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| FOR LAB USE ONLY |
| ACCESSION NO: |
| DATE & TIME RECEIVED: |
| TECHNICIAN: |

UAB MEDICAL GENOMICS LABORATORY
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NEUROFIBROMATOSIS TYPE 2 TEST REQUEST FORM

| | | | | |
|--|---|---|------------------------------------|-------------------|
| THIS FORM AND PHENOTYPIC CHECKLIST MUST BE FILLED OUT COMPLETELY | | | | |
| PATIENT NAME: | BIRTH DATE: | DAYTIME PHONE: | SEX: | SOC. SEC. NUMBER: |
| PATIENT'S ADDRESS: | CITY: | STATE: | ZIP CODE: | MED REC NUMBER: |
| EMAIL ADDRESS: | | PARENT OR GUARDIANS NAME (IF MINOR): | | |
| NF2 MUTATION ANALYSIS TESTING | | Physician's Name: | | |
| <input type="checkbox"/> Comprehensive NF2 Test (Test 1) <input type="checkbox"/> RUSH? <input type="checkbox"/> Copy Number Analysis by MLPA <i>only</i> <input type="checkbox"/> Targeted Mutation Analysis (Test 2) Proband _____ <input type="checkbox"/> Prenatal Targeted Analysis (Test 3) Proband _____ <input type="checkbox"/> Comprehensive Test on Tumor (Test 4) | | Physician's Address: _____ _____ _____ | | |
| Facility where specimen obtained: | | Phone: _____ Fax: _____ | | |
| Date: | | NPI Number: _____ Email: _____ | | |
| REQUIRED Diagnosis (ICD-9) Code (only in US): | | ADDITIONAL REPORTS TO: Name: _____ | | |
| Is Patient Pregnant? <input type="checkbox"/> No <input type="checkbox"/> Yes If yes, date of LMP: _____ | Please check if applicable: <input type="checkbox"/> Infectious diseases (AIDS, Hepatitis, etc) | | Mailing Address: _____ _____ | |
| Specimen type: | | Phone No: _____ Fax No: _____ | | |
| <input type="checkbox"/> Cheek Swabs; # Swabs: _____ | <input type="checkbox"/> Amniotic Fluid | <input type="checkbox"/> Cultured CVS | | |
| <input type="checkbox"/> Peripheral Blood (EDTA); # Tubes: _____ | <input type="checkbox"/> Direct CVS | <input type="checkbox"/> Cultured Amniocytes | | |
| | | <input type="checkbox"/> Tumor (specify): _____ | | |

BILLING INFORMATION

BILL INSTITUTION: (Please, provide name, address, and telephone number of entity responsible for payment)
 Purchase Order Number: _____ Contact Name: _____
 Billing address: _____ Phone #: _____
 _____ Fax #: _____
 _____ Email: _____

PAYMENT ENCLOSED:

Cashier's Check
 VISA® MasterCard® Discover® American Express®
 Card Number: _____ Expiration Date: _____
 Name as it appears on card: _____ 3-digit Security Code: _____
 Cardholder Signature: _____
 Cardholder Email Address: _____

BILL CONTRACTED INSURANCE COMPANY:
 Please include a copy of patient's insurance card. For a list of our contracted insurance companies, please visit our website at www.genetics.uab.edu/medgenomics, under "Billing". Please include a copy of pre-approval statement if payment has been authorized. **We also need the patient's credit card information so that any balance left after insurance pays may be applied to it. The RUSH fee must be paid up front by the patient.**

FILE INSURANCE CLAIM WITH NON CONTRACTED COMPANY
 Patient must pay full payment for test up front, (credit card or cashier's check) but UAB will file a claim for reimbursement with the patient's insurance company. Please send a copy of patient's insurance card, front and back. **The RUSH fee must be paid up front by the patient.**

Informed Consent for NF2 Testing

I, _____, hereby agree to participate in testing for Neurofibromatosis type 2 using a RNA/DNA-based cascade of tests. I understand that biological samples (blood, cheek cells) will be removed from me using standard techniques which carry very little risk. In addition, if prenatal diagnosis is being performed, fetal cells obtained by chorionic villus sampling or amniocentesis will be used. I understand that the blood, cheek cells or fetal samples will be used for the purpose of attempting to determine if I and/or members of my family are carriers of the disease gene. In addition I hereby give permission to collect biological samples from my minor children, named below, to be used for RNA/DNA-testing for the disease listed above.

| Child's name | Date of Birth | Gender (F/M) |
|--------------|---------------|--------------|
| _____ | _____ | _____ |

I understand that:

