Test Definition: PWAS

Prader-Willi/Angelman Syndrome, Molecular Analysis, Varies

Reporting Title: Prader Willi/Angelman Mol Analysis
Performing Location: Rochester

Additional Testing Requirements:

Mayo Clinic Laboratories highly recommends that this test be ordered along with a routine chromosomal microarray analysis, CMACB / Chromosomal Microarray, Congenital, Blood, if the diagnosis of Prader-Willi syndrome (PWS) or Angelman syndrome (AS) is not certain and chromosome analysis has not already been done.

All prenatal specimens must be accompanied by a maternal blood specimen. Order MATCC / Maternal Cell Contamination, Molecular Analysis, Varies on the maternal specimen.

Shipping Instructions:

Specimen preferred to arrive within 96 hours of collection.

Specimen Requirements:

Patient Preparation: A previous bone marrow transplant from an allogenic donor will interfere with testing. For instructions for testing patients who have received a bone marrow transplant, call 800-533-1710.

Submit only 1 of the following specimens:

Specimen Type: Whole blood
Container/Tube:
Preferred: Lavender top (EDTA) or yellow top (ACD)
Acceptable: Any anticoagulant
Specimen Volume: 3 mL
Collection Instructions:
1. Invert several times to mix blood.
2. Send whole blood specimen in original tube. Do not aliquot.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Prenatal Specimens
Due to its complexity, consultation with the laboratory is required for all prenatal testing; call 800-533-1710 to speak to a genetic counselor.

Specimen Type: Amniotic fluid
Container/Tube: Amniotic fluid container
Specimen Volume: 20 mL
Specimen Stability Information: Refrigerated (preferred)/Ambient

Acceptable:
Specimen Type: Confluent cultured cells
Container/Tube: T-25 flask
Specimen Volume: 2 Flasks
Collection Instructions: Submit confluent cultured cells from another laboratory.
Specimen Stability Information: Ambient (preferred)/Refrigerated

Specimen Minimum Volume:
Blood: 1 mL
Amniotic Fluid: 10 mL

Forms:
1. New York Clients-Informed consent is required. Document on the request form or electronic order that a copy is on file. The following documents are available:
-Informed Consent for Genetic Testing (T576)
-Informed Consent for Genetic Testing-Spanish (T826)
2. Molecular Genetics: Congenital Inherited Diseases Patient Information (T521
3. If not ordering electronically, complete, print, and send a Neurology Specialty Testing Client Test Request (T732) with the specimen.

Specimen Type	Temperature	Time	Special Container
Varies	Varies		

Ask at Order Entry (AOE) Questions:

Test ID	Question ID	Description	Type	Reportable
CULAF	CG767	Reason for Referral	Plain Text	No

Result Codes:

Result ID	Reporting Name	Type	Unit	LOINC®
52913	Result Summary	Alphanumeric		50397-9
52914	Result	Alphanumeric		82939-0
52915	Interpretation	Alphanumeric		69047-9
52916	Reason for Referral	Alphanumeric		42349-1
52917	Specimen	Alphanumeric		31208-2
52918	Source	Alphanumeric		31208-2
52919	Released By	Alphanumeric		18771-6

LOINC and CPT codes are provided by the performing laboratory.

Supplemental Report:

No

CPT Code Information:

81331-SNRPN/UBE3A, (small nuclear ribonucleoprotein polypeptide Nand ubiquitin protein ligase E3A) (eg, Prader-Willi syndrome and/or Angelman syndrome), methylation analysis
88235-Tissue culture for amniotic fluid (if appropriate)
88240-Cryopreservation (if appropriate)
81265-Comparative analysis using Short Tandem Repeat (STR) markers; patient and comparative specimen (eg, pre-transplant recipient and donor germline testing, post-transplant non-hematopoietic recipient germline [eg, buccal swab or other germline tissue sample] and donor testing, twin zygosity testing or maternal cell contamination of fetal cells (if appropriate)

Reflex Tests:

Test ID	Reporting Name	CPT Units	CPT Code	Always Performed	Orderable Separately
CULAF	Amniotic Fluid Culture/Genetic Test			No	Yes
MATCC	Maternal Cell Contamination, B			No	Yes
CULFB	Fibroblast Culture for Genetic Test			No	Yes
_STR1	Comp Analysis using STR (Bill only)			No	No (Bill only)
_STR2	Add'l comp analysis w/STR (Bill Only)			No	No (Bill only)

Result Codes for Reflex Tests:

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULAF	52304	Result Summary	Alphanumeric		50397-9
CULAF	52306	Interpretation	Alphanumeric		69965-2
CULAF	52305	Result	Alphanumeric		82939-0
CULAF	CG767	Reason for Referral	Alphanumeric		42349-1
CULAF	52307	Specimen	Alphanumeric		31208-2
CULAF	52308	Source	Alphanumeric		31208-2
CULAF	52309	Method	Alphanumeric		85069-3
CULAF	54641	Additional Information	Alphanumeric		48767-8

Test ID	Result ID	Reporting Name	Type	Unit	LOINC®
CULAF	52310	Released By	Alphanumeric		18771-6
MATCC	53285	Result Summary	Alphanumeric		50397-9
MATCC	53286	Result	Alphanumeric		40704-9
MATCC	53287	Interpretation	Alphanumeric		69047-9
MATCC	53288	Reason for referral	Alphanumeric		42349-1
MATCC	53289	Specimen	Alphanumeric		31208-2
MATCC	53290	Source	Alphanumeric		31208-2
MATCC	55150	Method	Alphanumeric		85069-3
MATCC	53291	Released By	Alphanumeric		18771-6
CULFB	52327	Result Summary	Alphanumeric		50397-9
CULFB	52329	Interpretation	Alphanumeric		69965-2
CULFB	52328	Result	Alphanumeric		82939-0
CULFB	CG770	Reason for Referral	Alphanumeric		42349-1
CULFB	CG899	Specimen	Alphanumeric		31208-2
CULFB	52331	Source	Alphanumeric		31208-2
CULFB	52332	Method	Alphanumeric		85069-3
CULFB	54625	Additional Information	Alphanumeric		48767-8
CULFB	52333	Released By	Alphanumeric		18771-6

Reference Values:

An interpretive report will be provided.