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"MAURO BASCHIROTTO"
INSTITUTE FOR RARE DISEASES

International Prader-Willi syndrome genetic screening initiative

IPWSO
International
Prader-Willi Syndrome
Organisation

Information for families

International Prader-Willi Syndrome Organisation (IPWSO) in collaboration with the non-profit organization "Mauro Baschirotto" Institute for Rare Diseases (B.I.R.D.) are offering a free genetic screening test for **Prader-Willi syndrome (PWS)** for people who are unable to access available testing in their country. The test methods used will detect about 99% of the cases of PWS.

In which cases should this test be performed?
If your doctor suspects the presence of Prader-Willi syndrome.

How do people send a sample for analysis?
Discuss this testing opportunity with your doctor. The doctor should contact the B.I.R.D. laboratory to find out how to send a sample for testing.

How does this procedure work?

- First your doctor will submit information about the person to be tested to the B.I.R.D. laboratory for approval. The data required relate to birth weight and length, clinical history and current weight and height. Some clinical photos will also be needed. The doctor will explain the test and ask you to sign an informed consent. You will be able to ask questions and receive answers regarding any aspect of the testing procedure.
- Once the case is approved for testing, a blood sample will be taken and sent to the B.I.R.D. laboratory.
- The tests are conducted free of charge.
- The turnaround time is usually 3-12 weeks, depending on the workload of the lab.
- The results will be written in English and sent by e-mail to the medical doctor sending the sample.
- The medical doctor sending the sample will inform the family about the result. The result will not be sent directly to the family.

If the genetic screening test is positive, genetic counseling is needed for the family. This is not provided by IPWSO. Genetic counseling should ideally be provided by a medical geneticist or genetic counselor. If none are available, then genetic counseling should be done by the physician who requested the genetic screening. A detailed review of the genetics of PWS can be found at: https://www.ncbi.nlm.nih.gov/books/NBK1330/

For assistance:
Medical Genetics Unit
"Mauro Baschirotto" Institute for Rare Diseases - B.I.R.D. Foundation n.p.o.
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To learn more about PWS and other free services offered by the International Prader-Willi Syndrome Organisation, visit www.ipwso.org or email office@ipwso.org.

All samples and all the documentation will be handled with maximum respect for privacy and the tests performed follow the best practice guidelines regarding genetic testing.