Introduction
Achondroplasia (MIM 100800) and hypochondroplasia (MIM 146000) are autosomal dominant skeletal disorders with mutations in the FGFR3 gene on chromosome 4p16.3.

Achondroplasia (ACH) has a birth incidence of between 1/15,000 and 1/77,000. Around 80-90% of cases are sporadic and there is an association with increased paternal age at the time of conception, suggesting that new mutations are generally of paternal origin. There are rare familial forms, as well as reported cases of germline and somatic mosaicism.

Hypochondroplasia (HCH) is genetically distinct from ACH and is clinically less severe, with no associated craniofacial abnormalities. Because of its mild nature, HCH can be difficult to diagnose and may be genetically heterogeneous. Approximately 70% of HCH patients have one of two mutations in the FGFR3 gene. Of the remaining 30%, some families are reported that do not link to chromosome 4p16.3

Referrals
We offer testing for confirmation of diagnosis in affected individuals and family members.

Prenatal testing
1) Prenatal testing is available to families in whom specific mutations have been identified - please contact the laboratory to discuss.
2) Prenatal testing to confirm a diagnosis of ACH suspected on antenatal ultrasound scan

Service offered
Achondroplasia: Testing for the common p.Gly380Arg (c.1138G>A and c.1138G>C) mutations in exon 8 of FGFR3. Together these account for around 99% of mutations.

Hypochondroplasia: Mutation screening of exons 6, 8, 11 and 13 of FGFR3 will detect approximately 75% of reported mutations. Includes testing for the common p.Asn540Lys (c.1620C>A and c.1620C>G) mutations in exon 11 (which account for around 70% of mutations) and the common achondroplasia p.Gly380Arg (c.1138G>A and c.1138G>C) mutations.

Technical
Direct sequence analysis of exons 8 (ACH) and 6, 8, 11 and 13 (HCH) detects the common mutations. This will also detect other mutations that may be present in these exons.

Target reporting time
2 weeks for routine ACH analysis and 4 weeks for routine HCH analysis. For urgent samples please contact the laboratory.
Patient details
To facilitate accurate testing and reporting please provide patient demographic details (full name, date of birth, address and ethnic origin), details of any relevant family history and full contact details for the referring clinician.