Chromosome Instability Disorders

Genetics

Chromosome Instability Disorders

Methodology	Spontaneous and mutagen induced chromosome breakage, G-banded chromosome analysis, Sister Chromatid Exchange analysis
Address to send samples	**Genetics Laboratory** **Viapath** **5th Floor Tower Wing** **Guy's Hospital** **Great Maze Pond** **London SE1 9RT**
Service Delivery Manager	**Richard Hall** Richard.hall@viapath.co.uk Tel: 020 7188 1709
Download referral form at:	http://www.viapath.co.uk/cytogenetics-laboratory.html
Samples accepted	**Blood samples (lithium heparin), amniotic fluid, chorionic villus biopsy and solid tissue samples**
Additional information/ special sample instructions	For sample and shipping information please contact the laboratory
Turnaround time	Blood samples -10 days, solid tissue - 28 days, prenatal samples - 14/28 days
Arrange in advance	**For new customers, please contact the laboratory prior to sending samples**
Cost	On application (discounts could be available for significant workloads)

The Genetics Department at Guy's Hospital is at the forefront of developing, validating and introducing the latest genetic techniques and has a proven track record for diagnostic service development leading to improvements in patient care. We have an extensive repertoire of genetics tests and are committed to providing a high-quality, cost-effective diagnostic service to patients.

Viapath provide a high quality clinical diagnostic service, offering one of the most comprehensive specialist pathology test portfolios in the UK across all pathology disciplines : **Genetics, Biochemistry, Haematology, Histopathology, Immunology, Microbiology** & **Virology**. Our recent major re-development programme included investment in a state-of-the-art Blood Sciences Laboratory on our King's College Hospital site, one of the largest integrated automated laboratories in Europe. For more information about our full range of specialist tests, consult our websites at **www.viapath.co.uk**

We are part of **King's Health Partners**, one of the UK's first **Academic Health Sciences Centres**.

Quality Accreditations
All our laboratories are accredited to CPA standards and JACIE standards where applicable. Our pathology services are regulated and licensed by the Human Tissue Authority (HTA) and the Medicines and Healthcare products Regulatory Agency (MHRA).

For more information on how we can support all your specialist & routine pathology needs, consult our websites, contact Senior Business Develoepmnt Manager **John Roberts** on **07766 205 577**, or E-mail **john.roberts@gstt.nhs.uk.**

Viapath
St. Thomas' Hospital,
Westminster Bridge Road,
London SE1 7EH

High quality, clinically excellent, cost effective pathology services.

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Our genetics laboratory is a supra-regional and international reference laboratory for chromosome instability testing by cytogenetic techniques: our experience encompasses the full range of classical and atypical cases reported in the literature. Specific testing strategies for the different disorders are employed to look for raised spontaneous and specific mutagen-induced damage and/or chromosome rearrangements or other anomalous behaviour. We offer both pre- and postnatal diagnosis on amniotic fluid, chorionic villi, blood and solid tissue samples.

Fanconi anaemia

The test detects defective DNA repair in response to alkylating agents by screening for increased spontaneous and mutagen induced chromosome breakage. The primary mutagen used is Diepoxybutane (DEB) which we find gives the best discrimination between affected and unaffected individuals but Mitomycin C (MMC) testing is available on request.

Radiosensitivity syndromes

Ataxia-telangiectasia and Nijmegen breakage syndrome patients are defective for repair of ionising radiation induced damage. Raised spontaneous and gamma ray induced chromosome breakage is screened for and G-banded metaphase preparations are examined for clonal chromosome rearrangements, particularly involving the immunoglobulin genes on chromosomes 7 and 14.

Other rare syndromes:

Bloom syndrome: This test looks for a greatly increased frequency of sister chromatid exchanges (SCEs) in cultured cells.

Roberts syndrome (SC Phocomelia): Screening cultured cells for premature centromere separation and the presence of random aneuploidy.

Premature chromosome condensation: An increased incidence of prometaphase cells is indicative of the presence of a mutation in the microcephalin gene MCPH1 and identifies patients for specific mutation testing.

ICF syndrome: Immunodeficiency, Centromeric region instability, and Facial anomalies syndrome is characterised by deletions and rearrangements involving the centromeric heterochromatic region of chromosomes, particularly chromosomes 1, 9, and 16, which can be screened for in mitogen-stimulated lymphocyte cultures.

Other syndromes: We can perform cytogenetic screening for raised levels of spontaneous chromosome breakage, chromosome rearrangements and aneuploidy. This may be of benefit if rare instability syndromes such as **Werner syndrome** and **variegated aneuploidy syndrome** are suspected, or if a defect in chromosome repair or processing is suspected in a patient with unusual presentation.
Please see separate information leaflets for details of other pre- and postnatal genetic tests.

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