**Laboratory of Personalized Genomic Medicine**
**Department of Pathology and Cell Biology**

COLUMBIA UNIVERSITY MEDICAL CENTER

630 West 168th Street
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New York, NY 10032
Tel: 212-305-9706
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# Informed Consent for Prader-Willi Angelman Syndrome by PCR

Please read the following form carefully and discuss with your ordering physician/genetic counselor before signing consent.

1. This is a genetic (DNA-based) test that detects the presence of maternally and/or paternally inherited region of chromosome 15, using a PCR-based assay.

2. The purpose of this analysis is to test for Prader-Willi and Angelman syndromes.
   2a. You (or the person for whom you are signing) may want genetic counseling before signing consent.

3. This is a test for genetic susceptibility ("genetic predisposition"). If the test is positive, you may wish to have further independent testing, consult your physician or have genetic counseling.

4. The condition(s) being tested for are Prader-Willi and Angelman syndromes, two different disorders affecting development and behavior.

5. When the paternal copy is absent, there is an approximately 99% chance of manifesting Prader-Willi Syndrome. When the maternal copy is absent, there is an approximately 99% chance of manifesting Angelman Syndrome. When a normal methylation pattern is present, the risk of having Angelman Syndrome or Prader-Willi Syndrome will depend upon clinical manifestations. Up to 16% of Angelman Syndrome patients, and up to 5% or Prader-Willi Syndrome patients will have normal results with this test.

6. The results of the above test become a part of the patient's medical record, and may be made available to individuals/organizations with legal access to the patient's medical record, on a strict "need-to-know" basis, including but not limited to the physicians and nursing staff directly involved in the patient's care, the patient's current and future insurance carriers, and other specifically authorized by the patient/authorized representative to gain access to the patient's medical records. Columbia University, NewYork-Presbyterian and Weill Cornell Medicine and their related entities participate in an Organized Health Care Arrangement (OHCA). This allows us to share health information to carry out treatment, payment and our joint health care operations, including integrated information system management, health information exchange, financial and billing services, insurance services, insurance, quality improvement, and risk management activities. Organizations that will follow this Notice include Columbia University, NewYork-Presbyterian sites, Weill Cornell Medicine and their related entities.

7. No additional tests will be performed on this sample, without specific, signed authorization by the patient.
After 60 days, unless consent is given the sample (DNA and received specimen) will be destroyed – please see below.

8. Medicare/Insurance Carriers may not pay for the test, in which case, the patient/responsible party will be billed for the test.

**Person obtaining consent**:

_____ _____ **Date:** _____

Print Name of Person Obtaining Consent  Signature of Person Obtaining Consent

**I have read and fully understood the above, and give my consent for this testing.**

**Patient** (person being tested):

_____ _____ **Date:** _____

Print Name of Patient/Authorized Representative  Signature of Patient/Authorized Representative

Relationship to Patient: _____

☐ _____ After 60 days, unless consent is given, the sample will be destroyed.

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**Note to Health Care practitioner obtaining consent to test for Prader-Willi Angelman Syndrome**

It is New York State Law that all genetic testing requires informed consent. Please ensure that the patient understands the contents of the informed consent and prior to signature. This is a Southern-Blot based test using the *SNRPN* probe with methylation sensitive restriction enzymes to distinguish maternal from paternal alleles. Individuals with only the maternal allele (either due to deletion of this region in the paternal chromosome 15, or to maternal uniparental disomy) will manifest Angelman Syndrome. The test does not distinguish between UPD or deletion. Furthermore, this test will miss cases caused by imprinting center defects, or mutations in individual genes.