

Additional file 1. Clinical characteristics of the 22 previous cases and present case with *TRIM8* variant

No.	Sex	Epilepsy	Age 1 ^a	DD ^b	Facial features	Brain magnetic resonance imaging	Others	Renal disease	Age 2 ^c	Renal biopsy	Renal outcome	<i>TRIM8</i> variants	Ref
1	M ^d	Yes	0.4	Yes	Mild facial features, long philtrum	Widespread moderate cortical atrophy, scattered T2 signal increase in the white matter	Hypospadias	None	NA	NA ^e	NA	p.Ala374Argfs*16	(10)
2	M	Yes	1.8	Yes	synophridia, sunken eyes, long philtrum	NM ^f	None	None	NA	NA	NA	p.Gln423*	(9)
3	M	Yes	0.2	Yes	Microcephaly, upper-slanted palpebral fissures, long-arched eyebrows, thin lips, large ear lobes	Normal	Hip dislocation, enuresis, constipation	None	NA	NA	NA	p.Cys367Trpfs*43	(8)
4	F ^g	Yes	3.4	Yes	None	Patchy confluent regions of T2 hyperintensity with small cysts in the frontal and peritrigonal white matter on both sides	Cheerful disposition	Nephrotic- range proteinuria	NM	None	NM	p.Ser444*	(9)

5	F	Yes	1.8	Yes	Microcephalic, left simian crease, fine hair	Normal brain structures with hypomyelination at the anterior limb of the bilateral internal capsule and bifrontal lobes of the white matter	None	Proteinuria	0.7	None	Spontaneously resolved	p.Gln459*	(9)
6	F	Yes	2	Yes	Gingival overgrowth, synophrys, long philtrum	Delayed myelination	Long extremities, joint hyperlaxity	Nephrotic-range proteinuria	4	Mesangial glomerulonephritis with nonspecific IgM deposits	NM	p.Tyr446*	(9)
7	M	Yes	2.4	Yes	NM	Normal	None	Nephrotic-range proteinuria	2.4	FSGS	ESRD	p.Tyr460*	(7)
8	M	Yes	5	Yes	NM	Identified an old infarct, other brain structures appeared normal	Right-side squint, chronic constipation	Nephrotic-range proteinuria	2.2	FSGS	ESRD	p.Tyr400ArgfsTer2	(6)
9	F	No	NA	No	None	Normal	None	nephrotic-range proteinuria	3	FSGS; cystic dilatation of distal tubules	ESRD	p.Tyr487*	(5)

10-19	4M 6F	Yes	1.5 to 4.5	Yes	Broad nasal bridge, broad forehead, plagiocephaly, low-set ears, high arched palate, micrognathia	Except for 1 normal case, the others showed different degrees of abnormality	Increased bladder pressure, hyperopia, astigmatism, amblyopia, sensorineural hearing loss, scoliosis	8 cases of nephrotic syndrome, 2 cases of nephrotic-r ange proteinuria	Birth to 13.7	Two cases of diffuse mesangial sclerosis, the others are FSGS	ESRD	p.Phe388Serfs*2, p.Gly401*, p.Gln411*, p.Gln414*, p.Gln423*, p.Gln445*, p.Tyr460*, Three cases are p.Gln459*	(4)
20 (twins)	M	No	NA	Yes (mild)	None	NM	Both hypotonia, Tourette's syndrome, preserved verbal intelligence quotient, difficulties with fine movements.	Both had nephrotic-r ange proteinuria	6	FSGS	ESRD	p.Tyr487*	(4)
21	F	No	NA	Yes (mild)	None	Normal	2 convulsions associated with hypertension (electroencephalogr -am results were normal)	Nephrotic- range proteinuria	3.1	FSGS	ESRD	p.Tyr487	(11)
22	F	No	NA	No	None	NM	None	Nephrotic- range proteinuria	3.3	Glomerular- segmental mesangial	Proteinuria decreased after	p.Gln485	(11)

										proliferative lesions	treatment		
23	M	No	NA	No	Low-set ears, micrognathia, thin lips, wide eye distance, stubby neck, large ear lobes	Slightly wider bilateral frontal extracerebral space, the left lateral ventricle is slightly fuller than the right lateral ventricle, and presence of an arachnoid cyst or cisterna magna	Reduced kidney size, hyperlipidemia, neurogenic bladder, astigmatism, myopia	Nephrotic-range proteinuria	6	FSGS; cystic dilatation of renal tubules	Stage 3 chronic kidney disease	p.Trp495*	Present case

^aAge 1: age at the time of onset of epilepsy (year)

^bDD: developmental delay

^cAge 2: age at the time of onset of renal disease (year)

^dM: male

^eNA: not applicable

^fNM: not mentioned

^gF: female