

Table SI. Reported missense mutations of the first glycine in the first extracellular domains of the connexin gene family

Gene	Protein	Substitution	Type of mutation	Phenotype	Ref.
<i>GJB2</i>	Cx26	p.Gly45Glu	Germline	KID syndrome	6, 7
		p.Gly45Glu	Somatic	PEODDN	8
<i>GJB3</i>	Cx31	p.Gly45Glu	Germline	EKV	4, 5, this case
<i>GJA8</i>	Cx50	p.Gly46Arg	Germline	Congenital cataract and microcornea	11
		p.Gly46Val	Germline	Congenital cataract and microcornea	12

Cx; connexin; KID; keratitis, ichthyosis and deafness; PEODDN: porokeratotic eccrine ostial and dermal duct naevus; EKV: erythrokeratoderma variabilis.