S1 Table. Clinical characteristics of the 34 patients with bi-allelic mutations in the *SMARCAL1* gene.

Family ID (reference if already published)	Mutation(s)	Diagnosis	Gender	Country of origin	Age at 1st manifestation (years)	Initial proteinuria	Initial GFR ml/min/1.73m^2	Histopathology	Age at ESRD (years)	Age at last observation (years)	Response to immunosuppresive treatment	Neurological signs and symptoms	Infections, immunodeficiency, autoimmune diseases ,lympho-proliferative disorders	Other extra-renal signs	Thyroid function	Height at 1st manifestation (SD)	Height at last observation (SD)
1	c.49C>T (p.Arg17*) hom.	phenotype	F	Germany	n/a	0.3g/dl	113	n/a	—	7.45 (death)	n/a	TIAs, migraine, multiple ischemic strokes, secondary focal seizures	—	developmental and speech delay, multiple pigmented nevi, malposition of teeth, IUGR	normal	n/a	-9.7
2	c.49C>T (p.Arg17*) c.836T>C (p.Phe279Ser)	SRNS-gene panel	F	Germany	9.5	14.1 g/g uPCr	93	collapsing FSGS	11.7	15.9	no response to steroids, MMF, *Rituximab*	isolated high-frequency hearing loss	low CD4-helper	neonatal asphyxia resulting in developmental and speech delay, mild/moderate ID, IUGR, preterm delivery	hypo-thyroidism	-3.4	-5.0
3	c.298A>T (p.Lys100*) c.2114C>T (p.Thr705Ile)	phenotype	F	Germany	7.2	4.9g/m^2/day	189	FSGS	11.8	16.0	no response to steroids, CsA, CP,MMF, *Tacrolimus, Rituximab*	—	T-cell deficiency	restrictive lung disease, multiple pigmented nevi, IUGR, preterm delivery	normal	-2.9	-3.9
4-I	c.836T>C (p.Phe279Ser) c.2542G>T (p.Glu848*)	phenotype	M	Germany	13.0	n/a	12	n/a	15.3	34.1	not treated with steroids/CsA/other	—	n/a	—	normal	n/a	-1.8
4-II	c.836T>C (p.Phe279Ser) c.2542G>T (p.Glu848*)	phenotype	M	Germany	12.4	2 g/day	75	FSGS	18.5	31.2	not treated with steroids/CsA/other	TIAs, multiple ischemic strokes, secondary seizures	lymphopenia, low CD4, CD8	restrictive lung disease, IUGR, preterm delivery factor V (Leiden) mutation	hypo-thyroidism	-2.7	-5.8
5-I	c.836T>C (p.Phe279Ser) c.2542G>T (p.Glu848*)	SRNS-gene panel	F	Poland	4.8	3.2 g/day	119	minimal change disease	8.7	9.1	no response to steroids transient response to CsA	TIAs, migraine	frequent respiratory infections; low CD4-helper	Painful muscle contractions and creatine kinase serum levels >2,000 IU/l during infection; IUGR, preterm delivery	latent hypo-thyroidism	-2.6	-4.5
6 (ref.5;13)	c.1026C>A (p.Tyr342*) c.2264T>G (p.Ile755Ser)	phenotype	M	Italy	3.6	n/a	47	collapsing FSGS	3.8	4.6 (death)	no response to steroids/CsA	TIAs, secondary seizures, lower extremity hypotonia	sepsis, persistent diarrhea, lymphopenia, low CD4, CD8	moderate ID, microdontia, agenesia of the 5th tooth (lower arch), solitary kidney, multiple pigmented nevi, IUGR, preterm delivery	n/a	-2.3	n/a

7	c.1382G>A (p.Gly461Asp) hom.	phenotype	M	Czech	4.1	16 g/day	153	FSGS/GGS	5.6	11.5	not treated with steroids/CsA/other	neonatal hypotonia	frequent respiratory infections low CD4, CD8	mild developmental delay, malformed deciduous molars, multiple pigmented nevi, IUGR	normal	-1.9	n/a
