------------|
| | Left | Right |
| Groin | <input type="checkbox"/> | <input type="checkbox"/> |
| Axilla | <input type="checkbox"/> | <input type="checkbox"/> |
| Submammary | <input type="checkbox"/> | <input type="checkbox"/> |

Age of Symptoms: _____

Please indicate location of spinal tumors (if present)

Additional comments/remarks:

