Genetic Testing

UCSF Benioff Children's Hospital Oakland

Updated 12-19-17

Rapid FISH: probes for Trisomy 13, 18, 21, and X and Y (use when RAPID RESULTS are desired within 1-3 days)
- In EPIC, order **Aneuploidy Panel by FISH**
- Before sending a sample to the lab, alert lab customer service at x2079 that this sample is coming and it needs Priority handling due to very short specimen stability (only 48 hours).
- Send **2 mL (1 mL minimum)** blood in Sodium Heparin (green top). This amount cannot be combined with the Blood Karyotype order.
- Fill out this clinical history form http://ltd.aruplab.com/Tests/Pdf/20 or attach a clinical note so that it can be sent with the sample.

SNP array: evaluation for copy-number changes, uniparental disomy or parental consanguinity, triploidy, mosaicism, chimerism and for multiple anomalies (including DiGeorge)
-Send the SNP array and not the FISH for 22 q11 deletion; other syndromes with similar clinical features are missed by FISH analysis. Additionally, 22 q 11 deletion is found in only 50% of patients with complete DiGeorge
-In EPIC, order: **MISC lab test, and in comments add "Cytogenomic SNP array, collect 2 ml EDTA, approved by Dr. Cham"**

Microarray:
Note: We will no longer be ordering "Array, CGH/CMA" in Epic until the BCHO labs change to an updated platform.

Karyotype: use for trisomies, including those not detected by FISH probes, when rapid analysis is not required; karyotype testing may be necessary as follow up to microarray for an unbalanced translocation
- In EPIC, order: **chromosome analysis, blood/karyotype**

DNA methylation: Prader Willi
- In EPIC, order: **Prader Willi PCR,** and not Prader Willi FISH

Organic Acids, Urine
In EPIC, order: **organic acids, QL, urine**

DNA sequencing tests:
-In EPIC, order: **MISC lab test.** Approval by Dr. Cham is required.