

Table SI. Mutations in SLURP1 and clinical phenotype

Mutation	Consequence	References	Age, years	PPK	Trans-grediens	Hyperhidrosis	Nail deformities	Pseudo-ainhum	Elbow/knee-pads	Perioral erythema	Score
c.1A>C	p.Met1Leu	Missense Eckl 2003 (6)	23	2	2	0	1	1	2	0	8
c.43T>C	p.Trp15Arg	Missense Nellen 2009 (10) <sup>a</sup>	17	2	1	1	0	0	0	0	4
c.43T>C	p.Trp15Arg	Missense Nellen 2013 (4)	n/a <sup>b</sup>	2	1	1	0	0	0	0	4
c.43T>C	p.Trp15Arg	Missense Zhao 2014 (1)	n/a <sup>b</sup>	2	1	1	0	0/1 <sup>c</sup>	0	0	4/5
c.58+1G>C	c.58+1G>C	Missense Sakabe 2014 (11)	56	2	3	1	0	0	1	0	7
c.82delT	p.Cys28fs*32 deletion	Bchetnia 2013 (12)	40	2	3	1	1	0	1	1	9
c.129C>A	p.Cys43Ter	Nonsense Muslumanoglu 2006 (13)	15	2	1	1	1	1	1	1	8
c.212G>C	p.Arg71Pro	Missense Nellen 2009 (10) <sup>a</sup>	17	2	1	1	0	0	0	0	4
c.244C>T	p.Pro82Ser	Missense Gruber 2011 (14)	27	1	2	1	0	0	0	0	4
c.256G>A	p.Gly86Arg	Missense Bchetnia 2014 (5)	28	2	1	1	0	0	0	0	4
c.256G>A	p.Gly86Arg	Missense Chao 2006 (15)	22	2	1	0	1	1	0	0	5
c.256G>A	p.Gly86Arg	Missense Oh 2011 (16) <sup>d</sup>	15	2	1	1	0	0	1	0	5
c.256G>A	p.Gly86Arg	Missense Taylor 2014 (17)	40	2	2	1	0	1	0	0	6
c.256G>A	p.Gly86Arg	Missense Zhang 2015 (7) <sup>e</sup>	4	2	2	1	0	0	2	0	7
			2	2	2	1	0	0	2	0	7
			11	2	2	1	1	0	1	0	7
c.286C>T	p.Arg96Ter	Nonsense Oh 2011 (16) <sup>d</sup>	15	2	1	1	0	0	1	0	5
c.293T>C	p.Leu98Pro	Missense Yerebakan 2003 (8)	8	2	1	1	0	0	1	0	5
c.296G>A	p.Cys99Tyr	Missense Bchetnia 2010 (9)	30	2	3	1	0	0	0	0	6
c.296G>A	p.Cys99Tyr	Missense Bchetnia 2013 (12)	13	2	1	0	1	0	0	1	5

<sup>a,d</sup>Patients were compound heterozygous for the p.Trp15Arg and p.Arg71Pro mutations (<sup>a</sup>) and p.Gly86Arg and p.Arg96Ter mutations (<sup>d</sup>), respectively.

<sup>b</sup>Age could not be determined. Both publications describe a group of MDM patients, and a general description of the phenotypes of inflicted patients is given. We computed the disease severity score based on these summarized clinical data. "Some, but not all, patients reported by Zhao et al. had pseudo-ainhum, which was not further specified in the description of their patients' clinical characteristics. Therefore, we scored "pseudo-ainhum" as 0/1. <sup>c</sup>Zhang et al. described 3 patients in great detail. A severity score was computed for each individual.

Resumé of clinical characteristics in reported patients of MDM, in whom *SLURP1* mutations have been identified. Papers describing series of patients, without individual descriptions of each patient have been omitted. Symptoms have been semi-quantitatively scored as follows: Age: age of patient at the time of examination (years); PPK: palmoplantar keratoderma, scored as mild (1) or severe (2); Transgrediens: extension of hyperkeratosis to the dorsal surfaces of hands and feet, scored as extending to fingers (1), dorsa of hands/feet (2), fore-arms/lower legs (3); Hyperhidrosis: hyperhidrosis, scored as either present (1) or absent (0); Nail deformities; nail deformities, such as subungual hyperkeratosis and nail dystrophy, scored as either present (1) or absent (0); Pseudo-ainhum; scored as either present (1) or absent (0); Elbow/knee-pads: hyperkeratotic plaques on elbows and knees (scored as 2), elbows or knees (scored as 1) or absent (scored as 0); perioral erythema: perioral skin symptoms, such as erythema, scaling or fissures, scored as either present (1) or absent (0). Score: adding numbers together yields a score that represents disease severity, with a higher score indicating a more severe disease course.