Table SI. Clinical features of TP63-related disorders

TP63	Our patient	AEC syndrome Ankyloblepha-ron-ectodermal defects-cleft lip/palate syndrome	ADULT syndrome Acro-dermo-ungual-lacrimal-tooth syndrome	EEC syndrome Ectrodactyly, ectodermal dysplasia, cleft lip/palate syndrome	Limb-mammary syndrome	SHFM4 Split-hand/foot malformation type 4	CL/P Isolated cleft lip/cleft palate
Dermal erosions (body)	X	X					
Dermal erosions (scalp)	X	X					
Ectodermal dysplasia		X	X	X		X	
Hypohidrosis		X	X		X		
Nail dysplasia	X	X	X	X	X		
Hypotrichosis		X	X	X			
Teeth abnormalities	X	X	X	X	X		
Ankyloblepharon		X					
Cleft lip/palate		X		X	X		X
Choanal atresia	X	(X)					
Split-hand/foot malformation/ syndactyly		X	X	X	X	X	
Lacrimal duct obstruction		X	X	X	X		
Hypopigmentation		X	X	X			
Hypospadias/genitourinary deformities	X	X		X			
Trismus		X					
Lentigines			X				
Hypoplastic breasts			X		X		
Hypoplastic nipples			X		X		

Clinical features of TP63-related disorders. This table shows a summary of clinical features present in our patient as well as common signs of TP63-related disorders. X indicates that a feature is present. (X) indicates that there are rare reports of this finding. This summary implies that the different entities are a range of disease severity of one single disorder. The clinical findings in our index case best match the diagnosis of AEC syndrome. However, many disease associated features are absent indicating that the differentiation of TP63-related disorders should be revised.