

## National Centre for Medical Genetics Summary of services available

www.genetics.ie

Molecular Genetics (DNA) Laboratory Tel: (01) 409 6733

## Molecular Genetic (DNA) tests

Tests currently provided in-house:

:

- Angelman Syndrome
- Familial Breast Cancer
- Cystic Fibrosis
- Duchenne/Becker Muscular Dystrophy
- · Torsion Dystonia, early onset
- Fragile X syndrome (including premature ovarian failure, Fragile X tremor-ataxia syndrome)
- Friedreich Ataxia
- Hereditary Non-polyposis Colorectal Cancer
- Huntington Disease
- Microsatellite Instability
- Prader Willi Syndrome
- Spinal Muscular Atrophy
- Uniparental Disomy

Hereditary Haemochromatosis testing is not available at NCMG.

We accept samples for all other molecular genetic tests and will send them out to reputable laboratories; invoices from the external laboratory will go directly to the referring clinician or institution.

Please refer to our website <u>www.genetics.ie</u> for further information.

Cytogenetics (Chromosome) Laboratory Tel: (01) 409 6970

## Constitutional (Chromosomes + FISH)

Tests currently provided in-house:

- New-borns and children (<5 yrs old) for chromosome analysis
- Microdeletion syndromes for FISH-only analysis (no age restrictions)
- On-going family studies

Prenatal samples (CVS and/or amniotic fluid) are accepted as normal.

## Oncology

The service provided remains a National service with limited sample acceptance restrictions.

Bone marrow and peripheral blood samples referred to the NCMG for haematological disorders are accepted .Information regarding bone marrow morphology and/or confirmed disease diagnosis will be required before analysis is undertaken.

Please refer to our website <u>www.genetics.ie</u> and leaflet.

Valid from July 2011, until further notice

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