Table 1 Management of ocular and ear–nose–throat (ENT) complications of congenital ichthyosis: recommendations with level of evidence and grade

Recommendations	Level of evidence	Grade	Type of recommendation	Details of the changes (if applicable)	References
Ophthalmic complications					
Monitoring					
• Regular monitoring from monthly to once or twice a year	4	D	UR	NA	
Ocular lubrication					
• Regular and long-term ocular lubrication with preservative-free topical medication if lagophthalmos (from once daily to half-hourly)	4	D	UR	NA	27, 36, 41
Ectropion					
• First-line treatment: ocular lubrification	4	D	UR	NA	42–45
• Emollients and massage of the eyelid	3	D	UR	NA	42–45
• Keratolytics on the eyelid (urea or *N*-acetylcysteine or topical tazarotene) must be tried before surgery	3	B	**MR**	Addition: 'must' be tried before surgery	46–53
• Oral retinoids: second-line therapy in cases of moderate-to-severe ectropion helps to reduce moderate-to-severe ectropion and prevent worsening	3	B	UR	NA	54–56
• Eyelid skin grafting: third-line therapy, may only be considered when symptomatic corneal exposure or epiphora persists despite adequate conservative treatments	3	B	UR	NA	57–65
• Hyaluronic acid gel filler injections may be beneficial. They should only be undertaken by experienced ophthalmologists	3	B	UR	NA	67, 68
ENT complications					
• Removal of earwax/cerumen by using eardrops, oil and mechanical techniques one to four times a year	3	D	UR	NA	69–71, 76–78
• Oral retinoids are not primary treatment					
• External otitis: cleansing and debridement measures associated with topical antibiotics	4	D	UR	NA	79
• Regular use of oils can protect the external auditory canal	4	D	UR	NA	71, 73–75
• If voice anomalies are detected, referral to a speech therapist to improve dysphonia and an otolaryngologist to visualize the vocal cords is warranted	2	D	**NR**	New item (voice anomalies)	69
• Regular hearing evaluations in case of ear canal obstruction: every 6 months for children younger than 6 years, yearly for adults	4	D	**MR**	Two additions: the mention of 'in case of ear obstruction', the frequency for adults	69, 78
• ENT referral in case of pruritus or pain of the ear, ear discharge, feeling of clogged ears or hearing loss	4	D	UR	NA	69, 70, 72

Adapted from the SIGN50 Guideline Developer's Handbook, NHS Scottish Intercollegiate Guidelines Network, revised edition 2019 (https://www.sign.ac.uk/our-guidelines/sign-50-a-guideline-developers-handbook). ENT, ear–nose–throat; MR, modified recommendations; NA, not applicable; NR, new recommendations; UR, unchanged recommendations;

We recommend that both children and adults with recurrent ear blockage undergo follow-up with an ENT specialist, including a hearing evaluation. For children, this should occur at least every 6 months, with the frequency adjusted as needed in adults and in cases of mild symptoms.[68,78]

External otitis resulting from ear canal obstruction can cause pain, discharge or even tympanic membrane damage, potentially leading to permanent hearing loss.[79] Management consists of cleansing and debridement, followed by use of topical medication (antibiotic drops, e.g. ciprofloxacin with or without corticosteroids).

Voice anomalies, affecting around 14% of CI cases, can interfere with speech and heighten the risk of psychosocial and developmental issues.[69] Congenital hoarseness sometimes persisting into childhood, requires referral to a speech therapist for dysphonia and an otolaryngologist for the evaluation of the vocal cords.[69]

In cases of pruritus or pain in the ear, ear discharge, a feeling of clogged ears or hearing loss, referral to an ENT specialist should be considered.

Pruritus and pain (action: update)

Pruritus and pain in CI are likely linked to skin inflammation.[80] Pain contributes to skin tightness, fissures and joint contractures, common concerns that significantly impact QoL but are not systematically assessed.[3,81]

Reducing pruritus and pain requires improving skin lesions through optimized topical (including cooling effects from wet-wrapping) and systemic therapies (such as oral retinoids).[4] While oral retinoids may offer antipruritic benefits, they can also cause irritation that worsens itching.[56,82–86] Antihistamines and antidepressants generally have little to no impact on pruritus,[87] except for sedative antihistamines, which may aid sleep.

Analgesics may be used for pain management, following guidelines established for treating pain in epidermolysis bullosa, other skin diseases[88–90] or pain in general (http://apps.who.int/iris/bitstream/10665/44540/1/9789241548120_Guidelines.pdf). Although very limited data are available, biofeedback may be useful.[91]