

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform Neurofibromatosis Type I/Legius Syndrome testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR NEUROFIBROMATOSIS TYPE 1/ LEGIUS SYNDROME

Patient's Name _____ Date of Birth ____/____/____ Gender F M

Physician _____ Physician's Phone (____) _____ Practice Specialty _____

Genetic Counselor _____ Counselor's Phone (____) _____

Patient's Ethnicity (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African-American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

Does the patient have SYMPTOMS? No Yes (check all that apply)

- Café au lait macules (circle number) 1 2 3 4 5 6 7+
- Specific osseous lesions such as tibial pseudarthrosis or sphenoid dysplasia; describe _____
- Overgrowth, describe _____
- Malignant peripheral nerve sheath tumor (MPNsT)
- Axillary or inguinal freckling
- Learning disabilities
- Dermal fibromas
- Vertebral dysplasia
- Optic glioma (age at diagnosis _____)
- Scoliosis
- Lisch nodules (iris hamartomas)
- Other _____

Does the patient have a FAMILY HISTORY of: NF1 LEGIUS Neither Unknown

If yes, attach a PEDIGREE or specify the RELATIONSHIP of the family members(s) to the patient and detail the symptoms/age of onset in each symptomatic relative.

Has DNA testing been performed for these family member(s)? No Yes Unknown

If yes, attach a copy of the relative's DNA laboratory result.

Has the patient undergone previous DNA testing for this disorder? No Yes Unknown

If yes, please describe test(s) and results _____

Circle the test below you intend to order.

2001952 Neurofibromatosis Type 1 (NF1) Deletion/Duplication

Six percent of NF1 is caused by large *NF1* gene/locus deletions.. Order to determine if a patient with a suspected diagnosis of neurofibromatosis type 1 has a causative large NF1 gene deletion/duplication.

Legius Syndrome (SPRED1) Sequencing and (NF1) Sequencing Exon 17

Eight percent of individuals with only café-au-lait spots and/or axillary freckling and no neurofibromas have a *SPRED1* mutation.

2001961 Familial Mutation, Targeted Sequencing

Tests for a *RET* sequence change previously identified in a family member. A copy of relative's DNA laboratory result is REQUIRED.

For questions, contact a genetic counselor at 800-242-2787 ext. 3922

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