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
  - No deletion detected
    - Interpretive report provided
- Abnormal methylation
  - No deletion detected
    - Interpretive report provided
- No abnormality detected on molecular or microarray analysis
  - Interpretive report provided
    - If clinical suspicion of Angelman syndrome remains, consider UBE3A gene sequencing, order UBE3Z / UBE3A Gene, Full Gene Analysis.

To characterize disease mechanism, order UNIPD / Uniparental Disomy