EXT1 and EXT2 Gene testing at the Department of Medical Genetics, University of Antwerp (Belgium)

Mutation analysis of the EXT1 and EXT2 genes will be performed by direct sequencing of all coding exons of both genes, including intron/exon boundaries. This results in the identification of a mutation in approximately 80% - 90% of the patients. Additional analysis to detect possible deletions involving the EXT1 or EXT2 gene will be performed by PCR analysis of intragenic EXT1 and EXT2 markers or FISH analysis (optional). Reporting of the results is expected within 2-4 months.

Costs:

Initial screening: 600 euro/sample for each gene analyzed. Normally both genes will be analyzed simultaneously. However, at request the analysis of only one gene or both genes stepwise is possible. If blood (heparin) is sent, FISH analysis is also performed.

Confirmation: 300 euro/sample for screening for a known mutation. Detailed information of the exact position of the mutation should be provided. Inclusion of a control sample is preferred.

Prenatal diagnosis: 600 euro/diagnosis. Prenatal diagnosis on chorion villi (CVS) must be announced in advance and can only be performed if adequate information is available. The lab should always be contacted before sending a CV sample. Maternal material (DNA or blood) is also required to test for possible maternal contamination (no additional cost).

FISH: 300 euro/ sample

Linkage analysis: If multiple members of a family are available linkage analysis can be performed to exclude/link one of the EXT genes. If this analysis is performed coupled with full mutation screening no additional cost will be charged and costs will thus be limited to 600 euro (index patient) and 300 euro for each additional family member that must be genotyped. It is possible to include individuals in the linkage analysis without genotyping them once the mutation has been identified. This should clearly be indicated at the request form. These individuals will not receive a result or invoice.

An institutional invoice address for each sample should be provided or otherwise a check in euro made payable to Department of Medical Genetics University Hospital of Antwerp should be included when sending the sample. We do not send invoices directly to the patient.

Universiteitsplein 1 2610 Antwerpen (BELGIE) Tel:+32(0)3 820 25 70 Fax:+32(0)3 820 25 66 http://www.uia.ac.be/dnalab

Diensthoofd : Prof. Markus NÖTHEN, MD
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Cytogenetica : Bernadette VAN ROY, MSc, Jan WAUTERS, PhD
DNA-onderzoek : Katrien STORM, MSc, Wim WUYTS, PhD, Guy VAN CAMP, PhD, Wim VAN HUL, PhD, Frank KOOY, PhD, Sven CICHON, PhD

Sample:

DNA analysis requires blood (20 ml – EDTA or heparin, but please include at least one tube heparin) or DNA (50 µg). For FISH analysis we need 10 ml (HEPARIN).

All samples should be sent to

Wim Wuyts, PhD
Dept. Medical Genetics
University of Antwerp
Building T (6th floor)
Universiteitsplein 1
2610 Wilrijk
Belgium

Samples should be shipped at room temperature and should arrive in our lab preferable within 48 hours. It is advised to contact the lab before sending any samples.

Requests/Reports

All requests for EXT mutation analysis should be sent by a genetic counselor or clinician with appropriate genetic knowledge with respect to multiple exostoses in order to ensure proper correspondence of the results to the patient. Reports will only be sent to the referring clinician or genetic counselor. No results will be mailed to the patient.

Clinical information

Clinical information is required. Please fill in attached clinical sheet for each patient

For additional questions, please contact Wim Wuyts.

Wim Wuyts, PhD

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MULTIPLE OSTEOCHONDROMAS (MO)

CLINICAL INFORMATION

- Name patient:
- Date of birth:
- sex:
-
- Height : at age:
- Age of onset of MO:
- Number of osteochondromas at ..... (age) years (please circle) :

1) 1
2) 2 to 5
3) 5 to 10
4) 10 to 20
5) >20

- Site of osteochondroma (please tick):

Site		Site		Site	
distal femur		distal tibia		foot	
proximal femur		proximal tibia		knee	
distal humerus		distal fibula		scapula	
proximal humerus		proximal fibula		clavicle	
pelvis		spine		other:.....	

- Did the patient develop a chondrosarcoma? no
  yes at age: .....
  location: .....

- Family history: 1) no family history
  2) family history: please include pedigree

MULTIPLE OSTEOCHONDROMAS (MO)

CLINICAL INFORMATION (2)

- Name patient:

- skeletal deformities:

1) no
2) yes: please specify:

Deformity		Functional impairment	
forearm		decreased range of forearm rotation	
forearm with radial head dislocation		decreased range of elbow flexion	
shortening of forearm		decreased range of knee flexion	
genu vaga		other:	
other:.....			

- complications (vessel entrapment, tendon entrapment.....):

1) no
2) yes: please specify: .....

.....

- additional comments/observations