

Table 1. Patient Characteristics

	P1	P2	P3	P4	P5	P6
Origin	Turkish	United Kingdom	French	French (West Indies)	Turkish	Turkish
Current age	6 y	2 y	22 y	10.5 y	died 93 rd day	3 m
Onset age	3 weeks	1 week	1 week	4 days	1 day	5 days
Severe diarrhea	yes, with hypovolemic shock	yes	yes, intermittent until 10 y	yes	yes	yes
Duodenal villous atrophy	yes	yes	yes	yes	ND	ND
Sub apical PAS accumulation	yes	yes	yes	yes	ND	ND
Sub apical CD10 accumulation	yes	yes	ND	yes	ND	ND
Electron microscopy	Vesicles or tubulovesicular structures MVI Basolateral microvilli large lysosomes/autophagosomes Defective microvilli anchored deep in the cytoplasm thickened terminal web region	Vesicles or tubulovesicular structures Defective microvilli Basolateral microvilli large lysosomes/autophagosomes	ND	ND	ND	ND
Treatment	TPN Total enterectomy	TPN	Exclusive enteral nutrition	TPN Small bowel transplantation	TPN	TPN
Cholestasis	yes	no	yes	no	yes	yes
Bone fragility	Fracture of the left inferior part of the femur at 7 th months and at 2 y Fracture of the right wrist at 1 y	no	Fracture of the scapula at 7 y Other fracture at 10 y	no	ND	One bone fracture upon delivery
Deafness	no	no	yes	no	no	ND

Other manifestations	no	no	blind: right anophthalmia, Peters anomaly & left pyramidal cataract / severe ID & behavioral disorder	ID	ND	ND
---------------------------------	----	----	---	----	----	----

ND: not determined; ID: intellectual disability; TPN: total parenteral nutrition; MVI, microvillus inclusion.