

Tubulopathy, genetic confirmed

S u b j e c t	A g e (y)	Hei ght (cm)	We igh t (kg)	BM I (kg/m ²)	Diagnosis	Mutation	Clinical presentation	Parental height (cm)		Bone age	Pub ertal stat us
								Pa ter nal	Ma ter nal		
1	7.1	112.7	17.2	13.5	Tubulair Mg reabsorptie defect	TRPM6	FTT	180	167	confirm age	A1 P1 M1
2	2.4	86.4	11.3	15.1	Gitelman syndrome	SLC12A3	FTT, salt craving, muscle cramps, decreased muscle strength	196	164	confirm age	A1 P1 M1
3	5.1	98.2	16.1	16.7	X-linked hypophosphatemic ricketts and SHOX mutation	Xp22.33p11.4 deletion (including SHOX and PHEX)	FTT, schisis, dilated left urether, patent foramen ovale, coeliac disease	171	164	delayed skeletal age 4y2m	A1 P1 M1
4	3.7	87.1	13.5	17.8	X-linked hypophosphatemic ricketts	c.1080-72C>T variant in intron 9 of PHEX gene	FTT, unstaible gait, frontal bossing, dysproportional body (shorted lower limbs)	180	164	shows metaphyseal chodrodysplasia, no skeletal age calculated	A1 P1 M1
5	5.0	102.0	14.2	13.6	Autosomal dominant Fanconi Syndrome	EHHADH gene	FTT, cholecystic lithiasis	unknow n due adoptio n		Not available	A1 P1 G1

Tubulopathy, not genetic confirmed

S u b j e c t	A g e (y)	Hei ght (c m)	We igh t (kg)	BM I (kg / m ²)	Diagnosis	Supplementation	Clinical presentation	Parental height		Bone age	Pub ertal stat us
								Pa ter nal	Ma ter nal		
1	2. 5	86, 5	10, 92	14, 6	renal tubular acidosis	Iron, vitamine D, sodiumbicarbonate	FTT, dysmorfic dental- and hair growth, development delay, periorbital oedema, polyuria, ferriprive anemia	/	/	confirm age	A1 P1 G1
2	1. 9	79	10, 335	16, 6	incomplete renal tubular acidosis	sodium bicarbonate	FTT, diarrhea, fatigue	17 2,5	16 9	delayed skeletal age, 1y3m	A1 P1 G1
3	3. 8	93	11, 4	13, 2	incomplete renal tubular acidosis	potassium citrate, sodium bicarbonate, Vitamine D	FTT	/	/	/	A1 P1 G1
4	2. 0	74, 5	7,8 5	14, 1	Fanconi syndrome secondary to mitochondrial respiratory chain defect	potasium and salt supplements, indometacine, nasogastric feeding	FTT, facial dysmorfics, motoric development delay, polyuria, recurrent urine infections by an ectopic kidney	/	/	/	A1 P1 M1

5	1 1. 6	136 ,7	31, 15	16, 5	salt losing nephropathy, hyperkalemia, hypercalciuria	Salt supplementation (sodium chloride)	FTT, polyuria	unknown due adoption		delayed skeletal age, 10y	A1 P1 G1
6	6. 1 1	108 ,5	17, 7	15	Salt- and magnesium losing nephropathy	Magnesium and sodium chloride	FTT, salt craving	17 0	15 3,8	delayed skeletal age, 5y9m	A1 P1 M1
7	9. 1	124 ,4	33, 5	22, 1	hypermagnesuria	Magnesium	FTT, born small for gestational age, urine tract infections, reduced muscle strength	16 7	15 7	delayed skeletal age, 8y10m	

