"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 non-profit organization. (BIRD) are offering diagnostic testing for **Prader-Willi syndrome (PWS)** without charge for people without affordable available testing in their country.
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 e-mail to the physician sending the sample. Genetic counseling regarding the results of the tests is available upon request.

ONLY SAMPLES ORDERED BY MEDICAL DOCTORS CAN BE ACCEPTED.

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
	Global developmental delay
6 – 12 years	Hypotonia with history of poor suck
	Global developmental delay
	Excessive eating (hyperphagia; obsession with food) with central obesity if uncontrolled
>13 years	Hypotonia with history of poor suck
	Cognitive impairment; usually mild intellectual disability
	Excessive drive to eat (hyperphagia; obsession with food) with central obesity if uncontrolled
	Hypothalamic hypogonadism OR
	Behavior problems (including temper tantrums and obsessive-compulsive features)

Please notice that the features described in this table are not the only ones present but in their absence the diagnosis of Prader-Willi syndrome is highly unlikely. (hypothalamic hypogonadism for instance is observed as genital hypoplasia at all ages but in younger individuals isn't included as a required criteria, in order to promode very early PWS diagnoses for infants the sole presence of hypotonia is a sufficient criteria for PWS testing).

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 freely available on the internet at the Gene Reviews page 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:

- The included International PWS Sample Sending - Clinical data form filled in
- Photographic material
- Additional clinical data (if available)

The information should be sent to: consulenze@birdfoundation.org ; uros.hladnik@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. Only exceptionally will we allow the testing of cases not featuring the required criteria to prompt DNA testing.

1. Sample collecting and preparation:

A medical professional should collect 8-12 blood spots (6-8 drops of fresh blood each) on thick laboratory filter paper (Whatman 903 or equivalent; do not use other types of paper as they can interfere with the lab procedures; if in doubt, please contact our lab). The spotted blood should be either without anticoagulant or with EDTA as anticoagulant (other types of anticoagulant may interfere with the test). 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 designed for laboratory use to protect it during transport. If the sample isn't completely dried before packing in the sterile bag, the DNA will degrade over time and there is a high risk of yeast infections. Clearly mark the full name of the tested person, together with the date of birth on the margin of the filter paper.

2. The International PWS Sample Sending - Clinical data form and photographic material

Please include the Clinical data form completely filled in and the photos of the tested person.

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. Foundation n.p.o.
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