Who we are

Majority owned by the NHS, but with the commercial freedom to invest in innovation, Viapath are on a mission to transform pathology services in the UK. We provide pathology services to the NHS, private hospitals and other organisations both across the country and internationally.

What we do

All our laboratories are either accredited or working towards accreditation by UKAS to ISO15189. To view our laboratory accreditation status please follow this link:

http://www.viapath.co.uk/about-viapath/quality-and-governance/accreditations

TEST OVERVIEW

Description
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: Ataxiatelangiectasia 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.

Clinical details
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.

Related condition or disease
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.

Synonyms or keywords
Chromosome breakage, chromosome instability, Fanconi anaemia, Diepoxybutane, DEB test, Mitomycin C, MMC, Radiosensitivity syndromes, Ataxia-telangiectasia, Nijmegen breakage syndrome, Bloom syndrome, Roberts syndrome, SC Phocomelia, Premature chromosome condensation, MCPH1, ICF syndrome, Werner syndrome, variegated aneuploidy syndrome

Department
Genetics Department

Laboratory
Cytogenetics Laboratory at Guy's

Location
Viapath at Guy's Hospital

ORDERING INFORMATION

Sample type and Volume required
Peripheral blood in a lithium heparin bottle, 5ml (2ml from babies). Amniotic fluid and solid tissue specimens in a dry

Storage and transport
Samples should arrive within 72 hours (preferably24 hours) of sampling. For prenatal diagnosis please discuss with the
sterile container (20ml), CVS (20mg please discuss with the laboratory). Cultured fibroblast cells are also accepted.

Call in advance
Please notify the laboratory before sending samples

Turnaround time
Blood samples - 10 days, solid tissue - 28 days, prenatal samples - 14/28 days

laboratory before sending any samples.

Cost
On application

Time limit
Do not spin down or freeze samples before sending.
How can we help?

We have a number of partnering options to suit your needs, whether you require this specific test or a range of services, we are here to help. Contact one of our friendly Business Development Managers for more information, or visit our website.