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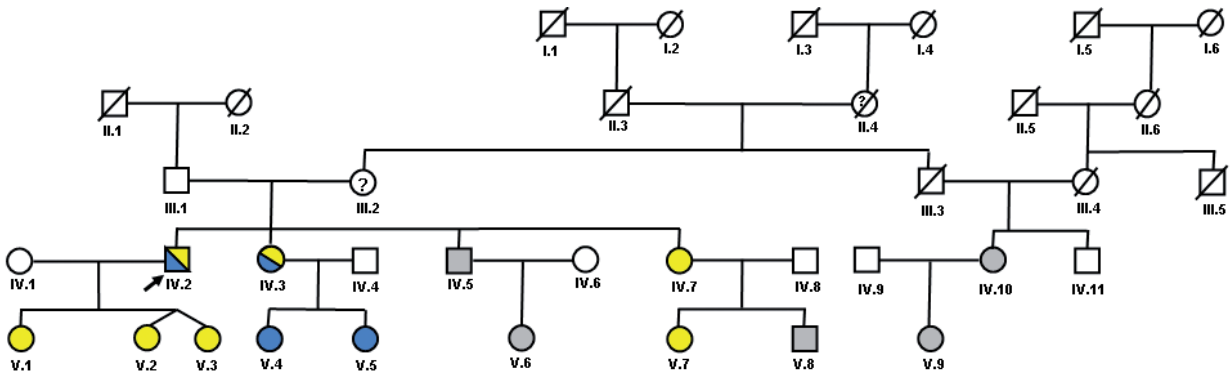

Fig. S1. Pedigree of the examined family members with Schöpf-Schulz-Passarge syndrome. *Squares*: males; *circles*: females. The *arrow* indicates the index patient (IV.2). *Coloured symbols* represent carriers of *WNT10A* mutations; *blue*: WNT10A p.Cys107*; *yellow*: WNT10A p.Arg248*. Examined family members carrying none of these mutations (*wild type*) are in *grey*. *Question marks* indicate individuals with unknown genetic status. The parents of the index patient, unfortunately, refused contact. His mother was reported to have symptoms on hands and feet. Her deceased mother, grandmother of patient 1, was reported to have had no dental anomalies, but had always suffered from sensitive hands and feet, and, despite wrapping her hands overnight and her husband being a shoemaker, hands including finger tips and feet had always fissured and skin could be peeled off in layers from her palms.

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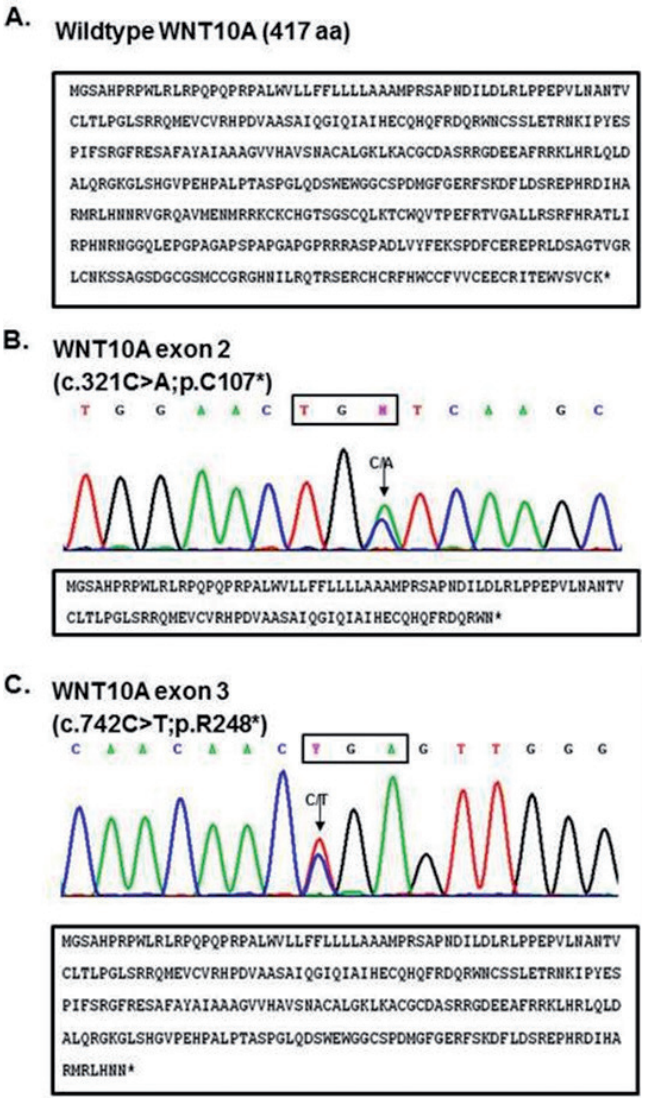

