Preparation of genomic DNA samples for Affymetrix Cytoscan HD Array Analysis

- The success of this assay requires the amplification of PCR fragments between 150 to 2000 bp in size throughout the genome. To achieve this, the genomic DNA must be of high quality, and must be free of contaminants that would affect the enzymatic reactions carried out.

- DNA must be free of PCR inhibitors: e.g heme and high concentrations of EDTA

- The genomic DNA extraction/ purification method should render DNA that is generally salt-free because high concentrations of certain salts can also inhibit PCR and other enzyme reactions.

- DNA must not be contaminated with other human genomic DNA sources, or with genomic DNA from other organisms. PCR amplification of the ligated genomic DNA is not human specific, so sufficient quantities of non-human DNA may also be amplified and could potentially result in compromised genotype calls.

- DNA must not be degraded: The approximate average size of genomic DNA may be assessed on a 0.8% or 1% agarose gel using an appropriate size standard control. High quality genomic DNA will run as a major band at approximately 10-20 kb on the gel.

1. Eppendorf Tube labeled with sample name on top and side of tube using permanent/alcohol proof marker (e.g. Sharpie)
2. Isolate DNA using a procedure with minimal salt and include an RNAse treatment.
3. Rehydrate isolated DNA with **1X Tris EDTA pH 8.0** (10mM Tris/0.1mM EDTA)
4. Preferred concentration of DNA is **50-100 ng/uL** in 1X TrisEDTA. **A total of 250 ng is the minimum sample amount for the assay. The lab requests 500 ng - 1 ug** to allow for pipetting error and quality control checks as needed.
5. Wrap each eppendorf tube with parafilm
6. Provide print out with sample name, DNA concentration, and A260/A280, especially important that we have the correct legible identifiers for coded samples.
7. The DNA must be high quality on quality check gel run with high MW ladder

8. Provide gel print out or email .tiff image
   sample lanes labeled and name of MW ladder
9. Do not freeze samples. Store 4C
10. Ship or deliver samples at 4C or room temp.