1. In ~92% of the NF2 **non-founder** patients fulfilling the NIH diagnostic criteria, the RNA/DNA cascade of tests detects an abnormality, called a mutation, in the NF2 gene, and the test is >99% accurate. Rare variations in the DNA of individuals can sometimes be found and can cause uncertainty in predicting the carrier status.
2. In other cases, the RNA-DNA test is unable to identify an abnormality although the abnormality may still exist. This event may be due to our current lack of knowledge of the complete gene structure or an inability of the current technology to identify certain types of mutations in the gene. The mutation detection system employed by the Medical Genomics laboratory for identifying NF2 mutations is the most sensitive yet developed. I have been informed of the likelihood of finding a mutation in the gene for which I am being tested. _____ (Initials)
3. I understand that the RNA/DNA NF2 analysis performed by the Medical Genomics Laboratory is specific for this disease and in no way guarantees my health or the health of my living or unborn children. The Medical Genomics Laboratory cannot be responsible for erroneous clinical diagnosis made elsewhere.
4. In order to perform accurate prenatal diagnosis, biological samples are required from the fetus as well as from the affected individual in the family and from the biological mother.
5. This test is relatively new and is being expanded and improved continuously. The test is not considered research, but is considered the best and newest laboratory service that can be offered. This testing is complex and utilizes specialized materials so that there is always some very small possibility that the test will not work properly or that an error will occur. There is a low error rate (perhaps 1 in 1000 samples) even in the best laboratories. My signature below acknowledges my voluntary participation in this test, but in no way releases the laboratory and staff from the Medical Genomics Laboratory from their professional and ethical responsibility to me.
6. I understand that my sample is not being banked. The laboratory does not return DNA samples to individuals or physicians. However, in some cases it may be possible for the laboratory to reanalyze my remaining DNA upon request. The request for additional studies must be ordered by my referring physician/counselor and there will be an additional fee.
7. Once my test result is completed, an aliquot of my DNA/RNA may be made anonymous (name and all other identifiers removed) and used for research purposes. I understand declining to participate in this research will in no way negatively impact my clinical testing or treatment.

PLEASE CHECK ONE OF THE FOLLOWING

- By checking this box, I indicate my desire to opt out of participation in anonymized research studies using my DNA/RNA sample.
8. Because of the complexity of RNA/DNA based testing and the important implications of the test results, results will only be reported to me through a physician, genetic counselor or certified genetics professional. The result reports are confidential and will only be released to other medical professionals or other parties with my express written consent. All laboratory data and personal information are strictly confidential and will not be released from the Medical Genomics Laboratory in full compliance with HIPPA. Participation in RNA/DNA testing is completely voluntary.
 9. I will receive a copy of this consent form.

Signature: _____
Witnessed by: _____
Date: _____

Physician's/Counselor's statement: I have explained RNA/DNA testing to this individual. I have addressed the limitations outlined above and have answered person's questions.

Signature: _____

NF2 PHENOTYPIC CHECKLIST FORM – UAB MEDICAL GENOMICS LABORATORY

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

DEMOGRAPHIC INFORMATION

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

Ethnicity: Mother: White African-American Native American Hispanic Asian Other
Father: White African-American 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

NF2 SIGNS AND SYMPTOMS

1) Ears

Deafness Bilateral Unilateral Age of symptoms: _____

Balance Dysfunction Age of Symptoms: _____

Audiometric Abnormality
If yes, please describe: Age of Symptoms: _____

Other
If Yes, please describe: Age of Symptoms: _____

2) Eyes

Blindness Age of Symptoms: _____

Lenticular opacities Age of Symptoms: _____

Lisch nodules Left Right Bilateral Unknown
Age of Symptoms: _____

3) CNS

Vestibular schwannomas Bilateral Unilateral Age of Symptoms: _____

Meningiomas
If yes, please provide detail on location: Age of Symptoms: _____

Spinal tumors: Absent by MRI Present, asymptomatic Present, symptomatic Unknown
Provide location of spinal tumors: C__ to C__, T__ to T__, L__ to L__
Histopathology proven schwannomas (vs neurofibromas)? Y/N

Cranial nerve involvement
If yes, please provide further details: Age of Symptoms: _____

4) 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 | Bilateral |
| Groin | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| Axilla | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |
| Submammary | <input type="checkbox"/> | <input type="checkbox"/> | <input type="checkbox"/> |

 Age of Symptoms: _____

Schwannomas (histopathology proven: Y/N) Age of Symptoms: _____

Please provide detail on location/ number of schwannomas

- Updated 8-10-09 -