"MAURO BASCHIROTTO"
INSTITUTE FOR RARE DISEASES

International Prader-Willi Syndrome Diagnostic Testing Initiative

IPWSO
INTERNATIONAL
PRADER-WILLI SYNDROME
ORGANISATION

International Prader-Willi Syndrome Organisation (IPWSO) in collaboration with the "Mauro Baschirotto" Institute for rare diseases n.p.o. (BIRD) are offering diagnostic testing for **Prader-Willi syndrome (PWS)**.
The test method to be used is methylation-specific PCR amplification of the CpG islands of the SNRPN gene located inside the 15q11-q13 region. This test will detect about 99% of the cases of PWS.
The tests are conducted free of charge on DNA isolated from dried blood spots. The turnaround time is usually 3-12 weeks, depending on the workload of the lab.
The results are written in English and are sent by mail and/or email to the physician sending the sample.
ONLY SAMPLES ORDERED BY MEDICAL DOCTORS CAN BE ACCEPTED.
Genetic counseling regarding the results of the tests is available upon request.

Please feel free to contact us for any further information.

Instructions for sending samples:

In order to be eligible for molecular testing through this program, the subject must have a clinical diagnosis or a strong clinical suspicion of Prader-Willi syndrome made by a medical doctor.

Criteria sufficient to suggest PWS have been published (Gunay-Aygun M et. al.; PEDIATRICS Vol. 108 No. 5, E92 November 1, 2001) and must be satisfied for us to conduct the testing:

Age at Assessment	Features Sufficient to Prompt DNA Testing
Birth to 2 years	Hypotonia with poor suck.
2 – 6 years	Hypotonia with history of poor suck and.
	Global developmental delay.
6 – 12 years	History of hypotonia with poor suck (hypotonia often persists) and.
	Global developmental delay and.
	Excessive eating (hyperphagia; obsession with food) with central obesity if uncontrolled.
>13 years	Cognitive impairment; usually mild mental retardation and.
	Excessive eating (hyperphagia; obsession with food) with central obesity if uncontrolled and.
	Hypothalamic hypogonadism and/or typical behavior problems (including temper tantrums and obsessive-compulsive features).

A detailed list of the major, minor and supportive features of PWS and natural history can be found in Holm VA et al., PEDIATRICS, vol. 91 number 2, pages 398-402, 1993 and at http://www.ncbi.nlm.nih.gov/books/NBK1330/

Before sending a sample, please contact "Mauro Baschirotto" Institute for Rare Diseases for acceptance by providing the following information:

- a short clinical description ; please include:
  - age, sex
  - whether there was the presence of neonatal hypotonia with poor suck and feeding difficulties in the first months of life,
  - weight at birth, age of initial weight gains, current weight
  - age at which developmental milestones were attained
  - clinical history
- prior genetic testing results (if any)
- photographic material (if possible)

The information should be sent to: consulenze@birdfoundation.org

The information will be reviewed and you will be informed if the sample can be accepted for testing. If it is, please follow the instructions below.

1. Sample collecting and preparation:

A medical professional should collect 8-12 blood spots (6-8 drops of fresh blood each) on thick filter paper for chromatography (Whatman or equivalent; do not use other types of paper, if in doubt please contact our lab). The spotted blood should be without anticoagulant or with EDTA. It is important the spotted paper is kept in a clean, dry and dark environment for a few days until it is completely dry (a clean carton box or a drawer are suitable; if no such place is available the samples can be covered with tissue paper, to avoid dust and contaminants, and left on an even surface to dry). Once dry, put the filter paper inside a sterile plastic bag for laboratory use to protect it during transport. Do not use sterile test-tubes.

2. Letter:

a. Patient Information
- name and surname (please underline the surname)
- birth date
- sex
- mail address

b. Sender information
- name and surname (please underline the surname)
- mail address
- telephone and fax numbers
- e-mail address

c. Results
- please specify where the results should be sent and preferred method (mail or email)

d. Test to be performed
- Please state that testing for Prader-Willi syndrome is requested

3. Sending the sample:

Please send the sample as a normal letter by regular mail and not by an express courier to the following address:

Medical Genetics Unit
"Mauro Baschirotto" Institute for Rare Diseases - B.I.R.D.
Via B. Bizio, 1 - 36023 Costozza di Longare (VI) - Italy

4. For assistance:

E-mail: Uros Hladnik, MD, spec. in medical genetics
uros.hladnik@birdfoundation.org
Telephone: +39 0444 555557
FAX: +39 0444 555034