

## Molecular Genetics Service Profile Achondroplasia (ACH)

### Introduction

- ◇ ACH (MIM 100800) is an autosomal dominant disease affecting 1 in 15,000 newborns. It is characterised by severe dwarfism with marked shortening of the proximal part of the limbs, macrocephaly with facial dysmorphism and lumbar lordosis. Radiographic features include square aspect of iliac bones, short femoral necks, widening of tibial metaphyses and fibulae longer than tibiae.
- ◇ ACH is caused almost exclusively (more than 99% of cases) by a recurrent mutation (G380R) in the transmembrane domain of Fibroblast Growth Factor Receptor 3 (FGFR3), (Rousseau *et al.*1994).

Contact details for the laboratory carrying out the genetic test for ACH  
**Unité INSERM 781, Hôpital Necker-Enfants Malades, 149 rue de Sèvres, 75743 Paris, Cedex 15, France.**  
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### Reasons for referral

- ◇ Mutation screening in patients with clinically confirmed or suspicion of ACH (sporadic or familial case).
- ◇ Prenatal diagnosis is **not** offered for ACH

### Samples

- ◇ Minimum 5µg of DNA from peripheral blood from your local laboratory. Blood samples (minimum of 5mls in EDTA) can also be sent to our laboratory by express mail (FedEx / UPS). Please contact our laboratory (as above) for further details.

### Technical

- ◇ Mutation analysis by direct bi-directional sequencing of exon 10 of *FGFR3*.

### Target turn-round time

- ◇ Routine analysis to confirm clinical diagnosis - 15 days. Turn-round times are from the receipt of all required samples and information, including appropriate clinical information and radiographs. Relevant clinical-radiographic expertise is currently offered at no cost through the use of the secure online submission system (the **ESDN Case Manager**). Testing is only performed after clinical and radiographic evidence has been reviewed using the **ESDN Case Manager**. To obtain a username and password for the **ESDN Case Manager** please email [info@esdn.org](mailto:info@esdn.org).

### Cost

- ◇ Full mutation screen - €100.

### References

- ◇ Rousseau F. *et al.* (1994). *Nature* **371**: 252-254.

### ESDN Administrator contact details

- ◇ Email: [info@esdn.org](mailto:info@esdn.org) Website: [www.esdn.org](http://www.esdn.org)

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