Table 1 Steps of ULC-acylceramide synthesis and CLE formation, the involved genes and diseases caused by their mutations

Step of ULC-acylceramide synthesis and CLE formation	Involved gene (molecule)	Related disease
elongation of fatty acids	*ELOVL6, 3, 7, 1* (ELOVL6, 3, 7, 1) *ELOVL4* (ELOVL4)	- ichthyosis, intellectual impairment, spastic quadriplegia
ω-hydroxylation of ultra-long-chain fatty acids	*CYP4F22* (CYP4F22)	LI
CoA addition to ultra-long-chain fatty acid	*SLC27A4* (FATP4)	ichthyosis prematurity syndrome
ceramide synthesis	*CERS3* (CERS3)	LI
ω-O-esterification with linoleic acid	*ABHD5* (ABHD5) *PNPLA1* (PNPLA1)	Dorfman-Chanarin syndrome LI
glucosylation of ceramide	*UGCG* (UGCG)	-
transport via lamellar granules	*ABCA12* (ABCA12)	HI, LI, CIE
oxidation of linoleic acid in ceramide	*ALOX12B* (12R-LOX) *ALOXE3* (eLOX3)	LI, CIE, pleomorphic ichthyosis
hydrolysis of oxidized linoleic acid	unknown lipase	-
covalent linking of ceramide to the outer surface of CCE	*TGM1* (TGase 1)*	-
deglucosylation of glucosylceramide	*GBA* (glucosylceramidase β)	Gaucher disease

*The role of TGase 1 in the covalent linking of ceramide to CCE remains controversial.