Molecular Genetics Service Profile
Nail-Patella Syndrome (NPS) or Osteoonychodysplasia

Introduction

◊ NPS (MIM 161200) is an autosomal dominant condition, characterized by dysplasia of nails, absence or hypoplasia of patellae, elbow deformity, eventually short stature, scoliosis and other musculoskeletal abnormalities. In some cases NPS presents with nephropathy of variable severity. Glaucoma and other ocular symptoms may also be part of the syndrome.

◊ Radiographic findings include iliac horns (triangular bony protuberances of the posterior ilium), hypoplasia of radial heads and/or capitella with subluxation or dislocation, absent or hypoplastic patellae.

◊ NPS is caused by mutations in the *LMX1B* gene. One of the essential features of the transcription factor encoded by *LMX1B* is involvement in the regulation of limb development.

Contact details for the laboratory carrying out the genetic test for NPS
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Reasons for referral

◊ Mutation screening for confirmation of a clinically/radiographically/histologically suspected diagnosis of NPS and to differentiate NPS from other similar syndromes e.g. with hypoplasia-dislocation of patellae.

◊ Prenatal diagnosis is not offered for NPS.

◊ Presymptomatic and carrier testing is not applicable.

◊ Screening for unknown mutations is labour intensive, therefore we cannot accept urgent referrals of this type.

Samples

◊ Minimum 100 μg of DNA from peripheral lymphocytes from your local laboratory. Blood samples (minimum of 5 mls in EDTA) can also be sent to our laboratory by express mail (FedEx / UPS).

Technical

◊ Mutation scanning of 8 *LMX1B* exons by fluorescent bi-directional sequencing (8 PCRs, 8 bi-directional sequencing steps).

Target turn-round time

◊ Mutation scanning of 8 exons - 10 weeks. Routine, single mutation test - 3 weeks. Urgent, single known mutation test 1 - 2 weeks. 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.

Cost

◊ Full mutation screen - €660.

References

◊ Dreyer *et al*. (1998) *Nat Genet* **19**: 47-50.

◊ McIntosh *et al*. (1998) *Am J Hum Genet* **63**: 1651-1658.

◊ Hamlington JD, Jones C, McIntosh I (2001) *Hum Mutat* **18**:453.

◊ Bongers EM, Gubler M-C, Knoers NV (2002) *Pediatr Nephrol* **17**:703-712.

ESDN Project Administrator contact details

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