



# PodoNet Registry

## Case Report Form – Basic Data

### Basic Patient Characteristics

Subject identifier  (AbCd)  
 Study ID  (ABC001)  
 Date of consent    (dd/mm/yyyy)  
 Date of birth   (mm/yyyy)

Sex  male  
 female

Parents Country of origin father   
 Ethnicity (father)  
 Caucasian  Asian  
 Hispanic  Indian  
 Subs. African/African American  Native American  
 Country of origin mother   
 Ethnicity (mother)  
 Caucasian  Asian  
 Hispanic  Indian  
 Subs. African/African American  Native American

Consanguinity  yes ⇒   
 No ⇒ Distance between parents birth place  km  
 Unknown ⇒ Distance between parents birth place  km

Total number of siblings

Blood sample for central DNA storage in Heidelberg collected?  
 Yes ⇒ Label for Sample P-   
 No  
 Unknown

Blood sample for DNA analysis sent to a partner laboratory?  
 Yes ⇒  Heidelberg  
 Ankara  
 Genova  
 Bochum  
 Paris  
 Bergamo  
 No ⇒

## Initial Manifestation of Disease

Date   (mm/yyyy)

- Presentation**
- Asymptomatic, incidental finding
  - Edema
    - Mild
    - Moderate
    - Severe
  - Hypertension
  - Renal failure
    - Acute
    - Chronic
  - Other

**Anthropometric data**

Weight  kg

Height  cm

**Serum biochemistry**

Creatinine  mg/dl

Protein  g/l

Albumin  mg/dl

Cholesterol  mg/dl

IgG  g/dl

C3  g/l

### Urine analysis

#### **Proteinuria**

- 24 h proteinuria  g/m<sup>2</sup>/day
- Dipstick result  neg.  (+)  +  ++  +++

- Albumin/creatinine and protein/creatinine ratio

Protein	<input type="text"/>	g/l
Albumin	<input type="text"/>	mg/dl
Creatinine	<input type="text"/>	mg/dl
Prot/Crea Ratio	<input type="text"/>	g/g
Alb/Crea Ratio	<input type="text"/>	g/g

- Hematuria**  Yes  No

## Extrarenal Findings

- Mental retardation
- Microcephaly
- CNS anomalies
- Peripheral nervous disorder
- Blindness/ visual impairment
- Short stature
- Facial dysmorphism
- Polydactylia
- Nail patella syndrome
- Spondyloepiphyseal dysplasia (Schimke's syndrome)
- Male pseudohermaphroditism (Denys-Drash or Frasier syndrome)
- Myopathy
- Cardiomyopathy
- Cardiac structure disorder
- Urogenital abnormalities
- Diabetes mellitus
- Malignant Disorder
- Autoimmune diseases

- Hepatitis B
- Hepatitis C
- HIV
- Connatal CMV
- Other TORCH infections
- Other

## Histopathological findings

**Date of biopsy**    (dd/mm/yyyy)

**Examination**

- Light microscopy
- Immunofluorescence
- Electron microscopy

**Total no. of glomeruli**

**No. of sclerosed glomeruli**

**Diagnosis**

- Minimal change glomerulonephritis
- Mesangioproliferative glomerulonephritis
- Focal-segmental glomerulonephritis
  - Tip lesion
  - Collapsing
  - Hilar
- Global glomerulosclerosis
- Diffuse mesangial sclerosis
- Other

**Specific comments**

## Genetic Studies

**NPHS1**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**NPHS2**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**NPHS3**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**WT1**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**LAMB2**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**TRPC6**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**ACTN4**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**SMARCAL1**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**GMS1**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**CD2AP**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**COQ2**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**PDSS2**

- Mutation(s) found:
  - Homozygous
  - Heterozygous
  - Compound heterozygous
- No mutation
- Not performed
- Pending


**Other**

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## Family History – Glomerular Disease in Relatives

### Relative

- |   |   |
|---|---|
| <input type="checkbox"/> Brother              | <input type="checkbox"/> Sister                 |
| <input type="checkbox"/> Father               | <input type="checkbox"/> Mother                 |
| <input type="checkbox"/> Paternal Grandfather | <input type="checkbox"/> Paternal Grandmother   |
| <input type="checkbox"/> Maternal Grandfather | <input type="checkbox"/> Maternal Grandmother   |
| <input type="checkbox"/> Paternal Uncle       | <input type="checkbox"/> Paternal Aunt          |
| <input type="checkbox"/> Maternal Uncle       | <input type="checkbox"/> Paternal Aunt          |
| <input type="checkbox"/> Paternal male cousin | <input type="checkbox"/> Paternal female cousin |
| <input type="checkbox"/> Maternal male cousin | <input type="checkbox"/> Maternal female cousin |
| <input type="checkbox"/> Son                  | <input type="checkbox"/> Daughter               |
| <input type="checkbox"/> Grandson             | <input type="checkbox"/> Granddaughter          |
| <input type="checkbox"/> Nephew               | <input type="checkbox"/> Niece                  |
| <input type="checkbox"/> Other                |   |

### First manifestation

- Small proteinuria
- Gross proteinuria
- Nephrotic syndrome
- Chronic renal failure
- Dialysis

### Age at disease onset

years

### Histopathology

- Minimal change glomerulonephritis
- Focal segmental glomerulosclerosis
- Mesangioproliferative glomerulonephritis
- Diffuse mesangial sclerosis
- Other
- Unknown

### Genetic examination

- No
- Yes ⇒
  - No mutation found
  - Mutation(s) found ⇒
  - Result unknown

### Current status

- Small proteinuria
- Gross proteinuria
- Nephrotic syndrome
- Remission
- Chronic renal failure
- Dialysis
- Transplantation
- Deceased
- Unknown

### Blood sample for central DNA storage in Heidelberg collected?

- Yes ⇒ Label for Sample P-
- No
- Unknown

### Blood sample for DNA analysis sent to a partner laboratory?

- Yes ⇒
  - Heidelberg
  - Ankara
  - Genova
  - Bochum
  - Paris
  - Bergamo
- € No ⇒