EDDNAL Laboratory Registration form

Please complete this page once and the following page(s) once per each disease. This form can be returned to EDDNAL either via email (eddnal@skypro.be) or via fax +32 24 77 29 40 (attn. EDDNAL).

Person completing this form_____Date_____

I. Laboratory Specific Information*

- Name of Laboratory_____
- Name of Institution_____
- Head of the Laboratory _____

  Degree(s)_____Field/Speciality_____

  Email_____

  Phone_____Fax_____
- Complete Mailing Address _____

  _____

  _____
- Lab URL (if lab has a web site)_____

Is your laboratory certified by an official organisation in your country that allows you to perform mutation analysis? Y / N

If **YES**, by which one?_____

II. Contact Person Information*

Contact Name 1 _____

Degree(s) _____Field/Speciality_____

Email_____

Phone_____Fax_____

Contact Name 2 (optional) _____

Degree(s) _____Field/Speciality_____

Email_____

Phone_____Fax_____

* *This information will be posted on the EDDNAL web-site.*

EDDNAL Disease Registration form

Please complete this form (second page is optional) for <u>each disease</u> for which your laboratory performs <u>diagnostic</u> 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 <u>only</u> research, please complete the Laboratory Registration Form and Parts I and VI of the Research form.*