MEDICAL GENOMICS LABORATORY: *NF1/SPRED1* 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

NIH criteria:
☐ >6 CAL spots >5mm, postpubertal >15mm
☐ >2 neurofibromas or 1 plexiform NF
☐ Axillary or inguinal freckling
☐ First degree relative diagnosed with NF1 by above criteria
☐ Optic glioma
☐ >2 Lisch nodules
☐ A distinct osseous lesion
Does patient fulfill NIH diagnostic criteria for NF1? ☐ Yes ☐ No

Clinical diagnosis:
☐ NF1
☐ Spinal NF
☐ NF Noonan
☐ Multiple CAL spots
☐ Isolated neurofibromas
☐ Single NF1 feature
☐ Legius syndrome
☐ Familial multiple CAL spots
☐ Segmental NF1
☐ Watson Syndrome
☐ Unknown

Family history: ☐ Sporadic (proband is a "founder" ☐ Familial (proband is a "non-founder") ☐ Unknown
Consanguinity: ☐ Yes ☐ No ☐ Unknown

GENERAL INFORMATION

Height: ____cm Head circumference: ____cm Weight: ____kg

NF SIGNS AND SYMPTOMS

1) CAL spots: ☐ 0 ☐ 1-5 ☐ ≥6 to 100 ☐ >100
General impression on the borders of the CAL-spots:
☐ typical well-defined smooth borders diameter:
☐ irregular margins, ragged borders diameter:
for detail on size and location of the CAL-spots and other hyper/hypopigmentation areas: page 3.

2) Skin fold freckling: ☐ None

	Left	Right	Comments (e.g. very faint,…..):
Groin	☐	☐	_____
Axilla	☐	☐	_____
Submammary	☐	☐	_____

3) Lisch nodules: ☐ None ☐ Left ☐ Right ☐ Unknown

4) Cutaneous neurofibromas (soft nodules that project above the skin) : histopathologically confirmed: Y / N
☐ 0 ☐ 2-6 ☐ 6-99 ☐ 100-500 ☐ >500

5) Intradermal neurofibromas (soft depression within the skin w/ pinkish overlying discoloration): histopathologically confirmed: Y / N
☐ 0 ☐ 2-6 ☐ 6-99 ☐ 100-500 ☐ >500

6) Subdermal neurofibromas (firm nodules palpable underneath the skin): histopathologically confirmed: Y / N
☐ 0 ☐ 2-6 ☐ 6-99 ☐ 100-500 ☐ >500

7) Plexiform neurofibromas: histopathologically confirmed: Y / N

☐ None ☐ visible from outside ☐ with hyperpigmentation ☐ without hyperpigmentation ☐ internal

☐ Head ☐ Neck ☐ Trunk ☐ L Arm ☐ L Hand ☐ L Leg ☐ L Foot
☐ Abdomen ☐ Pelvis ☐ Genital area ☐ R Arm ☐ R Hand ☐ R Leg ☐ R Foot

8) Spinal neurofibromas (neurofibromas arising from the spinal nerve root): histopathologically confirmed: Y / N

☐ Unknown ☐ Absent by MRI ☐ Present, asymptomatic ☐ Present, symptomatic

If present: please provide detail on **figure** page 4
☐ unilateral or ☐ bilateral;
C_____ T_____, L_____, S_____ regions.

9) Optic glioma: ☐ Unknown ☐ Absent by MRI ☐ Present by MRI, **symptomatic** ☐ Nerve (L and/or R) ☐ Chiasm ☐ Present by MRI, **asymptomatic** ☐ Nerve (L and/or R) ☐ Chiasm

10) Other neoplasms: ☐ None
☐ Hypothalamic glioma ☐ Brainstem glioma ☐ Other glioma
☐ MPNST ☐ JMML ☐ Rhabdomyosarcoma
☐ Pheochromocytoma ☐ Colonic polyps ☐ Lipoma
☐ juvenile xanthogranuloma ☐ Breast cancer
☐ schwannoma ☐ Other, specify:_____

11) Skeletal Abnormalities: ☐ None
☐ Long bone dysplasia ☐ Pseudoarthrosis ☐ Sphenoid wing dysplasia
☐ Bone cysts ☐ scoliosis ☐ Dysplastic vertebrae
☐ pectus excavatum ☐ pectus carinatum ☐ Other:_____

12) Cardiovascular disease: ☐ Absent ☐ Unknown ☐ Present: ☐ Hypertension ☐ Pulmonic stenosis ☐ Aortic stenosis ☐ Renal artery stenosis ☐ Moya moya ☐ ASD ☐ VSD ☐ Other_____

13) Development: ☐ Normal ☐ Exam not done

☐Abnormal ☐ ADD ☐ Hyperactivity ☐ Learning disability ☐ speech delay ☐ autism ☐ pervasive developmental delay
IQ: Full scale _____, Verbal _____, Performance _____.

