

Table S1. Clinical features of family members with WNT10A gene mutations

Pedigree number	II-2	III-2	III-4	III-6	IV-1	IV-2	IV-3
Sex	M	F	F	M	M	F	F
Age	83	53	51	47	24	21	22
Genotype							
p.Cys107X	+/+	+/-	+/-	+/-	+/-	-/-	+/-
p.Phe228Ile	-/-	+/-	-/-	+/-	+/-	+/+	-/-
Phenotype							
Eyelid cysts	Yes	Yes*	Yes*	No	No	No	No
Palmoplantar keratoderma	Yes	Yes	Mild	Yes	No	No	No
Dystrophic fingernails	Yes	Fragility	No	Fragility	No	No	No
Dystrophic toenails	Pterygium	Poor growth					
Deciduous dentition	Yes	Yes	Mild onycholysis	Mild	No	No	No
Permanent dentition	Microdontia	Microdontia	Normal	Microdontia	Microdontia	Widely spaced	Normal
	Oligodontia	Widely spaced	Normal	Widely spaced	Oligodontia	Mild hypodontia	Normal
	Conical teeth	Retained deciduous dentition		Hypodontia	Conical teeth	Retained deciduous teeth	
	Dentures aged 14 years	No	No	Conical teeth	Dental implants		
Hypotrichosis (head)	Androgenetic alopecia present – Hamilton V1			No	No	No	No
Hypotrichosis (other)	Sparse eyelashes			Sparse eyebrows	No	No	No
Abnormalities of sweating	Hyperhidrosis	Hyperhidrosis	No	Hyperhidrosis	No	None reported, but easy overheating	No
Benign adnexal tumours	Multiple seborrhoeic keratoses	Multiple seborrhoeic keratoses		Yes	No	No	No
	Eccrine hidrocystoma	No		No	No	No	No
Malignancies	Cutaneous:			No	No	No	No
	Malignant melanoma						
	Squamous cell carcinoma						
	Basal cell carcinomas						
Photophobia	Mild	Mild	No	Mild	No	No	No
Retinal abnormalities	Retinal detachment (aged 78 years; cause unknown)	No	No	No	No	No	No
Other features	Absent dermatoglyphics	Facial erythema and easy flushing	Facial erythema and easy flushing	No	No	No	No
	Facial erythema and easy flushing		Telangiectasia				
	Smooth tongue						

*Suspected apocrine hidrocystomas but not histologically proven.