

## Molecular Genetics Service Profile Marshall Syndrome

### Introduction

- ◇ Marshall Syndrome (MIM 154780) is an autosomal dominant disorder characterized by sensorineural hearing loss, high myopia, vitreoretinal degeneration, retinal detachment, cleft palate, mid-facial hypoplasia, flat nasal bridge, short nose, anteverted nostrils, long philtrum, short to normal stature.
- ◇ ≥ 50 % of Marshall Syndrome cases are caused by mutations in *COL11A1*. Possible other loci are not known.
- ◇ Genotype-phenotype correlations: Splicing mutations in *COL11A1* that involve 54-bp exons in the 3'-half of the gene result in typical Marshall phenotypes. Other *COL11A1* mutations can cause more Stickler-like phenotypes. The major differences between the two phenotypes are: Hearing deficit, facial features and short stature are more common in Marshall syndrome. Eye findings are usually more severe in Stickler than in Marshall syndrome (Annunen *et al.* 1999, Majava *et al.* 2007).

Contact details for the laboratories carrying out the genetic test for Marshall Syndrome  
Dept of Medical Biochemistry and Molecular Biology, University of Oulu, Aapistie 7 90220 Oulu, Finland.  
<http://www.biocenter.oulu.fi/projects/mannikkoalakokko.html>  
Dr. Minna Männikkö Tel: +358 8 5375751. Fax: +358 8 537 5811.  
Email: [minna.mannikko@oulu.fi](mailto:minna.mannikko@oulu.fi)

### Reasons for referral

- ◇ Mutation analysis in patients for confirmation of a clinically suspected diagnosis of Marshall Syndrome. Screening for unknown mutations is labour intensive, therefore we cannot accept urgent referrals of this type.
- ◇ Evaluation of at-risk relatives for management reasons and genetic counselling. In this case the mutation in the index case must be known.
- ◇ Prenatal diagnosis is not offered.

### Samples

- ◇ Minimum 100µg of genomic DNA from peripheral lymphocytes (or cultured cells) from your local laboratory. Blood samples (minimum of 5ml in EDTA) can also be sent to our laboratory by express mail (DHL/FedEx/TNT/UPS).

### Technical

- ◇ Mutation scanning of exons 1-68 of *COL11A1* by fluorescent bidirectional sequencing.

### Target turn-round time

- ◇ Mutation scanning of *COL11A1* – 20 weeks. Routine, single mutation test - 4 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](mailto:info@esdn.org).

### Cost

- ◇ Full mutation screen €2500.

### References

- ◇ Annunen *et al.* (1999) *Am J Hum Genet* **65**: 974-983, Majava *et al.* (2007) *Am J Med Genet A* **143**:258-264.

### ESDN Administrator contact details

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

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