Table SI. Skin manifestations of patients with holocarboxylase synthetase deficiency (HCSD)

Gene mutation	Ethic	Sex	Onset	Skin lesions (age at observation)	Ref.
p.L216R/p.L216R	Australian/Maori	M	3 d	Severe eczema involving the trunk, nappy area, scalp, facial areas and auditory canals (NR)	S1
	Samoan	F	0 d	Erythematous rash on the buttock (early infancy)	S2
	Samoan	F	0 d	Erythematous rash in the nappy area and intertriginous regions (early infancy), persistent rash at the outer canthi (2 y)	S2
p.L237P/c.780delG	Japanese	F	2 d	Psoriatic dermatitis (6 m)	This case
	Japanese	M	1 d	Alopecia (22 d), non-bullous congenital ichthyosiform erythrodermatitis-like lesions (22 d), periorificial dermatitis (4 m), seborrhoeic dermatitis (5 y 6 m), psoriatic dermatitis (5 y 6 m)	S3
c.1053_1054insC/p.T462I	Spanish	F	1 d	Erythematous and cracked skin (0 d)	S4
p.R565X/p.G326E	NR	F	NR	Alopecia (2 y)	S5
p.V363D/c.1681+1G>A	NR	F	1 y 4 m	Periorificial and intertriginous dermatitis (16 m)	S6
p.R508W/p.R508W	Chinese	M	11 m	Generalized pustular psoriasis (11 m)	S7
	Chinese	M	1 y 6 m	Generalized skin rash involving the trunk, limbs and face, particularly the mouth corners and lower eyelids (6 y)	S8
	Thai	F	8 m	Dermatitis around the mouth, eyes, neck and perineum, but none around the nose area (NR)	S9
p.R508W/p.G505R	Thai	F	1 m	Dermatitis around the mouth, eyes, neck and perineum, but none around the nose area (NR)	S9
p.R508W/U	Thai	M	9 m	Dermatitis around the mouth, eyes, neck and perineum, but none around the nose area (NR)	S9
	NR	M	5 m	Erythematous rash on the buttocks and neck gradually spread to all flexures (6 w)	S10
p.N511I/p.G693A	Hispanic	M	1 d	Alopecia, atopic dermatitis (6 m)	S5
p.V550M/p.V550M	Jewish/Turkish	M	NR	Scaly red skin eruption mainly in skin creases diagnosed as seborrheic dermatitis with candidiasis (3 m)	S10
p.G581S/p.V550M	Italian	M	5 m	Erythematous dermatitis localized in the diaper and intertriginous areas (5 m), diffuse erythematous, scaly and exudative skin lesions over the entire body (11 m)	S1
p.N511K/p.G582R	Italian	M	6 m	Erythematous squamous exudative lesions in the armpit, buttock, and neck areas (6 m), facial, axillary and gluteal dermatitis (23 m)	S1
c.1992delC/IVS10+5G>A	French	F	5 m	Alopecia and periorificial dermatitis (8 m)	S11
p.Y663H/p.Q379X	NR	F	0 d	Taut, shiny, and thickened skin with a cellophane-like appearance and perioral creases (0 d)	S12
NR	Caucasian	F	0 d	Ichthyosis (0 d)	S13
NR	NR	M	15 d	Desquamative dermatitis of the trunk, axillary fold, neck, and face (15 d)	S14
NR	Caucasian	F	6 m	Periorificial dermatitis (4 y)	S15

NR: not recorded; U: unidentified; M: male; F: female; d: days; m: months; y: years.

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