H9.1-B13, v3, in voege 30/4/18

Clinical information sheet

Neurofibromatosis (NF)

NNFF INTERNATIONAL NF1 GENETIC MUTATION ANALYSIS CONSORTIUM SUBMISSION FORM

CENTRUM MEDISCHE GENETICA UZ GENT

FORM USE

Name of investigator (required field): _____

Institution (required field): _____

Please include phone number and/or e-mail address (one of them is required so that we may contact you if we have questions about your submission):

Investigator's e-mail address: _____

Investigator's Phone Number: _____

PART I DEMOGRAPHIC INFORMATION

Mutation Database Number: _____

See above under Definition of General Terms. Enter this number if this is not the first submission for a given patient.

I.1 NNFFIDB Number: _____

Clinical Database Participants Only

See above under Definition of General Terms. If this patient is known to have been entered into the Clinical Database, please complete this information, otherwise leave it blank.

I.2 Sex

☐ Male

☐ Female

In the case of ambiguous gender, use the chromosomal definition of sex.

I.3 Date of Birth: _____

If there is any uncertainty as to birth date, enter the date which is commonly used by the government on health records. Date of birth is used by the Central Database Centre as the primary variable to ensure that a patient or family member is only entered once into the Database. It is therefore essential that this information be accurate.

I.4 Date of Death

Enter exact date of death. If only year, or month and year, is known, enter that information and leave the other fields blank.

☐ Deceased, Date of Death: _____

☐ Not applicable (still alive)

☐ Deceased, but date unknown

I.5 Maternal ethnic origins?

☐ Asian – Japanese
☐ Asian – Chinese
☐ Asian – Indian Sub-continent
☐ Asian – Other
☐ Black
☐ Latin American – Other
☐ Native American – Other
☐ White
☐ Other – Specify: _____
☐ Other – Unknown

I.6 Paternal ethnic origins?

☐ Asian – Japanese
☐ Asian – Chinese
☐ Asian – Indian Sub-continent
☐ Asian – Other
☐ Black
☐ Latin American – Other
☐ Native American – Other
☐ White
☐ Other – Specify: _____
☐ Other – Unknown

PART II

II.1 N.I.H. Diagnostic Criteria

Please indicate which of the diagnostic criteria are met for NF1 in this patient. (Check all that apply)

☐ Six or more cafe au lait macules: Prepubertal > 5mm,Postpubertal > 15mm
☐ Two or more neurofibromas of any type or one plexiform neurofibroma
☐ Freckling in the axillary or inguinal regions
☐ Optic glioma
☐ Two or more Lisch nodules
☐ A distinct osseous lesion such as sphenoid dysplasia
☐ A first-degree relative with NF1 by the above criteria

II.2 PEDIGREE

Please use the space on this page to provide a pedigree using the symbols listed on the form. Omit names and local identification/lab numbers but include the Mutation Database Number and Clinical Database (NNFFID) Number where available.

The format for describing the pedigree should look similar to the following:

Individual	Gender	NF-Affected	Tested for Mutation

Proband			
Sibling 1			
Sibling 2			
etc.			
Father			
Mother			
etc.			

Please enter your pedigree information in the following text box:

PART III NF SIGNS AND SYMPTOMS

III.1 Date of Exam: _____

If exact date of latest exam is unknown complete the month and year fields as appropriate.

III.2 Superficial neurofibromas (check all types that are present)

- ☐ None
- ☐ Cutaneous
- ☐ Subcutaneous
- ☐ Pendulous
- ☐ Present – type unknown
- ☐ Unknown

Use the "Unknown" option if it is not known whether or not the patient has superficial neurofibromas.
A subcutaneous neurofibroma is located beneath the skin and will move little when the surrounding skin is pulled laterally. A cutaneous neurofibroma is located superficially in the skin and will move with the surrounding skin when the skin is pulled laterally. A pendulous neurofibroma is located above the skin surface and appears as a tag, or is connected to the body by a stalk.
Use the following space to record additional relevant information or to indicate any unusual findings such as pigmentation, atypical biopsy results or unusual distribution:

