NORTH YORK GENERAL

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GENETICS PROGRAM
MOLECULAR GENETICS LABORATORY

TESTING CURRENTLY AVAILABLE

Aneufast for chromosomes 13, 18, 21, X and Y aneuploidies
Dentatorubral Pallidoluysian Atrophy (DRPLA)
Familial Thrombophilia: Factor V Leiden and Factor II c.*97G>A
FMR1-related disorders:

- Fragile X syndrome (FXS)
- Fragile X-associated Tremor/Ataxia syndrome (FXTAS)
- Fragile X-associated Premature Ovarian Insufficiency (FXPOI)

Hereditary Breast and Ovarian cancer:

- Ashkenazi Jewish Founder panel
- Familial variants (including deletion/duplication)
- Comprehensive Hereditary Cancer Panels (including deletion/duplication):
  - Comprehensive Breast AND Ovarian Cancer (18 gene panel)
    ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11 & TP53
  - Comprehensive Breast Cancer (11 gene panel)
    ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, PALB2, PTEN, STK11 & TP53
  - High Risk Breast and Ovarian Cancer (*BRCA1* and *BRCA2* genes only)

Friedreich Ataxia (FRDA)
Hemochromatosis: H63D and C282Y
Huntington Disease (HD)
Infertility

- Y Chromosome Microdeletion

Identity:

- Maternal Cell Contamination
- Specimen Misidentification

Microarray

- Postnatal SNP array
- Prenatal SNP array
- Oncology SNP array

Oculopharyngeal Muscular Dystrophy (OPMD)
Spinal and Bulbar Muscular Atrophy (SBMA)
Spinocerebellar Ataxia 1 (SCA1)
Spinocerebellar Ataxia 2 (SCA2)
Spinocerebellar Ataxia 3 (SCA3)
Spinocerebellar Ataxia 6 (SCA6)
Spinocerebellar Ataxia 7 (SCA7)
Spinocerebellar Ataxia 8 (SCA8)
Spinocerebellar Ataxia 17 (SCA17)
Uniparental Disomy Chromosome 14 (UPD 14)
Uniparental Disomy Chromosome 15 (UPD 15)