Prader-Willi and Angelman Syndromes: Laboratory Approach to Diagnosis

Clinical suspicion of Prader-Willi or Angelman syndrome

- Order PWAS / Prader-Willi/Angelman Syndrome, Molecular Analysis (EDTA blood only)
- If not previously performed, order CMACB / Chromosomal Microarray, Congenital, Blood (EDTA and sodium heparin blood required)

- Abnormal methylation
- Deletion detected
- Interpretive report provided

- Abnormal methylation
- No deletion detected
- Interpretive report provided

- Abnormal methylation
- Duplication detected
- Interpretive report provided

- No abnormality detected on molecular or microarray analysis

To characterize disease mechanism, order UNIPD / Uniparental Disomy

- Interpretive report provided
- If clinical suspicion of Angelman syndrome remains, consider UBE3A gene sequencing, order UBE3Z / UBE3A Gene, Full Gene Analysis.