

FG syndrome (Opitz-Kaveggia syndrome)

OMIM: 305450

Gene: MED12

Locus: Xq13

OMIM: 300188

SERVICE: mutation analysis of exon 21 of MED12 gene

TESTING: **Diagnostic*:** clinically affected males
Presymptomatic: male relatives patients at risk of developing FG (known mutation)
Carrier: female relatives of clinically affected patients (known mutation)
Prenatal: at risk of having an affected male (known mutation)
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Clinical Geneticists only**
The laboratory does NOT accept referrals directly from patients

TARGET REPORTING TIME AND COSTS

(Non UK National Health Service patients are subject to a surcharge. Payment must be agreed prior to testing – please include invoice form A)

Diagnostic:	8 weeks	£99 (sequencing one exon)
Presymptomatic/Carrier:	2 weeks	£145
Prenatal	3 days	£350 (including maternal contamination studies)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exon 21 of the MED12 gene
- There is a single recurrent mutation (c.2881C>T; p.Arg961Trp) which accounts for 10% of the cases of FG syndrome

SAMPLE REQUIREMENTS

- 1-5ml blood in EDTA or 50ul DNA (concentration ~500ng/ul)
- All patient samples must be labelled with **name, date of birth and Hospital/NHS number**
- Samples should be accompanied by a FULLY completed request card (available from the laboratory)
- Please include details of test, family history, patient address & postcode, GP, referring clinician and unit/hospital
- **Samples and paperwork must include three unique and matching patient identifiers**

SHIPPING DETAILS

- DNA can be sent by first class post
- Blood must be appropriately packaged and preferably sent by courier to arrive as soon as possible
- Do not freeze prior or during postage

CONSENT

It is the responsibility of the referring clinician to ensure consent has been obtained for:

- testing and storage
- the use of the sample and the information generated from it to be shared with members of the patients family and their health professionals

After testing, part of this sample might be used anonymously for the development of new tests and to monitor the quality of laboratory results.

CONTACT DETAILS

Genetics Laboratories, Box 143
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Cambridge CB2 0QQ
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Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

UKGTN testing criteria

UK Genetic Testing Network

Name of disease(s): FG (Opitz-Kaveggia) syndrome
Name of gene(s): MED12

Patient name:	Date of birth:
Patient postcode:	NHS number:
Name of referrer:	
Title/Position:	
Department/Hospital:	
Contact email/telephone number:	

Referrals will only be accepted from one of the following:
(Please indicate with a tick which category refers to the referrer).

Referrer	Tick if this refers to you.
Consultant Clinical Geneticist	

Minimum criteria required for testing to be appropriate:

Criteria	Tick if this patient meets criteria
Male	
6 of the 8 criteria below:	
• Mental retardation	
• Hypotonia	
• Small and simple ears	
• Constipation and/or anal anomalies	
• Tall and prominent forehead	
• Downslanting palpebral fissures	
• Broad thumbs and halluces	
• Abnormalities of the corpus callosum	
Additional features that may be present :	
• Characteristic behaviour (friendly, hyperactive, attention-seeking)	
• Frontal hair upsweep	
• Relative macrocephaly	
• Ocular hypertelorism	
• Family history consistent with X-linked inheritance	

If the patient does not fulfil these criteria and you still feel that testing should be performed please contact the molecular genetics laboratory