Fig. S2. Graphical illustrations of the WNT10A mutations detected. The WNT10A gene comprises four exons and is located on the long (q) arm of chromosome 2 (2q35). It encodes a protein of 417 amino acids belongs to a large family of secreted signalling proteins. (A) Wild-type protein sequence of *WNT10A* (417 aa). (B) Sequence map of exon 2 showing the c.321C>A; p.Cys107* mutation in the human *WNT10A* gene. (C) Sequence map of exon 3 showing the c.742C>T; p.Arg248* mutation in the human *WNT10A* gene. Both nonsense mutations introduce premature stop codons TGA (indicated by a *box*).

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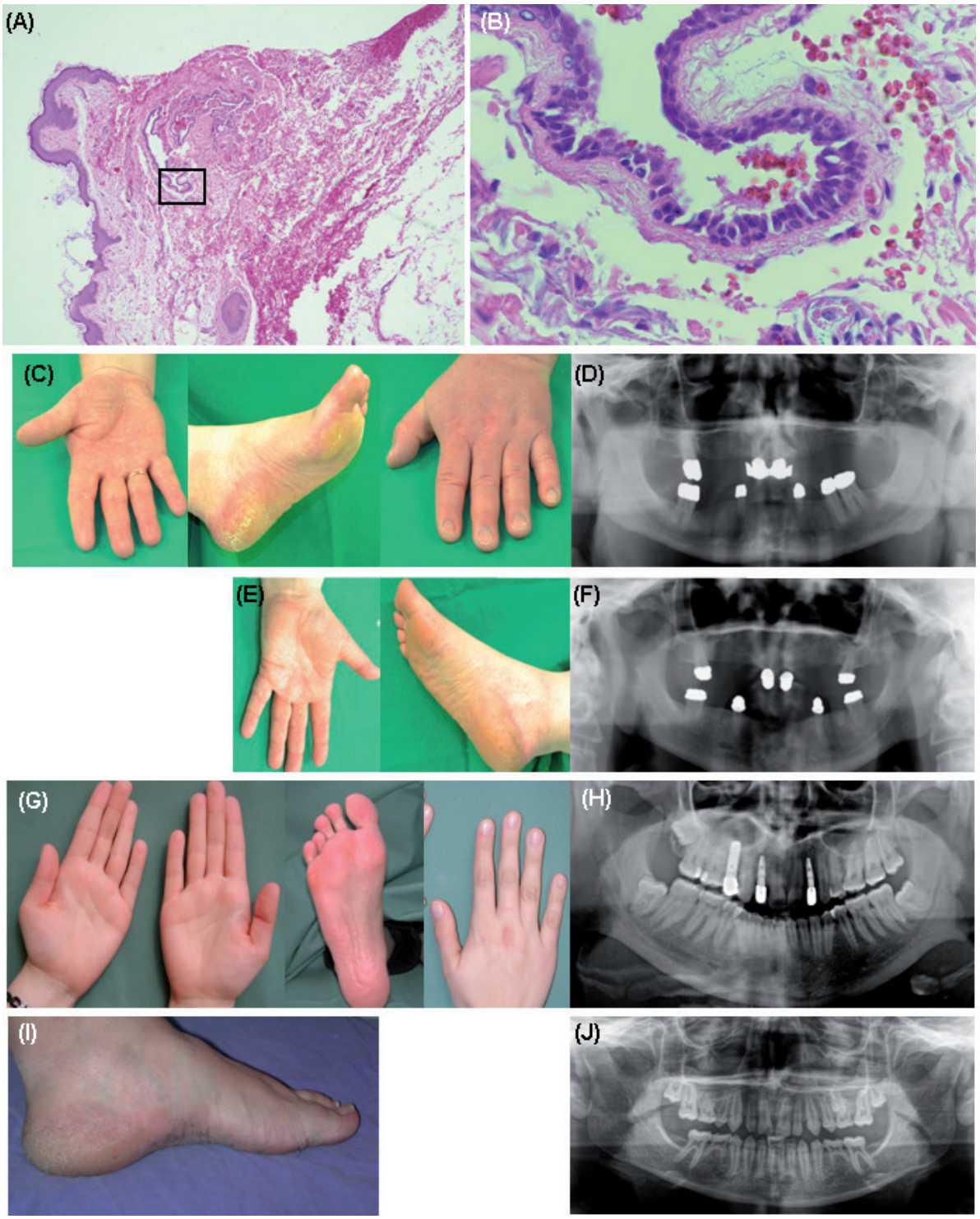

