

Renal clinical phenotype at first MDT (Fill in to best of knowledge at time of first MDT)			
a. Renal impairment: Fill in values even if normal			
☐ Yes ☐ No ☐ Not Assessed		Date of last test:	
i. Urea: (mmol/L)		ii. Creatinine: (umol/L)	
iii. eGFR: (mL/min/1.73m ²)		iv. CKD stage:	
v. current RRT: None ☐ Haemodialysis ☐ Yes ☐ No Peritoneal dialysis ☐ Yes ☐ No		vi. transplant: ☐ Yes ☐ No ☐ donor brain death ☐ donor cardiac death ☐ living related donor ☐ living non-related donor	
b. Hypertension: Fill in values if available			
☐ Yes ☐ No ☐ Not Assessed			
i. Date of last measurement:		ii. Last measurement:	
iii. Medical management: ☐ Yes ☐ No ☐ Not Applicable (Specify) _____ _____			
c. Blood biochemical abnormalities: Fill in values only if abnormal			
☐ Yes ☐ No ☐ Not Assessed			
i. Sodium	☐ Yes ☐ No	Values: (mmol/L)	Date:
ii. Potassium	☐ Yes ☐ No	Values: (mmol/L)	Date:
iii. Chloride	☐ Yes ☐ No	Values: (mmol/L)	Date:
iv. Bicarbonate	☐ Yes ☐ No	Values: (mmol/L)	Date:
v. Calcium	☐ Yes ☐ No	Values: (mmol/L)	Date:
vi. Magnesium	☐ Yes ☐ No	Values: (mmol/L)	Date:
vii. Phosphate	☐ Yes ☐ No	Values: (mmol/L)	Date:
viii. Bilirubin	☐ Yes ☐ No	Values: (umol/L)	Date:



ix. Liver ALP	☐ Yes ☐ No	Values: (U/L)	Date:
x. Liver GGT	☐ Yes ☐ No	Values: (U/L)	Date:
xi. Liver ALT	☐ Yes ☐ No	Values: (U/L)	Date:
xii. Liver AST	☐ Yes ☐ No	Values: (U/L)	Date:
xiii. Albumin	☐ Yes ☐ No	Values: (g/L)	Date:
xiv. LDH	☐ Yes ☐ No	Values: (U/L)	Date:
xv. Urate	☐ Yes ☐ No	Values: (mmol/L)	Date:
xvi. PTH	☐ Yes ☐ No	Values: (pmol/L)	Date:
xvii. Vitamin D	☐ Yes ☐ No	Values: (nmol/L)	Date:
d. Blood haematological abnormalities: Fill in values only if abnormal			
☐ Yes ☐ No ☐ Not Assessed			
i. Hb	☐ Yes ☐ No	Values: (g/L)	Date:
ii. Platelets	☐ Yes ☐ No	Values: (x10 ⁹ /L)	Date:
iii. MCV	☐ Yes ☐ No	Values: (fL)	Date:
iv. MCH	☐ Yes ☐ No	Values: (pg)	Date:
v. Ferritin	☐ Yes ☐ No	Values: (ug/L)	Date:
vi. Iron	☐ Yes ☐ No	Values: (umol/L)	Date:
vii. Haptoglobin	☐ Yes ☐ No	Values: (g/L)	Date:
e. Urine abnormalities: Fill in values only if abnormal			
i. Haematuria	☐ Yes ☐ No ☐ Not Assessed		
Microscopic	☐ Yes ☐ No	Date:	
Macroscopic	☐ Yes ☐ No	Date:	
ii. Proteinuria	☐ Yes ☐ No ☐ Not Assessed		



24hr protein concentration	☐ Yes ☐ No	Values: (mg/24h)	Date:
Protein:creatinine ratio	☐ Yes ☐ No	Values: (g/mol creat)	Date:
Albumin:creatinine ratio	☐ Yes ☐ No	Values: (g/mol creat)	Date:
B2 microglobulin	☐ Yes ☐ No	Values: (ug/L)	Date:
iii. Calcium (24hr or Calcium:Creatinine ratio)	☐ Yes ☐ No ☐ Not Assessed		
	Values: (g/L)	Date:	
iv. Phosphate (24hr or Phosphate:Creatinine ratio)	☐ Yes ☐ No ☐ Not Assessed		
	Values: (mmol/24h or mmol/mol creat)	Date:	
v. Urate (24hr or Urate:Creatinine ratio)	☐ Yes ☐ No ☐ Not Assessed		
	Values: (mmol/24h or mmol/mol creat)	Date:	
vi. Oxalate (24hr or Oxalate:Creatinine ratio)	☐ Yes ☐ No ☐ Not Assessed		
	Values: (mmol/24h or mmol/mol creat)	Date:	
vii. Cysteine (24hr or Cysteine:Creatinine ratio)	☐ Yes ☐ No ☐ Not Assessed		
	Values: (mmol/24h or mmol/mol creat)	Date:	
viii. Glycosuria			
☐ Yes ☐ No ☐ Not Assessed		Date:	
ix. Amino aciduria			
☐ Yes ☐ No ☐ Not Assessed		Date:	
f. Renal biopsy			
☐ Yes ☐ No ☐ Unknown		Date of biopsy:	