8 (ref.9)	c.1439C>T (p.Pro480Leu) c.2264T>G (p.Ile755Ser)	phenotype	F	Italy	4.7	n/a	107	n/a	6.3	9.0 (death)	no response to steroids, CsA	—	catheter-related sepsis, pulmonary Aspergillosis ITP lymphopenia, low IgG	restrictive lung disease, developmental delay,mild ID, microdontia, IUGR, preterm delivery	hypo-thyroidism	-3.6	-5.4
9 (ref.6)	c.1439C>T (p.Pro480Leu) c.2542G>T (p.Glu848*)	phenotype	F	Czech	2.2	1-2 g/m^2/d	114	FSGS/GGS	—	5.7 (death)	not treated with steroids/CsA/other	TIAs, multiple ischemic strokes, secondary seizures, triplegia with motoric aphasia	fungal sepsis self-limited neonatal thrombocytopenia; Evans syndrome (age 5yrs) low CD4, CD8	mild developmental delay, multiple pigmented nevi, IUGR	normal	-4.2	-7.4
10	c.1681C>T (p.Arg561Cys) hom.	SRNS-gene panel	F	Turkey	17.2	2.1 g/day	81	FSGS	—	18.6	no response to steroids, Rituximab partial response to CsA	—	—	—	normal	-4.2	-4.1
11	c.1736C>A (p.Ser579*) hom.	phenotype	M	Lebanon	4.1	28g/g uPCr	59	FSGS	—	6.6 (death)	no response to steroids partial response to CsA	TIAs, multiple ischemic strokes, secondary seizures, hemiparesis	frequent respiratory infections PTLD of the colon (EBV assoc.) non Hodgkin B-cell lymphoma low CD4-helper	mild developmental delay, moderate ID; ASD-II; PFO, dystopia of the right kidney, bilateral hernia inguinalis, IUGR, preterm delivery t-PA deficiency and elevated PAI-I (hom.)	hypo-thyroidism	-4.5	-6.3
12-I	c.1736C>A (p.Ser579*) hom.	phenotype	F	Lebanon	4.0	18.3g/g uPCr	145	FSGS	5.7	6.8 (death)	no response to steroids partial response to CsA, *Tacrolimus*	—	PTLD of the CNS and lung (EBV assoc.) non Hodgkin B-cell lymphoma T-cell deficiency	restrictive lung disease requiring O_2 therapy; ASD, PFO, IUGR, preterm delivery	hypo-thyroidism	-4.8	-6.0
12-II	c.1736C>A (p.Ser579*) hom.	phenotype	M	Lebanon	3.0	3.4g/g uPCr	107	not performed	—	6.3 (death)	no response to steroids partial response to *Tacrolimus*	—	frequent respiratory infections low CD4, CD8	restrictive lung disease requiring O_2 therapy; multiple pigmented nevi, IUGR	hypo-thyroidism	-5.5	-7.4
13	c.1739G>T (p.Gly580Val) c.2423C>G (p.Pro808Arg)	phenotype	M	Macedonia	2.8	+++ (dipstick)	68	not performed	4.0	7.3 (death)	not treated with steroids/CsA/other	periventricular white matter defects, secondary seizures,	frequent respiratory infections mild leukopenia	microdontia, mild developmental delay, no perinatal data	latent hypo-thyroidism	-3.6	-7.3

14	c.1756C>T (p.Arg586Trp) hom.	phenotype	M	Turkey	9.6	9.6g/g uPCr	158	FSGS	10.0	18.1	no response to steroids, CsA, CP	—	meningitis, MDS susp. mild leucopenia, low CD4	post-transplant BK virus asscociated nephropathy (BKVAN); multiple pigmented nevi, IUGR	normal	-3.7	-6.9
15 (ref.8)	c1859G>A (p.Trp620*) c.2542G>T (p.Glu848*)	phenotype	M	Poland	4.4	9.1g/day	91	hilar FSGS	8.6	11.2	no response to steroids, CsA partial response to MMF	—	frequent respiratory infections post-transplant ITP mild leucopenia, low CD4, CD8, CD3, IgG	multiple pigmented nevi, IUGR, preterm delivery	normal	-2.0	-4.9
