Renal panel for Steroid Resistant Nephrotic Syndrome (SRNS), Alport syndrome and rare inherited renal disease

Clinical Background and Genetics
- The Steroid Resistant Nephrotic Syndrome (SRNS) gene panel has been designed for the analysis of genes associated with SRNS and related renal conditions including Alport syndrome.
- SRNS is defined as:
  1. Presence of nephrotic syndrome (Serum albumin < 25g/l and urine albumin > 4 mg/m2/h or urine albumin/creatinine ratio >100 mg/mmol), that is either:
  2. Persistent to treatment with steroids, or
  3. Present in the first 3 months of life, or
  4. Focal segmental glomerulosclerosis (FSGS) on biopsy.

Service offered
- 69 genes are targeted using a custom designed SureSelect Target Enrichment System kit and sequenced using a MiSeq (Illumina) analyser. Analysis is performed using an open source in-house pipeline (alignment: BWA; modification variant and calling: GATK; variant annotation: Annovar).

Referrals
- Referrals are accepted nationally

Target reporting Time
Diagnostic screen of 69 genes: 84 days (12 weeks)
Clinically urgent samples: 6 weeks indicative RT.
Please indicate urgent samples

Known Familial Variant:
- 42 days (6 weeks) or 14 days (2 weeks)
- urgent cases (Sanger sequencing or MLPA)

Please contact the laboratory for up to date prices

Clinical Advice
- If clinical discussion is required contact:
  Prof. Moin A Saleem FRCP, PhD, Professor of Paediatric Renal Medicine, University of Bristol. Email: m.saleem@bristol.ac.uk

References
# Steroid Resistant Nephrotic Syndrome (SRNS), Alport syndrome and rare inherited renal disease NGS Panel Proforma

<table>
<thead>
<tr>
<th>Patient Name:</th>
<th>Consultant Name:</th>
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<tbody>
<tr>
<td>DOB:</td>
<td>Sex: M/F</td>
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<td>NHS Number:</td>
<td>Hospital Number:</td>
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<td>Date requested:</td>
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**Indications for testing:** nephrotic syndrome (Serum albumin < 25g/l and urine albumin > 4 mg/m2/h or urine albumin/creatinine ratio >100 mg/mmol).

- Is this patient resistant to treatment with steroids? **Yes/No**
- Presentation: Congenital/Infantile/Childhood/Juvenile/Adult
- Histology/biopsy: Membranous/MPGN/TBMN/FSGS/DMS/other/not done
- Age of onset?
- Ethnic background, if known?**
  - *This aids in interpretation of rare genetic variants.*
- Is there a family history of nephrotic syndrome?
  - If yes, please provide more details in the box below.
- Is there a family history of consanguinity?
- Initial response to steroids? **Yes/No/Partial**
- Response to second line immunosuppression? **Yes/No/Partial**
- Extra-renal features? If Yes please circle below
  - Ocular abnormalities
  - Deafness
  - Nail Patella Syndrome
  - Alport Syndrome
  - Pierson Syndrome
  - Metabolic disease
  - Epilepsy/tremor/ataxia
  - Abnormal genitalia/wilms tumour/gonadoblastoma
  - Psychomotor delay/mental retardation
  - Haematological abnormality
  - Other-please detail **Yes/No**

Please list any other relevant clinical and histological features in this patient and relevant family history:

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**It has been assumed that, in submitting a sample and request for testing, that the referring clinician has counselled the patient appropriately that:**

- Multiple genes will be targeted and analysed in the proband
- The test may or may not find the cause of the condition
- That any genetic changes detected will fall into one of the following categories:
  1. Known genetic variants compatible with the patient’s phenotype
  2. Novel genetic variants, which may be clinically relevant but which may require further investigation including family studies
  3. Novel genetic variants that may be related to the phenotype but which we are unable to interpret the clinical significance of at present.

Signed: ____________________________________________________________
Print: _____________________________________________________________

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Information document No. 75, Version 7
Active date of this version 23/07/2019
Approved by: Maggie Williams