Fig. S3. Clinical and radiological findings in patients 1–5. (A–D) patient 1 (IV.2); (A) Overview and (B) detail of histology of lid lesions revealed apocrine hidrocystoma (haematoxylin (H&E) stain). (C) The skin of the palms and soles showed diffuse erythrokeratoderma with scaling particularly in pressure-bearing areas. All nails were fragile, some hypoplastic with longitudinal furrows and ridging. (D) Panoramic X-ray displaying 6 permanent teeth (16, 11, 21, 36, 35, and 47 according to the Fédération Dentaire Internationale tooth numbering system), 2 persistent deciduous teeth (73 and 83), but no periodontal or other pathology. All teeth were capped to support the upper and lower removable dentures. His beard hair appeared fine, his scalp showed androgenic pattern baldness, he had sparse and short eyelashes, almost missing eyebrows and scanty body and axillary hair (not shown). (E, F) Patient 2 (IV.3). (E) The skin of her nose and her palms and soles were erythematous with fine scaling. (F) Panoramic X-ray showing only 8 permanent teeth (17, 11, 21, 27, 37, 34, 44, and 47) all being capped to support upper and lower removable dentures. There was no history of periodontitis. (G, H) patient 4 (V.3); (G) Her palms and soles were mildly erythematous. The skin of her hands was very dry and would fissure easily without the daily use of skin lotion. In addition, the skin of her elbow crease was lichenified. Her finger nails and also her toe nails were very soft and could easily be pulled off. She therefore uses nail polish routinely to stabilize her nails. (H) Panoramic X-ray: 5 permanent teeth (15, 14, 12, 22, and 25) did not develop. Gaps had been closed orthodontically and by the insertion of dental implants. (I) Patient 3 (IV.7) with callused skin of the soles. Agenesis of 3 permanent teeth (14, 37, and 47) and no signs of periodontitis (not shown). (J) Patient 5 (V.7); panoramic X-ray showing agenesis of 5 permanent teeth (15, 37, 35, 45 and 47).

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Since this is a very extensive table, the format and content has not been edited by ActaDV.

Table SI. Characteristics of 38 reported cases of Schöpf-Schulz-Passarge Syndrome

