Cockayne Syndrome (CS)

Clinical Background and Genetics
Cockayne Syndrome (CS) is an autosomal recessive neurodegenerative disorder characterised by progressive growth failure, microcephaly, mental retardation, retinal degeneration, sensorineural deafness, and photosensitivity. CS has a variable rate of progression. The spectrum of CS phenotypes can be divided into three general clinical presentations:

Cockayne Syndrome type I: "Classic" CS (early-onset) in which the major features of the disease become apparent by one or two years of age. This is the most common type.

Cockayne Syndrome type II: A more severe and less common form of the disorder with abnormalities recognised at birth or in the early neonatal period.

Cockayne Syndrome type III: Milder/later-onset forms that are still poorly defined.

Cockayne Syndrome has been associated with mutations in two genes; ERCC6 (also known as CSB) and ERCC8 (also known as CSA or CKN1).

Service offered
- Full gene screening of ERCC6 by direct sequence analysis, this includes all 21 exons (includes non-coding exon 1) and extends to the upstream putative branch site and downstream splice sites. 75% of mutations of CS mutations are identified in ERCC6.
- Full gene screening of ERCC8 by direct sequence analysis (for patients negative after ERCC6 analysis), this includes the coding exons (1-12) and extends to the upstream putative branch site and downstream splice sites. 25% of mutations of CS mutations are identified in ERCC8.
- Familial tests for known mutations.
- The ERCC6 and ERCC8 genes are also included in a 62 gene panel for chromosome breakage syndromes. This panel also includes the genes ERCC1, ERCC2 and ERCC5 which have been implicated with disorders phenotypically similar to Cockayne Syndrome. Please contact the laboratory for further information on this service.

Quality
- This is a UKGTN approved service. BGL participates in the EMQN scheme for DNA sequencing.

Referrals
- Referrals are accepted nationally from clinical Geneticists only. Please contact the laboratory for up to date prices.

Target reporting Time
- Diagnostic screen of each gene 56 days
- Gene panel testing 112 days
- Known Mutation 14 days
- Screening for 2 known mutations 14 days
- Prenatal/urgent 3 days

References