_____

III.3 Plexiform neurofibromas (check all locations that apply)

- ☐ None
- ☐ Orbit
- ☐ Face
- ☐ Head/Neck
- ☐ Trunk – dorsal
- ☐ Trunk – ventral
- ☐ Arm
- ☐ Leg
- ☐ Present – location unknown
- ☐ Unknown

If a plexiform lesion extends over more than one of the locations listed above, choose the area involved with the greatest portion of the neurofibroma for each plexiform.
Use the following space to record additional relevant information or to indicate any unusual findings such as pigmentation, atypical biopsy results, unusual location:

_____

III.4 Internal neurofibromas

- ☐ Absent
- ☐ Present – asymptomatic
- ☐ Present – symptomatic
- ☐ Possible
- ☐ Unknown

Use the following space to note the location and size of internal neurofibromas and briefly describe any symptoms they are causing:

_____

III.5 Neurological Problems

☐ Absent
☐ Present (please describe in following comment field)
☐ Unknown

Use the following space to record any neurological problems, whether or not they appear related to NF1:

III.6 Education completed

☐ Not applicable (too young for school)
☐ Still in school at or above age level
☐ Still in school behind age level
☐ Grade 5 or less
☐ Grade 6-9
☐ Grade 10-high school completion
☐ Undergraduate degree
☐ Higher degree
☐ Unknown

Educational level is at or above the age appropriate level if the child is in a regular school and is passing all subjects. Child is behind age appropriate level if he is in a special school for the learning disabled, needs substantial classroom assistance or is failing subjects or grades in a regular school.
Please use the space below to note any special circumstances which would explain early school leaving or failing grades in a child or adult. Examples include a child who has repeated a grade as a result of extensive hospitalizations or immigrants whose first language is not that of your school system.

III.7 Developmental examination

☐ Not Done
☐ Normal (provide I.Q. if known): _____
☐ Abnormal (provide I.Q. if known): _____
☐ Unknown

Accept results of any testing done by a qualified professional. Comment on areas of abnormality such as significantly delayed verbal or spacial abilities:

III.8 Cardiovascular disease

☐ Absent
☐ Present (please describe with a comment)
☐ Unknown

Comment on any clinical findings, ECG, angiogram, ultrasound abnormalities etc. and on any specific diagnosis such as aortic stenosis, ASD, hypertension, renal artery stenosis etc.

III.9 Skeletal anomalies

☐ Absent
☐ Present (please describe with a comment)
☐ Unknown

Comment on abnormalities such as pseudoarthrosis, congenitally bowed tibia, scoliosis, dysplastic vertebrae, dysplastic sphenoid wing, bony overgrowth etc.

III.10 Gastrointestinal complications

☐ Absent
☐ Present (please describe with a comment)
☐ Unknown

Do not include uncertain diagnoses. Do not include tumours under this item.
Comment on abnormalities such as pseudoarthrosis, congenitally bowed tibia, scoliosis, dysplastic vertebrae, dysplastic sphenoid wing, bony overgrowth etc.

III.11 Noonan phenotype

☐ Absent
☐ Present (please describe with a comment)
☐ Possible
☐ Unknown

Use "Possible" option if patient has borderline features of Noonan Syndrome. Use a comment to specify features, e.g. short stature, low set ears, midface hypoplasia, hypertelorism, ptosis, webbed neck, pulmonic stenosis, etc.

III.12 Other dysmorphic features

☐ Absent
☐ Present (please describe with a comment)
☐ Unknown

Use a comment to specify the dysmorphic feature, e.g. large ears, prominent jaw. Do not include the effects of plexiform neurofibromas or other overgrowth which have been described in items III.3 or III.9 above.

III.13 Neoplasms

☐ Absent
☐ Present (please describe with a comment)
☐ Unknown

Specify type of neoplasm e.g. pheochromocytoma, sarcoma, Schwannoma. Specify location, histology, age of onset, and course of any neoplasm with a comment.