Author	Year	N	Sex	Age	Cysts	Since age	Hypo-dontia	1° denti-tion	2° denti-tion	Comment	Hypo-trichosis	Since age	Palmo-plantar hyper-keratosis	Since age	Sweating	Since age	Oncho-dystrophy	Since age	Consan guinuity	Pedigree presented	Malignant tumors	At age
Schöpf et al.	1971	2	F	68	X	60	X	Lost early	X	Only 3 permanent teeth (12, 11, 48)	X	25	X	12			X	12	Yes	X		
			F	66	X	Late in life	X		X		X		X				X		Yes	X		
Burket et al.	1984	1	M	35	X	30	X	N.a.	X	Only 4 (rudimentary) permanent molars, persistent deciduous teeth until his late 20s, no hx of periodontitis			X	12			X	12	No			
Font et al.	1986	1	M	67	X	20	X			Fewer permanent teeth, most of them widely spaced, no hx of periodontitis			X	20			X		No		BCC nose	66
Nordin et al.	1988	3	F	80	X	65	X	X	X	4 lower deciduous teeth missing, lost at age 8-9, only 4 cone-shaped upper incisors (12, 11, 21, 22) developed	X		X	15	X	Always	X	Childhood	No	X		
			M	75	X	60	X			Small deciduous teeth, 2 lower permanent incisors missing, 2 upper permanent incisors cone-shaped, other permanent teeth small and widely spaced		20	X	15	X	Youth	X	Childhood				
			M		X								X				X				BCC? (clinical diagnosis)	71
Perret et al.	1989	1	M	75	X		X		X	Denture at age 18 because of hypoplastic teeth	X	20	X								SSC	67, 75
Monk et al.	1992	3	F	62	X		X	X	X	Primary dentition reported "abnormal", no permanent teeth, edentulous at exam; "bird-like facies"	X		X	47			X		Yes		Hypernephroma, BCC	
			M	58	X		X	X	X	"unusual" primary dentition, no permanent dentition, edentulous at exam; "bird-like facies"	X								Yes			
			M	51	X		X		X	One single permanent tooth only, edentuloue at exam, "bird-like facies"	X		X				X		Yes			
Küster & hammerstein	1992	1	F	43	X	43	X	N.a.	X	Most permanent teeth mising, first dentures at age 14, 5 permanent teeth present at exam	X	Childhood	X	20			X		No	X		
Craigen et al.	1997	1	M	56	X	54	X		X	No permanent teeth, total dental extraction at age 18	X	Life-long	X	Life-long			X	Life-long	No	X		
Starink	1997	4	F	78	X	77				Small conical widely spaced teeth	X			23			X		No		Malignant ESFA	
			F	59	X					At age 16 all teeth were removed, because of conical shape	X		X	20					No			
			F	73	X					Conical teeth	X		X	Childhood			X		No		BCC of nose, adenocarcinoma of breast	
			F	71	X		X		X	Only part of the permanent teeth developed	X		X	25			X		No			
Verplancke et al.	1998	1	M	53	X		X	N.a.	X	No permanent teeth developed	X	18	X	14			X		No			
Dot et al.	2000	1	M	71	X	55	X	X	X	Missing primary incisors and conical canines; only 8 permanent teeth, conical upper permanent canines	X	30	X	20			X	45	No		BCC, M. Bowen	45 and 70
Gkolfinopoulos et al.	2001	2	F	49	X		X	N.a.	X	Permanent teeth did not erupt, "bird-like facies", full dentures	X		X	25			X		No		No	
			M	56	X		X		X	Permanent teeth did not erupt (atrophic roots on OPG), full dentures	X	30	X	18	X	Child-hood	X		Yes		No	

