New Laboratory Offerings
See Molecular Laboratory Requisition Form

Female Infertility Panel:

- **Fragile X Associated Premature Ovarian Failure**: Approximately 1/180 females in the US are carriers of a premutation in the *FMR1* gene. Twenty percent of these women will develop POF while having none of the learning disability associated with the presence of the full mutation in that same gene. Thus women with POF may be at risk of carrying the premutation gene and also be members of a family at risk for Fragile X mental retardation. Approximately 7% of all women with POF have been found to carry a premutation allele. **Requires whole blood in EDTA (purple top)**
- **Chromosome Analysis**: Infertility may be caused by the presence of chromosome rearrangements that lead to failure of egg development. **Requires whole blood in sodium heparin (green top)**
- **Chromosome Mosaicism**: This test is indicated when mosaicism, the presence of two or more cell lines with different chromosome patterns, is suspected based upon clinical findings. Infertility may be caused by mosaicism for chromosome abnormalities in a small percentage of cells that would not be detected in the usual chromosome analysis. **Will use sample from chromosome analysis above**

Male Infertility Panel: for males with oligospermia or azoospermia

- **Y Deletion:** Molecular analysis for possible deletions within the Azoospermia Factor (AZF) loci on the long arm of the Y chromosome. Mutations within this region have been associated with male infertility and several of the genes within this interval encode key spermatogenesis-related genes. NOTE: not all males with abnormal sperm production have disturbances of these genes. **Requires whole blood in EDTA (purple top).**
- **CFTR Mutation Analysis:** Congenital bilateral absence of the vas deferens is associated with the presence of mild CFTR mutations. **Will use same sample as the Y deletion analysis above.**
- **Chromosome Analysis:** Infertility may be caused by the presence of chromosome numeric abnormalities of rearrangements that lead to failure of sperm development. **Requires whole blood in sodium heparin (green top).**

Fragile X Associated Ataxia/Tremor: Approximately 1% of males and a smaller percentage of women presenting with late onset progressive tremor /ataxia have been found to carry a premutation in the *FMR1* gene. This syndrome presents in males as early as the 5th decade and in women in the 6th decade. These symptoms progress to significant ataxia and other neurological symptoms over the next two decades. Approximately 1/400 males and 1/180 females are premutation carriers. Fifty percent of these carrier males and a much smaller percentage of carrier females will display features of this disorder by the time they are in their 7th decade. **Requires whole blood in EDTA (purple top).**

FLT3 and NPM1: The presence of mutation in one or both of these genes provides prognostic information useful in treatment option decisions for patients with AML who have normal cytogenetic analysis. **Requires whole blood or bone marrow in EDTA (purple top).**