14) Education: ☐ Too young for school ☐ At or above age level ☐ Below age level ☐ Unknown ☐ HS completion ☐ College graduate ☐ Higher degree

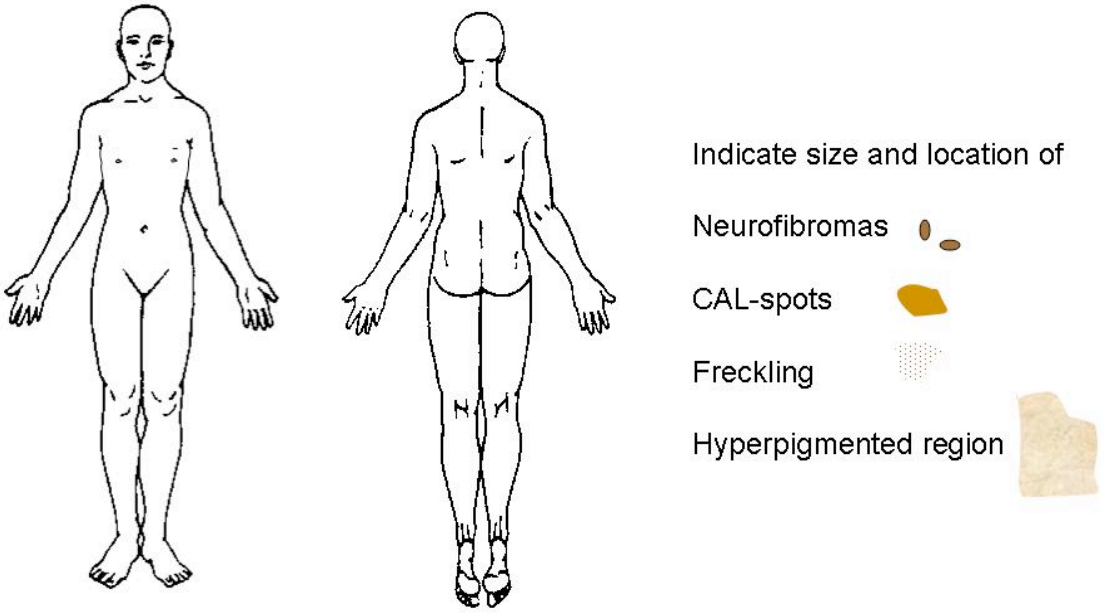
15) Noonan phenotype:

☐ Absent ☐ Possible ☐ Unknown

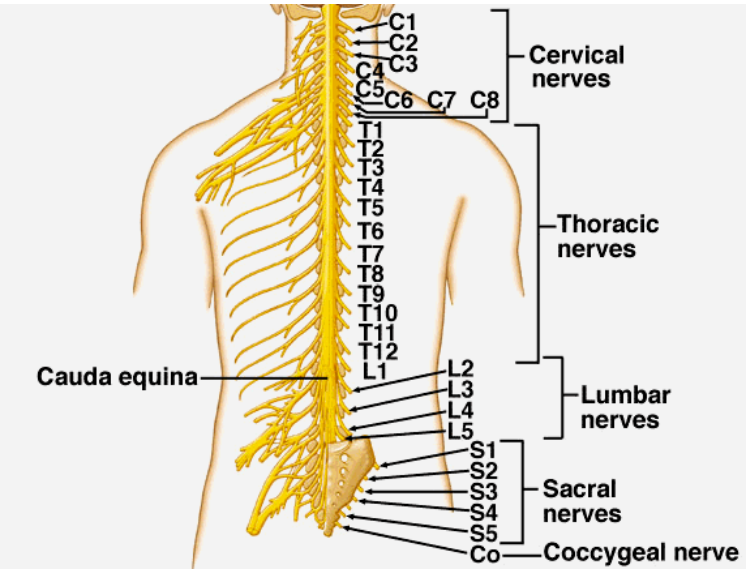
☐ Present: ☐ Short stature ☐ Low set ears ☐ Pulmonic Stenosis
☐ Hypertelorism ☐ Webbed neck peak jet velocity ____m/s
☐ Midface hypoplasia peak gradient _____mmHg across pulmonic valve
☐ PTPN11 testing done? Result _____

16) Segmental NF phenotype: ☐ Absent ☐ Possible

location/size of pigmentary lesions and/or neurofibromas



17) location of spinal tumors (if present)



18) any other comments/remarks: