Table SI. Features of patients with hearing loss and unusual mucocutaneous findings due to Phe142Leu in *GJB2*

	Patients				
Characteristics	Family 1, Patient II;1	Family 2, Patient II;1	Family 3, Patient II;1	Family 3, Patient I;2	Family 4, Patient II;1
Reference	Brown et al., 2003 (6)	Rednam et al., 2011 (7)	Ibanez et al., 2013 (8)	Ibanez et al., 2013 (8)	Present case
GJB2 gene (NM_004004.5)	c.424T>C	c.424T>C	c.426C>A	c.426C>A	c.426C>A
Connexin 26 (NP_00399.2)	p.(Phe142Leu)	p.(Phe142Leu)	p. (Phe142Leu)	p.(Phe142Leu)	p.(Phe142Leu)
Inheritance	Het; de novo	Het; de novo	Het; mat	Het; NA	Het; *de novo*
Age at description, years	2	6	Birth	23	41
Sex	F	F	F	F	F
Sensorineural hearing loss	+	+	+	+	+
Cutaneous involvement					
Lesions at birth	–	–	+	–	+
Psoriasiform dermatitis	+	+	–	–	+
Acne with cicatricial sequelae	–	–	–	–	+ (adulthood)
Palmoplantar keratoderma	–	–	–	–	+
Calcinosis cutis	+ (both heels)	–	–	–	–
Erythematous lesions	+ (periorificial, face, trunk upper extremities and diaper areas)	–	+ (intertriginous areas, scalp)	+ (head, neck, trunk)	+ (face, trunk, limbs, periorificial)
Whitish lesions	–	–	+ (papules on the scalp and upper limbs)	+ (external auditory canal, labial commissures and subauricular areas)	–
Skin histopathology					
Hyperkeratosis	+ (orthokeratotic)	NA	+ (epidermal and follicular)	NA	+
Parakeratosis	+ (focal)	NA	+ (epidermal and follicular)	NA	+ (focal)
Acanthosis	+	NA	–	NA	–
Papillomatosis	+	NA	–	NA	–
Foci of acute and chronic inflammation	+ (superficial derma and epidermis)	NA	+ (perifollicular inflammatory infiltrate)	NA	+ (superficial derma and epidermis)
Adnexal involvement					
Alopecia	+	–	+	–	–
Nail dystrophy	–	–	+	–	–
Oropharyngeal involvement					
Multiple dental lamina cysts	+	–	–	–	–
Teeth anomalies	+ (focal enamel hypoplasia of primary teeth)	NA	NA	NA	+ (premature loss of permanent teeth)
Angular cheilitis	+	–	+	–	+
Gingivitis	+	+	–	–	–
Erythema, focal ulceration, adherent white plaques	+	–	–	–	+
Endoscopy of upper gastrointestinal tract					
Inflammation of oesophageal mucosa	+ (denuded and friable mucosa)	+ (esophagitis)	+ (parakeratosis)	+	–
Oesophageal stricture	+	–	–	+ (oesophagocoloplasty)	–
Tumoral lesions					
Squamous cell carcinoma	–	+ (hard palate)	–	–	–
Recurrent infections	+ (urinary tract infections, perirectal abscesses, otitis)	+ (perirectal abscesses)	+ (2episodes of sepsis)	–	+ (candidiasis)
Other	Iron deficiency anaemia, reactive thrombocytosis; mild delay in myelination	–	Double left excretory renal system; anaemia and transient neonatal hypocalcaemia; cyst of choroid plexus	–	Verrucous haemangioma; lymphocytopaenia with decreased CD4, CD8 and natural killer cells

+ Present; – absent; het: heterozygous; mat: maternal; NA: not available; F: female; pat: paternal. In the family by Ibanex et al. (8) the father, homozygote c.35delG in *GJB2*, transmitted this additional mutation to his daughter.