Testing for Genetic Disorders

Methodology	QF-PCR, aCGH, G-banded chromosome analysis, FISH, Sanger and Next Generation Sequencing
Address to send samples	**Genetics Laboratory** **GSTS Pathology** **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.gsts.com/cytogenetics-laboratory.html
Samples accepted	**Blood samples in EDTA and/or lithium heparin**
Additional information/ special sample instructions	For sample and shipping information please contact the laboratory
Turnaround time	QF-PCR - 3 working days, aCGH - 21 days, G-banded chromosome analysis - 14 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.

KingsPath & **GSTS** 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 redevelopment 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 website 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 Develpemnt Manager **John Roberts** on **07766 205 577**, or E-mail **john.roberts@gstt.nhs.uk.**

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Our regional genetics laboratory uses state of the art molecular cytogenetic and molecular techniques for detection of genetic imbalance and diagnosis of monogenic disease in children and adults with developmental disability, dysmorphism, congenital abnormality or specific syndromes.

Cytogenetic investigations:

- **QF-PCR** (quantitative fluorescence PCR) for the rapid diagnosis of neonates or children with a suspected common chromosome aneuploidy syndrome (Down, Edwards, Patau, Turner and Klinefelter)
- **Array CGH** (array comparative genomic hybridisation) for high resolution analysis for imbalance across the genome, recommended as a first test in place of traditional G-banded chromosome analysis. We were the first centre in the UK to introduce array CGH as a first line test, and thus have a wealth of experience in interpreting and reporting the results; our innovative hybridisation and analysis strategies mean that our prices are very competitive.
- **Karyotyping** (G-banded chromosome analysis) **and FISH** for characterisation of chromosome abnormalities detected by other cytogenetic techniques and follow-up to establish recurrence risks.

Molecular genetic investigations:

Postnatal testing for a wide range of specific monogenic diseases, using appropriate technologies, including next generation sequencing for detection of mutations.

Please see separate information leaflets for details of chromosome instability testing, prenatal diagnosis and subfertility investigations.

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Genetics