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.

Nijmegen 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