Bristol Genetics Laboratory

North Bristol NHS Trust

Chromosome Breakage Disorders Cytogenetics

(See also chromosome breakage disorders NGS)

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Sample Required:

Adult: 3-5ml blood / bone marrow in lithium heparin.
Paediatric: at least 1ml lithium heparin (preferably >2ml)

Prenatal testing MUST be arranged with the laboratory well in advance.

Samples should be accompanied by a FULLY completed request form (available as download at www.nbt.nhs.uk/genetics or from the laboratory).

Please include details of test, family history, address and POSTCODE, NHS number, referring clinician and unit/hospital.

Consent and Storage:

All genetic testing requires consent. It is the responsibility of the referring clinician to ensure that appropriate consent has been obtained.

Stored material from all referrals may be retained for quality assurance purposes and may be used anonymously for the development of new tests for the disorder in question.

Clinical Background and Genetics

Fanconi Anaemia

Clinical features All marrow elements are usually affected, resulting in anaemia, leukopenia and thrombopenia. Also noted are pigmentary changes in the skin and malformations of the heart, kidney, and limbs(aplasia of the radius, thumb deformity).

Diagnostic method Increased levels of chromosome damage are found when cultures are exposed to alkylating agents (DEB is used at this lab).

Ataxia Telangiectasia

Clinical features Progressive cerebellar ataxia, telangiectases especially of the conjunctiva, and proneness to sinopulmonary infection.

Diagnostic method Increased levels of chromosome damage are evident when exposed to the radiomimetic drug Bleomycin. Also increased spontaneous levels of T- cell receptor rearrangements are found on chromosomes 7 and 14.

Nijmegan Breakage Syndrome

A rare disorder with a combination of both Ataxia telangiectasia and Fanconi clinical and cytogenetic features

Quality

BGL participates in the UKNEQAS scheme for Fanconi and has UKGTN approval for the Fanconi and Ataxia Telangiectasia service.

Reports are usually available within 28 days.

For up-to-date prices please contact the laboratory

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