

Gitelman syndrome (GMS) **OMIM: 263800**
Gene: SLC12A3 **Locus: 16q13** **OMIM: 600968**

SERVICE: **mutation and dosage analysis of the SLC12A3 gene**

TESTING: **Diagnostic:** **clinically affected patients**
Presymptomatic: **patients at risk of developing GMS (known mutations)**
Carrier: **relatives of clinically affected patients (known mutations)**
*samples will only be accepted with a completed 'testing criteria' form (see attached)

REFERRALS: **Clinical Geneticists, Renal Physicians & Endocrinologists 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	£625 (sequencing and dosage)
Presymptomatic/Carrier:	2 weeks	£175 (sequence two exons)

TECHNICAL INFORMATION

- PCR and fluorescent sequence analysis of exons 1-26 and splice site boundaries of the SLC12A3 gene
 - Multiplex ligation dependent probe amplification analysis of exons 1-26 of the SLC12A3 gene (excluding exon 6)
- Point mutations or small insertions/deletions are responsible for around 99% of cases.

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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Addenbrooke's Hospital
Cambridge CB2 0QQ
Tel: +44 (0) 1223 348866
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Website: www.cuh.org.uk/genetics-labs



Accredited Medical Laboratory
Reference No: 1275

Cambridge testing criteria

Name of disease(s): **Gitelman syndrome (GMS)**

Name of gene(s): **SLC12A3**

Patient name:

Date of birth:

Patient postcode:

NHS number:

Name of referrer:

Title/Position:

Department/Hospital:

Contact email/telephone number:

Referrals only will 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 Renal Physician	
Consultant Endocrinologist	
Consultant Clinical Geneticist	

Minimum information required for testing to be appropriate:

Criteria	Tick if this patient meets criteria or give values
1. Pretreatment hypokalaemia with renal K wasting	
2. hypomagnesaemia	
3. metabolic alkalosis	
4. hypocalciuria	
5. renin level (w/ normal ranges please)	
6. aldosterone level (please state if K low)	
7. Result of diuretic screen if performed	
8. Ultrasound result	

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