16	c.1931G>A (p.Arg644Gln) c.2542G>T (p.Glu848*)	SRNS-gene panel	M	Poland	3.2	0.16g/day	206	minimal change disease	—	8.3	no response to steroids partial response to CsA	—	mild leucopenia, low CD4	microdontia, delayed permanent teething, multiple pigmented nevi, IUGR	n/a	0.3	-1.6
17	c.1933C>G (p.Arg645Gly) c.2425G>A (p.Gly809Arg)	phenotype	M	France	10.8	n/a	<60	FSGS	12.2	13.1	no response to steroids, CsA, *Tacrolimus*	ischemic stroke, demyelinating peripheral neuropathy, epilepsy (absence)	catheter-related sepsis SCID	femoral head necrosis, multiple pigmented nevi, IUGR, preterm delivery	normal	n/a	-3.7
18	c.1940A>C (p.Lys647Thr) hom.	SRNS-gene panel	F	Morocco	5.7	6.3g/g uPCr	128	minimal change disease	—	6.7	no response to steroids, CsA MMF,	migraine-like headaches, possible episode of TIA, epilepsy (absence)	mild leucopenia, low CD4, CD8	severe photophobia, macular opacities, IUGR	normal	-2.2	-2.8
19	c.1940A>C (p.Lys647Thr) hom.	SRNS-gene panel	F	Algeria	5.3	1.4g/day	~60	collapsing FSGS	5.6	6.6	no response to steroids; no CsA/other treatment	—	mild leucopenia, lymphocyte subpopulation tests not performed	chronic vomiting and diarrhea, IUGR	normal	n/a	-4.4
20-I	c.2070+2insT c. 2462T>G (p.Ile821Ser)	phenotype	M	France	3.4	n/a	n/a	FSGS	5.7	9.8 (death)	no response to steroids partial response to CsA	TIAs, secondary seizures	frequent respiratory infections mild leucopenia, lymphopenia, low IgG	restrictive lung disease requiring O_2 therapy during infections; multiple pigmented nevi, malposition of teeth, IUGR, preterm delivery	hypo-thyroidism	-2.1	-4.7
20-II	c.2070+2insT c. 2462T>G (p.Ile821Ser)	phenotype	M	France	3.2	0.7g/l	105	minimal change disease	6.2	8.0 (death at 10 yrs)	no response to steroids partial response to CsA	TIAs, ischemic stroke, secondary seizures, hemiplegia	frequent respiratory infections SCID	hypertrophic and dilated cardiomyopathy with acute and transient heart failure (age 6yrs) multiple pigmented nevi, IUGR, preterm delivery	normal	-2.0	-5.0
21	c.2149_2150dupAT (p.Leu718Serfs*13) c.2542G>T (p.Glu848*)	phenotype	M	Germany	2.1	8.2g/g uPCr	112	minimal change disease	—	3.4	no response to steroids, CsA	—	n/a low CD4	photophobia, multiple pigmented nevi, IUGR, preterm delivery	latent hypo-thyroidism	-2.4	-4.3

22 (ref.7)	c.2207delT (p.Val736Glyfs*75) hom.	phenotype	M	Saudi Arabia (Yamani)	6.8	7.4g/l	129	FSGS	—	10.4	no response to steroids, CsA, CP	—	frequent respiratory infections	loose, fragile, and yellow-stained primary teeth, polyhydramnion, IUGR	hypo-thyroidism	-5.3	n/a
23	c.2244+5 G>A hom.	phenotype	M	United Kingdom	4.5	n/a	n/a	not performed	—	5.5	not treated with steroids/CsA/other	n/a	n/a	mild developmental delay,mild ID, no perinatal data	n/a	n/a	n/a
24-I	c.2290C>T (p.Arg764Trp) hom.	SRNS-gene panel	M	Saudi Arabia	8.4	+++ (dipstick)	28	FSGS	8.7	10.7	no response to steroids, MMF	generalized tonic-clonic seizures secondary to ESKD	lymphocyte subpopulation tests not performed	multiple pigmented nevi, IUGR	hypo-thyroidism	-4.6	n/a
