Table SII. Autosomal dominant syndromic genodermatoses associated with heterozygous *GJB2* mutations

Name/description	Onset	SNHL	Palmoplantar keratoderma	Other skin features	Hair and nails	Mucous	Eyes	Infections	Other
Vohwinkel syndrome	Infancy	+	Diffuse, focal or striate, with honeycomb pattern; transgrediens; pseudoainhum and spontaneous auto-amputations of the digits	Starfish keratoses on knuckles and wrists, alopecia	Alopecia, nail dystrophy	–	–	–	Hyperhidrosis
Burt-Pumphrey syndrome	Early childhood	+	Diffuse, focal or punctate hyperkeratosis	Knuckle pads	Leukonychia	–	–	–	Breast and axillary cysts
Palmoplantar keratoderma with deafness	Infancy to early childhood	+	Diffuse or focal	–	–	–	–	–	–
Keratitis ichthyosis deafness syndrome (KID/HID)	Birth or infancy	+	Diffuse hyperkeratosis	Ichthyosiform erythrokeratoderma or hystrix-like ichthyosis	Cicatricial alopecia, scarce body hair	Oral leukokeratosis	Keratitis, corneal ulcerations, blepharitis	Chronic bacterial and fungal infections	High risk of SCC
Clouston's syndrome-like	Shortly after birth	+	–	Atopic eczema, sharply demarcated erythematous plaques with some desquamation on scalp	Mild scalp hypotrichosis with lank, blonde hair; nail dystrophy	Redness and swelling of the oral mucosa and gingiva; perianal erythema	–	–	Developmental delay
Keratoderma-deafness-mucocutaneous syndrome	Birth or infancy	+	Focal hyperkeratosis (not constant)	Erythematous patches and plaques; acne; calcinosis cutis	Alopecia, nail dystrophy	Angular cheilitis, oropharyngeal and gastrointestinal inflammation	–	Bacterial and fungal cutaneous and mucous infections	SCC (1 case)

SCC: squamous cell carcinoma; SNHL: sensorineural hearing loss.