Lab Dept:	**Anatomic Pathology**
Test Name:	**PRADER-WILLI / ANGELMAN SYNDROME MOLECULAR ANALYSIS METHYLATION**

General Information

Lab Order Codes:	PWMA
Synonyms:	Prader Willi/Angelman Syndrome Methylation Study; Methylation PCR Prader-Willi/Angelman Syndrome
CPT Codes:	81331 – SNRPN/UBE3A, methylation analysis
Test Includes:	Methylation-Sensitive Multiple Ligation-Dependent Probe Amplification (MLPA) with an interpretive report of findings.

Logistics

Test Indications:	Confirmation of diagnosis in patients suspected of having either Prader-Willi or Angelman Syndrome based on clinical assessment or previous laboratory analysis.
Lab Testing Sections:	Anatomic Pathology - Sendouts
Referred to:	Mayo Clinic Laboratories (MML: PWAS)
Phone Numbers:	MIN Lab: 612-813-6280 STP Lab: 651-220-6550
Test Availability:	Daily, 24 hours
Turnaround Time:	14 – 21 days
Special Instructions:	A "Molecular Genetics – Congenital Inherited Diseases Patient Information Sheet" (Mayo Supply T245) is required for all orders. An "Informed Consent Form for DNA Testing" (Mayo Supply T576) is available. Please contact the laboratory for forms.

Specimen

Specimen Type:	Whole blood
Container:	Lavender top or Yellow top ACD tube
Draw Volume:	3 mL (Minimum: 1 mL) blood

Processed Volume:	Same as Draw Volume
Collection:	Routine venipuncture
Special Processing:	Lab Staff: **Do Not centrifuge.** Submit specimen in original vacutainer tube at room temperature. Specimen must reach Mayo within 96 hours of collection. Forward promptly at room temperature.
Patient Preparation:	None
Sample Rejection:	Mislabeled or unlabeled specimens; incorrect anticoagulant

Interpretive

Reference Range:	An interpretive report will be provided.
Critical Values:	N/A
Limitations:	In addition to disease-related probes, the multiple ligation-dependent probe amplification technique utilizes probes localized to other chromosomal regions as internal controls. In certain circumstances, these control probes may detect other diseases or conditions for which this test was not specifically intended. Results of the control probes are not normally reported. However, in cases where clinically relevant information is identified, the ordering physician will be informed of the result and provided with recommendations for any appropriate follow-up testing. Rare variants (ie, polymorphisms) exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered. Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Errors in the interpretation of results may occur if information given is inaccurate or incomplete. Methylation status cannot be assessed on chorionic villus specimens. Rare cases of Prader-Willi syndrome or Angelman syndrome (AS) result from a subtle balanced translocation inherited from one of the parents. These may not be detected by this assay. A negative molecular test result, especially in the case of a clinical suspicion of AS, does not rule out the diagnosis, because point alterations may not be detected by these methods.
Methodology:	Methylation-sensitive multiple ligation-dependent probe amplification (MLPA)
References:	Mayo Clinic Laboratories November 2024

Updates: 1/13/2009: CPT updates
2/7/2013: CPT updates
11/20/2024: Minor updates