24-II	c.2290C>T (p.Arg764Trp) hom.	family screening	F	Saudi Arabia	5.1	4g/l	180	FSGS	—	7.2	no response to steroids, CsA	—	lymphocyte subpopulation tests not performed	multiple pigmented nevi, IUGR	latent hypo-thyroidism	-3.6	-4.9
25	c.2308C>T (p.Gln770*) hom.	phenotype	M	Jordan	4.2	9.3g/g uPCr	252	FSGS	—	6.0 (death)	no response to steroids, MMF	ischemic stroke, migraine	Sepsis low CD4	multiple pigmented nevi, clinodactyly, syndactyly of toes, IUGR, preterm delivery	latent hypo-thyroidism	-5.5	-6.6
26	c.2542G>T (p.Glu848*) c.2459G>T (p.Arg820Leu)	SRNS-gene panel	F	Poland	2.6	3g/day	40	FSGS	3.1	10.1	no response to steroids, CsA	migraine-like headaches, focal seizures	acute disseminated encephalomyelitis (ADEM) no meningitis mild leucopenia	multiple pigmented nevi, IUGR, preterm delivery	normal	-4.4	-7.0
27-I	c.2459G>A (p.Arg820His) hom.	SRNS-gene panel	M	Turkey	8.2	'nephrotic range'	52	FSGS	9.2	9.2	no response to steroids, CsA	—	lymphocyte subpopulation tests not performed	no perinatal data	normal	-2.9	-3.3
27-II	c.2459G>A (p.Arg820His) hom.	family screening	F	Turkey	6.1	1.1g/g uPCr	200	FSGS/GGS	6.6	8.0 (death)	no response to steroids, CsA	—	frequent respiratory infections lymphocyte subpopulation tests not performed	no perinatal data	n/a	-4.6	n/a
5-II	c.2542G>T (p Glu848*) hom.	phenotype	M	Poland	1.4	++ (dipstick)	n/a	FSGS	4.6	6.1	no response to steroids, CsA	—	frequent respiratory infections low CD4	microdontia multiple pigmented nevi, oligohydramnion, IUGR, preterm delivery	hypo-thyroidism	-0.1	-4.7
28	c.2542G>T (p Glu848*) del(2)(q34-q36)	phenotype	F	Germany	2.5	1.9g/g uPCr	165	not performed	—	6.4 (death)	not treated with steroids/CsA/other	multiple ischemic strokes, secondary focal seizures,	pancytopenia in infancy T-cell (CD4, CD8) deficiency	developmental delay,moderate ID, corneal opacities, microcornea, oligohydramnion, IUGR, preterm delivery	normal	-4.5	-7.4

Legend:

n/a – not available; hom – homozygous; M – male; F – female; uPCr – urinary protein/creatinine ratio; uAlbCr – urinary albumin/creatinine ratio; FSGS – focal segmental glomerulosclerosis; GGS – global glomerulosclerosis; ESRD – end-stage renal disease; CsA – *ciclosporin A*; MMF – *Mycophenolate mofetil*; CP – *Cyclophosphamide;* TIA – transient ischemic attack; CD – a class of glycoproteins (Cluster of Differentiation); tPA – tissue plasminogen activator; PAI-I – plasminogen activator inhibitor-1 (also known as endothelial plasminogen activator inhibitor or serpin E1); PTLD – Post-transplant lymphoproliferative disorder; CNS – central nervous system; EBV – Ebstein-Barr virus; ITP – immune thrombocytopenic purpura; IgG – immunoglobulin G; MDS – myelodysplastic syndrome; SCID – severe combined immunodeficiency; IUGR – intrauterine growth retardation; ID – intellectual disability; ASD – atrial septum defect; PFO – patent foramen ovale; SD – standard deviation;