

## EDDNAL Disease Registration form

Please complete this form (second page is optional) for each disease for which your laboratory performs diagnostic testing.

### I. Disease Information

1. Disease Name/Abbreviation \_\_\_\_\_
2. Gene(s)/Locus(I) \_\_\_\_\_
3. OMIM# \_\_\_\_\_
4. Synonyms (optional) \_\_\_\_\_
5. Contact person for this disease \_\_\_\_\_

### II. Testing Information

1. Type of diagnosis:  
 Symptomatic                       Asymptomatic carrier status  
 Presymptomatic                       Prenatal
2. Material needed:  
 RNA                       DNA                       Other (please specify): \_\_\_\_\_
3. Methodology:  
*A. Direct Testing*  
 PCR-SSCP                       PCR-DGGE                       PCR-RFLP  
 DHPLC                       PTT                       Southern blotting  
 Sequencing: automated / manual                       Panel of mutations (Number)  
 Commercial kit – name: \_\_\_\_\_  
 Homemade technique  
 FISH analysis  
 Other: \_\_\_\_\_  
*B. Indirect Testing*  
 Intragenic                       Extragenic  
*C. Other methods*  
 Methylation                       Protein truncation test  
 Uniparental disomy                       Other: \_\_\_\_\_

### III. Additional Information

1. Target report time : \_\_\_\_\_ weeks
2. Do you accept samples from foreign countries? Y / N  
If YES :
  - What is the price of this service \_\_\_\_\_ €
  - Would you perform this analysis for free in special circumstances (developing countries, scientific interest,)? Y / N
  - Do you need clinical and/or genetic information before performing this test? Y / N
  - Do you need a preliminary agreement on reimbursement and clinical relevance before samples are sent? Y / N
  - Additional comments/other sample requirements: \_\_\_\_\_

## EDDNAL Disease Registration form (cont.)

*This page is optional. Please provide this information if it is available and at your discretion.*

### IV. Experience

1. Number of tests performed since 1996 for this disorder:

Symptomatic: \_\_\_\_\_ Asymptomatic carrier status: \_\_\_\_\_  
Presymptomatic: \_\_\_\_\_ Prenatal: \_\_\_\_\_

2. Number of positive tests for this disorder:

Symptomatic: \_\_\_\_\_ Asymptomatic carrier status: \_\_\_\_\_  
Presymptomatic: \_\_\_\_\_ Prenatal: \_\_\_\_\_

3. Do you offer genetic counselling for this disorder: Y / N

### V. Quality Assessment

1. Are you certified by a quality assessment association for this analysis? Y/N

If **YES**, please give the name: \_\_\_\_\_

### VI. Research\*

1. Please give a brief description of your research. \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

2. Citations relevant to your laboratory's research on this disease: (MUST include first author last name/initials, full journal name, volume, page, year) \_\_\_\_\_  
\_\_\_\_\_  
\_\_\_\_\_

3. Would you be interested in receiving DNA samples from patients affected with this diseases on a research basis? Y / N

\* Should your laboratory be engaged in only research, please complete the Laboratory Registration Form and Parts I and VI of the Research form.