		Variable Units	R76W			Other *HNF4A*			P value
			n	Median	IQR	n	Median	IQR	
Features of a renal Fanconi syndrome	Low molecular weight proteinuria	Urinary α-1 microglobulin mg/L	6	205	83.5-287.8	20	6.0	6.0-6.8	0.00005
		Urinary β-2 microglobulin mg/L	6	62.0	36.0-87.4	20	0.3	0.1-0.3	0.0001
		Urinary retinol binding protein mg/L	6	100.5	56.3-150.5	20	2.0	2.0-2.0	0.000003
	Hypouricaemia	Serum urate μmol/L	4	96.0	84.8-108.0	16	268.0	233.3-321.3	0.002
	Glycosuria	Serum glucose mmol/L	6	4.8	3.4-11.2	17	7.2	5.3-9.8	0.2
		Urine glucose mmol/L	6	55.7	37.6-119.6	20	0.3	0.3-1.5	0.001
	Phosphaturia	Serum phosphate mmol/L	6	1.09	0.73-1.22	18	1.15	1.03-1.27	0.4
		Urinary phosphate:creatinine mmol/mol	6	4.22	2.98-6.07	20	2.1	1.5-3.0	0.009
	Renal tubular acidosis	Serum bicarbonate mmol/L	6	22.0	20.3-26.3	18	23.4	22.2-24.5	0.5
Additional/atypical features	Renal function	Serum creatinine μmol/L	6	114.0	82.5-146.0	20	84.0	67.8-102.8	0.03
	Other biochemistry	Serum magnesium mmol/L	5	1.10	0.91-1.16	18	0.82	0.77-0.88	0.01
		Urinary magnesium:creatinine mmol/mol	6	0.69	0.49-1.04	20	0.4	0.34-0.5	0.02
		Urinary oxalate:creatinine mmol/mol	6	0.050	0.040-0.125	20	0.034	0.028-0.046	0.03
		Serum calcium mmol/L	6	2.14	2.01-2.22	18	2.39	2.31-2.47	0.001
		Urinary calcium:creatinine mmol/mol	6	0.97	0.47-1.15	20	0.5	0.38-0.65	0.05
		Serum PTH pmol/L	5	4.4	4.3-4.8	17	3.0	2.1-4.0	0.002
			R76W			Other *HNF4A*			P value
	Imaging	Nephrocalcinosis on renal ultrasound	5	5/5		10	0/10		0.0003

Supplementary Table 1. Comparison of biochemical and radiological features between patients with the R76W mutation and patients with other HNF4A mutations

Urinary amino acid umol/mmol Cr	R76W (n=6)		Other HNF4A (n=15)		p value
	Median	IQR	Median	IQR	
Hydroxyproline	79.5	54.0-107.8	6.0	3.0-9.0	0.001*
Proline	69.0	47.5-143.3	1.0	1.0-3.0	0.0004*
Phosphoethanolamine	2.5	0.75-5.75	2.0	1.0-5.0	0.9
Threonine	138.0	69.8-265.5	18.0	11.0-30.0	0.002*
Serine	149.5	90.0-221.8	34.0	30.0-56.0	0.02
Aspargine	48.5	26.0-140.0	16.0	8.0-30.0	0.06
Glutamic acid	195.5	86.5-220.3	3.0	2.0-5.0	0.0004*
Glutamine	137.0	80.3-233.3	37.0	16.0-61.0	0.002*
Glycine	1101.5	957.5-1428.5	197.0	105.0-310.0	0.0005*
Alanine	231.0	162.5-349.0	50.0	26.0-80.0	0.001*
Citrulline	61.5	38.0-144.0	1.0	1.0-2.0	0.0002*
Valine	17.5	3.8-33.8	3.0	2.0-5.0	0.01
Cystine	97.5	75.0-137.0	5.0	4.0-10.0	0.0004*
Methionine	5.5	4.0-7.8	5.0	4.0-7.0	0.5
Isoleucine	3.0	0.8-6.5	3.0	2.0-5.0	1.0
Leucine	15.0	9.3-45.0	7.0	5.0-8.0	0.005
Tyrosine	23.0	13.3-43.8	10.0	8.0-19.0	0.02
Phenylalanine	17.0	8.0-31.3	7.0	4.0-11.0	0.03
Ornithine	93.5	51.3-184.8	2.0	1.0-3.0	0.0004*
Lysine	400.5	329.3-780.3	14.0	11.0-30.0	0.0004*
Histidine	243.0	137.3-282.3	73.0	31.0-96.0	0.001*
Tryptophan	6.5	3.3-15.3	4.0	2.0-10.0	0.5
Arginine	64.5	23.8-92.3	2.0	1.0-3.0	0.0004*
Mean Z score	22.9		0.2		

Supplementary Table 2. Comparison of urinary amino acids between patients with R76W mutation and patients with other HNF4A mutations

Data are median (umol/mmol creatinine).
P-value calculated using Mann Whitney U test.
*Significance after a Bonferroni correction for analysing 23 amino acids, $P < 0.002$.