i. Light microscopy: _____

ii. Electron microscopy: _____

iii. Immunohistochemistry: _____

g. Abdominal ultrasound

☐ Yes ☐ No ☐ Unknown

Date of last scan: _____

i. Kidney lengths (centiles): _____

ii. Kidney cysts (number, maximum size, and location): _____

iii. Structural anomalies of kidney and urinary tract: _____

iv. Other anomalies (e.g. liver, pancreas): _____

h. CT/MRI

☐ Yes ☐ No ☐ Unknown			Date of last scan:
i. Anomalies: _____ _____ _____ _____			
Non-renal clinical phenotype			
a. Audiology		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
b. Ophthalmology		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
c. Endocrinology		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
d. Skeletal		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
e. Cardiology		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
f. Respiratory		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
g. Neurology		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
h. Dermatology		☐ Yes ☐ No ☐ Not Assessed	



Features: _____ _____			
i. Haematology		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
j. Immunology		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
k. Oncology		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
l. Reproductive		☐ Yes ☐ No ☐ Not Assessed	
Features: _____ _____			
m. Developmental/Learning/Behavioural issues		☐ Yes ☐ No ☐ Not Assessed	
Features: : _____ _____			
n. Dysmorphology		☐ Yes ☐ No ☐ Not Assessed	
Features: : _____ _____ _____			
o. Growth parameters		☐ Yes ☐ No ☐ Not Assessed	
Height:	Values: (cm)	Centile:	Age:
Weight:	Values: (kg)	Centile:	Age:

Head circumference:	Values: (cm)	Centile:	Age:
Growth velocity:		Value:	Age:
Body mass index:		Age assessed:	
Genetic investigations			
a. Prior to Renal genetics MDT clinic: attach reports			
None	☐ Yes ☐ No		
i. Chromosomes (karyotype/microarray)	☐ Yes ☐ No ☐ Unknown/missing	Result:	Date:
ii. Single gene(s)	☐ Yes ☐ No ☐ Unknown/missing	Result: +ve / -ve / inconclusive Gene(s): Nucleotide: Protein: Zygosity Heterozygous/Homozygous/Hemizygous s	Date: Variant class: 1, 2, 3A, 3B, 3C 4, 5
Variant type Missense/Nonsense (prem stop)/Frameshift/splicing/Synonymous/Exon deletion/Exon duplication/Whole gene deletion/whole gene duplication/In frame Deletion			
iii. Gene panel	☐ Yes ☐ No ☐ Unknown/missing	Result: +ve / -ve / inconclusive Gene/s: Nucleotide: Protein: Zygosity Heterozygous/Homozygous/Hemizygous	Date: Variant class 1, 2, 3A, 3B, 3C 4, 5
Gene panel name:			
Variant type Missense/Nonsense (prem stop)/Frameshift/splicing/Synonymous/Exon deletion/Exon duplication/Whole gene deletion/whole gene duplication/In frame Deletion			
Additional Variant information:			
iv. Whole Exome	☐ Yes ☐ No ☐ Unknown/missing	Result: +ve / -ve / inconclusive Gene/s: Nucleotide: Protein: Zygosity Heterozygous/Homozygous/Hemizygous	Date: Variant class 1, 2, 3A, 3B, 3C 4, 5
Gene panel name:			

Variant type: Missense/Nonsense (prem stop)/Frameshift/splicing/Synonymous/Exon deletion/Exon duplication/Whole gene deletion/whole gene duplication/In frame Deletion			
Additional Variant information:			
v. Whole Genome ☐ Yes ☐ No ☐ Unknown/missing Gene panel name:		Result: +ve / -ve / inconclusive Gene/s: Nucleotide: Protein: Zygosity Heterozygous/Homozygous/He mizygous	Date: Variant class 1, 2, 3A, 3B, 3C 4, 5
Variant type: Missense/Nonsense (prem stop)/Frameshift/splicing/Synonymous/Exon deletion/Exon duplication/Whole gene deletion/whole gene duplication/In frame Deletion			
Additional Variant information:			
vi. Unknown ☐ Yes ☐ No		Information	
Additional information on unknown genetic tests			