



Name:

| date | | | | |
|---|-------------|----------|--------------|-----|
| height | cm | weight | | kg |
| blood pressure | | U-Stix: | | |
| blood produit | | Proteine | _ Leucocytes | Ery |
| laboratory values | | | | |
| reason for first visit/ medical history | / course of | disease | | |
| | | | | |
| | | | | |
| | | | | |
| | | | | |
| | | | | |
| | | | | |
| | | | | |
| | | | | |
| | | | | |
| medical treatment/drugs | | | | |
| | | | | |
| | | | | |
| | | | | |

| daily fluid intake | | | mic | turition | /day |
|------------------------------------|---|----|-----|----------|------|
| edema | 0 | No | 0 | Yes | |
| Continence of urine | 0 | No | 0 | Yes | |
| recurring urinary tract infections | 0 | No | 0 | Yes | |
| gross hematuria | 0 | No | 0 | Yes | |



Medical history

Perinatal history

| gestation week at birth | birth length |
|------------------------------|-----------------------------|
| birth weight | head circumference at birth |
| particularities at birth | |
| peri/postnatal abnormalities | |

Prenatal history

| oligohydramnios | N | ? | Υ | first occured in week of gestation; intervention: | | | | | |
|--------------------------|-------|--------|-------|--|--|--|--|--|--|
| gestational diabetes | Ν | ? | Υ | first occured in week of gestation; treatment: | | | | | |
| drug therapy during | N | ? | Υ | what drugs? | | | | | |
| pregnancy | | | • | | | | | | |
| other abnormalities duri | ng pi | regna | ancy | | | | | | |
| | | | | | | | | | |
| prenatal sonographic ab | norn | naliti | es (p | lease enter weeks of gestation at onset of symptoms) | | | | | |
| | | | | | | | | | |

Family history

| father, year of birth: | | | disease: | | | | | |
|------------------------|---|-----------------------|-------------------------|-----|-------|--------------------|------|----------------|
| height | | | weight | | | ethnicity | | |
| renal disease | 0 | renal cysts | o chron. kidney disease | 0 | dia | alysis | 0 | ADPKD |
| o NONE | 0 | ARPKD | o renal transplantation | 0 | Ne | phronophthisis | 0 | HNF1ß |
| other renal disease: | | | | par | rtici | pating in any re | egis | ter study? |
| | | | | | | | | |
| mother, geb.: | | | disease: | | | | | |
| mother, geb.: | | | disease: weight | | | ethnicity | | |
| | 0 | Nierenzysten | | 0 | Dia | ethnicity alyse | 0 | ADPKD |
| height | 0 | Nierenzysten ARPKD | weight | 0 | | | 0 | ADPKD HNF1ß |

consanguinity O No O Unclear O Yes degree of relationship?

Siblings

| | date of birth | gender | relevant disease |
|---|---------------|--------|------------------|
| 1 | | | |
| 2 | | | |
| 3 | | | |
| 4 | | | |



Extended family history

Are other members of the family affected by the patient's disease or similar symptoms? Please indicate if the person is participating in a patient register study?

| | | | | Which family members? (Nr. see below) |
|--------------------------------|------|----|-------|---------------------------------------|
| renal cysts | N | ? | Υ | |
| chronic renal failure | N | ? | Υ | |
| dialysis | N | ? | Υ | |
| hepatic cysts | N | ? | Υ | |
| chronic hepatic failure | N | ? | Υ | |
| diabetes before the age of 50 | N | ? | Υ | |
| gestational diabetes | N | ? | Υ | |
| gout's disease | N | ? | Υ | |
| ARPKD | N | ? | Υ | |
| ADPKD | N | ? | Υ | |
| BBS | N | ? | Υ | |
| nephronophthisis | N | ? | Υ | |
| HNF1ß nephropathy | N | ? | Υ | |
| other: | | | | |
| maternal grandfather | | 6) | pat | ernal grandfather 11) other: |
| 2) maternal grandmother | | 7) | pat | ernal grandmother |
| 3) maternal uncle | | 8) | pat | ernal uncle |
| 4) maternal aunt | | 9) | pat | ernal aunt |
| 5) maternal cousin | | 10 |) pat | ernal cousin |
| Space for family tree (if need | ded) | | | |
| | | | | |



Physical examination

Dermatology, skeleton and left-right axis

| postaxial hexadactyly | N | ? | Υ | brachydaktyly | N | ? | Υ | |
|-----------------------|-----|---|---|-----------------------------------|---|---|---|--|
| syndactyly | N | ? | Υ | abnormal nails on fingers or toes | N | ? | Υ | |
| thoracic hypoplasia | N | ? | Υ | short ribs | N | ? | Υ | |
| cone shaped epiphyses | N | ? | Υ | dental abnormalities | N | ? | Υ | |
| acanthosis nigricans | N | ? | Υ | striae | N | ? | Υ | |
| exanthema / rash | N | ? | Υ | situs ambigous | N | ? | Υ | |
| Situs ambiguus | N | ? | Υ | Situs inversus totalis | N | ? | Υ | |
| other: | u u | | | | | | | |

Regular orthopedic consultation \circ No \circ Yes (name of specialist):

Gastroenterology

| chronic constipation | N | 3 | Υ | Leberzysten | N | 3 | Υ | |
|---------------------------|---|---|---|------------------------------|----------|---|---|--|
| chronic diarrhea | N | ? | Υ | jaundice | N | ? | Υ | |
| palpable hepatomegaly | N | ? | Υ | ascites | N | ? | Υ | |
| palpable splenomegaly | N | ? | Υ | esophageal varices | N | ? | Υ | |
| elevated liver enzymes | N | ? | Υ | hyperphagia | N | ? | Υ | |
| cholelithiasis | N | ? | Υ | Hirschsprung disease | N | ? | Υ | |
| gastrointestinal bleeding | N | ? | Υ | endoskopy (date and result): | <u> </u> | ı | | |
| other: | | 1 | 1 | | | | | |

regular Gastroenterological - $$\circ$$ No $$\circ$$ Yes (name of specialist): consultation

Pulmology / ORL

| recurring infections of | N | 2 | Υ | anosmia | N | 2 | v | |
|--------------------------|----|---|---|--------------------|----|---|---|---|
| respiratory tract or ORL | IN | ŗ | ľ | anosinia | IN | ŗ | ľ | |
| asthma bronchiale | N | ? | Υ | paracentesis | N | ? | Υ | |
| polyposis nasi | N | ? | Υ | hearing loss | N | ? | Υ | |
| diurnal tiredness | N | ? | Υ | hearing aid device | N | ? | Υ | |
| persistent snoring | N | ? | Υ | | N | ? | Υ | |
| other: | • | | | · | • | | | 1 |

 O No

FIRST VISIT

O Yes:



Cardiology

cardiac abnormalities

| dextrocardia | O | NO | | O | Yes: | | |
|-----------------------------------|--------|------|----|---|--------|------------|---|
| 24 h RR measurerment? | 0 | No | | 0 | Yes | date | performed |
| done? | | | | | | 0 | no hypertension |
| | | | | | | 0 | hypertension |
| | | | | | | 0 | nocturnal fall of blood pressure normal |
| | | | | | | 0 | nocturnal fall of blood pressure not normal |
| | | | | | | | other |
| 5 L L L 2 | | | | | ., | 0 | results enclosed |
| Echocardiography done? | 0 | No | | 0 | Yes | | done normal |
| | | | | | | | left ventricular hypertrophy |
| | | | | | | | |
| ECG done? | 0 | No | | 0 | Yes | date | other: |
| | | | | | | 0 | normal O pathologic: |
| other: | | | | | | | |
| Central nervous system | | 1 | | 1 | T | | |
| ataxia | | N | 3 | Υ | | | |
| muscular hypotonia | | N | ? | Υ | | | |
| microcephalus | | N | ? | Υ | | | |
| mental retardation | | N | ? | Υ | | | |
| epileptic seizures | | N | ? | Υ | last e | episode _ | how often? |
| EEG performed | | N | ? | Υ | last E | EEG | O normal |
| LLO periorifica | | | | | O ab | normal: | |
| autoaggression | | N | 3 | Υ | | | |
| autism spectrum disorder | | N | ? | Υ | | | |
| disorder of impulse control | | N | ? | Υ | | | |
| learning difficulties | | N | ? | Υ | | | |
| Year of schooling and school | perfor | manc | e: | | | | |
| other | | _ | | | | | |
| regular neurological consultation | | 0 | No | | 0 Y | es (name d | of specialist):. |



Developmental milestones

| free sitting | free walking | continence of urine |
|---------------|-----------------------------|---------------------|
| free standing | please enter months of life | |

Ophthalmology

| pupils | light | light reaction | | | | | | | | | |
|--------------------|-------|----------------|-----|------------------------|---|---|----------|--|--|--|--|
| loss of vision | N | ? | Υ | iris coloboma | N | ? | Υ | | | | |
| vision aid | N | ? | Υ | optic nerve atrophy | N | ? | Υ | | | | |
| strabism | N | ? | Υ | achromatopsia | N | ? | Υ | | | | |
| astigmatism | N | ? | Υ | visual field narrowing | N | ? | Υ | | | | |
| night blindness | N | ? | Υ | nystagm | N | ? | Υ | | | | |
| oculomotor apraxia | N | ? | Υ | cataract | N | ? | Υ | | | | |
| other: | I | | 1 1 | | ı | ı | <u> </u> | | | | |

regular ophthalmologic consultation

No No

O Yes (Name of specialist):.

Sexual development

| o female | menarche | PHB | | | | | | |
|----------|---------------------|--------------------------|---|---|-----------------|---|---|---|
| | PHG | testicular volume (l/r)/ | | | | | | |
| o male | maldeszensus testis | N | ? | Υ | hypospadia | N | ? | Υ |
| | mikropenis | N | ? | Υ | scrotum bifidum | N | ? | Υ |
| other: | | | | | | | | |



Renal ultrasound

| Name: _ | | date | examiner |
|---------|--|------|----------|
|---------|--|------|----------|

renal malformation or dystopy:

| renal agenesis | right / left / bilateral | N | ? | Υ | fused kidney | N | ? | Υ |
|----------------|--------------------------|---|---|---|---|---|---|---|
| megaureter | right / left / bilateral | N | ? | Υ | duplex kidneys right / left / bilateral | N | ? | Υ |
| hydronephrosis | right / left / bilateral | N | ? | Υ | crossed dystopia | N | ? | Υ |
| dysplasia | right / left / bilateral | N | ? | Υ | horseshoe kidney | N | ? | Υ |
| other: | | • | • | • | | • | • | |

RIGHT KIDNEY

LEFT KIDNEY

| localization | o orthotopic | o orthotopic |
|---|----------------------------|-------------------------------------|
| | o dystopic: | o dystopic: |
| outer shape | o smooth | o smooth |
| outer snape | o (fetal) lobulation | o (fetal) lobulation |
| renal length | cm | cm |
| renal width 1 | cm | cm |
| renal width 2 | cm | cm |
| volume | ml | ml |
| renal pelvis | mm | mm |
| ronal paranchyma | o normal | o normal |
| renal parenchyma | o high echogenity | high echogenity |
| corticomedullary | o normal | o normal |
| differentiation | o reduced | o reduced |
| | o abolished | abolished |
| | o Yes | o Yes |
| renal cysts | o No | o No |
| | o Unclear | o Unclear |
| - localization of custo | o cortical | o cortical |
| localization of cysts | o medullar | o medullar |
| | o < 5 cysts | o < 5 cysts |
| number of cysts | ○ > 5 cysts | o > 5 cysts |
| | o size < 10 mm (all cysts) | o size < 10 mm (all cysts) |
| size of cysts | o size > 10 mm (min 1cyst) | o size > 10 mm (min 1cyst) |
| lower urinary tract | | |
| | o Yes | o Yes |
| urinary tract obstruction | o No | o No |
| | o Unclear | o Unclear |
| other: | | |



Abdominal ultrasound

| Name: | | | date | e | | exam | niner _ | | | |
|------------------------------------|-------------|---------------|--------|----------|---------|-------|-------------|-------|--|--|
| | | | | | | | | | | |
| inhomogeneous hepatic parenchyma | 0 | No | 0 | unclear | 0 | Yes | | | | |
| hepatic steatosis | 0 | No | 0 | unclear | 0 | Yes | | | | |
| periportal fibrosis | 0 | No | 0 | unclear | 0 | Yes | | | | |
| ascites | 0 | No | 0 | unclear | 0 | Yes | | | | |
| biliary duct dilatation | 0 | No | 0 | unclear | 0 | Yes | 0 | | trahepatic trahepatic | |
| biliary duct cysts | 0 | No | 0 | unclear | 0 | Yes | | | | |
| collateral circulation | 0 | No | 0 | unclear | 0 | Yes | | 0 0 0 | umbilical omentum minus lower liver border spleen | |
| portal flow < 16 cm/sec | 0 | No | 0 | unclear | 0 | Yes | | | | |
| hepatic cysts | 0 | No | 0 | unclear | 0 | Yes | | | | |
| liver size in medioclavicular line | | | | cm | | | | | | |
| pancreatic cysts | 0 | No | 0 | unclear | 0 | Yes | | _ | | |
| spleen size | | | | cm | | | | | | |
| other: | | | | | | | | | | |
| | | | | | | | | | | |
| Any abnormalities of female | <u>e ge</u> | <u>nitals</u> | in abd | ominal u | ıltrası | ound: | | | | |
| | | | | | | | | | | |
| | | | | | | | | | | |
| | | | | | | | | | | |
| | | | | | | | | | | |