Hampton et al.	2005	1	M	69	X		50	X			Full dental clearance @ age 14	X	15	X	20			X	10				
Castori et al.	2008	2	F	65	X		25 (?)	X	N.a.	X	Only 8 permanent teeth, "bird-like appearance"	X	Childhood	X	Childhood			X	Childhood	Yes	X		
			M	73	X		50	X	N.a.	X	Complete absence of permanent dentition	X	Early childhood	X	43	X	Adult-hood	X	Early childhood	Yes	X		
Bohring et al.	2009	1	F	25	X			X	X	X	17 permanent teeth missing (including wisdom teeth), 12 persistent deciduous teeth, 3 permanent teeth present	X		X		X		X				Porocarcinoma of left heel	
Szepetiuk	2009	1	M	56	X		46	X			"bird-like apppearance", dental alterations present "at birth", majority of permanent teeth absent	X		X	15			X	Birth	No		PMH: several SSC, BCC, s/p cardiac transplantation in 1992, med:immunosuppression	
Nagy et al.	2010	1	F	70	X		40	X	X	X	Reduced no. Of deciduous teeth, no permanent teeth	X	Childhood			X		X	Early adulthood	No			
Petrof et al.	2011	1	F	59	X		50	X	X	X	Reduced primary dentition, lack of secondary dentition			X	30-40	X	30-40	X		Unknown	Fostered child		
Wedgeworth et al.	2011	1	M		X			X		X	Severe hypodontia, dentures from the age of 14, conical-shaped teeth									No		Malignant melanoma, cutaneous SCC, BCC	
Tziotzios et al.	2014	7	F	58	X			X	X	X		X		X		X		X					
			M	50	X			X	X	X		X		X		X		X					
			M	43	X							X		X		X		X					
			F	36	X			X	X	X		X		X		X						BCC	
			M	57	X			X		X				X				X				Basosquamous CA	
			M	28				X	X	X				X		X		X					
			F	49	X			X		X		X		X									
Vilas-Sueiro et al.	2015	1	M	70	X		Many years	X		X	No 2nd teeth since childhood			X				X					
Painsi et al.	2017	1	F	86	X			X	X	X		X		X	Life-long	X	Childhood	X		No		Squamous cell CA of right palm excised	50
Total		**38**																					
Mean				**59,44**			**50**						**22**		**22,1**								
Min							**20**						**15**		**12**								
Max							**77**						**30**		**47**								
Median							**50**						**20**		**20**								
Present study	**2018**	**4**	**M**	**53**	**X**			**X**	**N.a.**	**x**	**Only 6 permanent teeth, 2 persisting deciduous teeth**	**X**	**17**	**X**	**20**	**N.a.**		**X**		no	**x**	**Basal cell carcinoma of cheek**	**50**
N.a. = no abnormalities			**F**	**51**	**X**			**X**		**X**	**Only 8 permanent teeth**	**X**		**(x)**		**Almost no sweating**		**X**	**50**	No	**X**		
			F	**46**	**X**									**X**						No	**X**		
			F	**25**				**X**		**X**	**Agenesis of 5 permanent teeth**			**X**				**X**		No	**X**		
			F	**8**				**X**		**X**	**Agenesis of 5 permanent teeth**									No	**X**		

The format and content has not been edited by ActaDV in this Table.

Table SII. Reported WNT10A mutations and genotypes in patients diagnosed with Schöpf-Schulz-Passarge syndrome. The most commonly identified mutation is c.321C>A;p.Cys107*. So far, three heterozygous and eight homozygous WNT10A genotypes resulting from 11 different mutations have been described.

								First allele		Second allele	
Autors	**Year of publi-cation**	**Number of patients**	**Homo-zygous**	**Compound hetero-zygous**	**Mis-sense**	**Non-sense**	**Exon**	**Nucleotide substitution**	**Amino-acid substitution**	**Nucleotide substitution**	**Amino-acid substitution**
Bohring et al.	2009	1	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Nagy et al.	2010	1	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Castori et al.	2011	1	X		X		4	c.796G>T	p.Gly266Cys	c.796G>T	p.Gly266Cys
		3	X		X		3	c.391G>A	p.Ala131Thr	c.391G>A	p.Ala131Thr
Petrof et al.	2011	1	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Wedgeworth et al.	2011	1	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Tziotzios et al.	2014	1	X			X	4	c.1168G>T	p.Glu390*	c.1168G>T	p.Glu390*
		1	X		X		4	c.1084T>C	p.Cys362Arg	c.1084T>C	p.Cys362Arg
		1		X	X	X	2,4	c.321C>A	p.Cys107*	c.810C>A	p.Ser270Arg
		1		X	X	X	2,3	c.321C>A	p.Cys107*	c.391G>A	p.Ala131Thr
		3	X			X	2	c.321C>A	p.Cys107*	c.321C>A	p.Cys107*
Vilas-Sueiro et al.	2015	1	X		X		3	c.682T>A	p.Phe228Ile	c.682T>A	p.Phe228Ile
					X		4	c.831G>T	p.Trp277Cys	c.831G>T	p.Trp277Cys
Painsi et al.	2017	1	X		X		4	c.1135C>T	p.Arg379Cys	c.1135C>T	p.Arg379Cys
Pauly et al.	2018	1		X	X		2,4	c.318C>G	p.Asn106Lys	c.1036T>C	p.Cys246Arg
present study	2018	2		X		X	2,3	c.321C>A	p.Cys107*	c.742C>T	p.Arg248*
		1				X	3			c.742C>T	p.Arg248*