Non-syndromic ichthyoses

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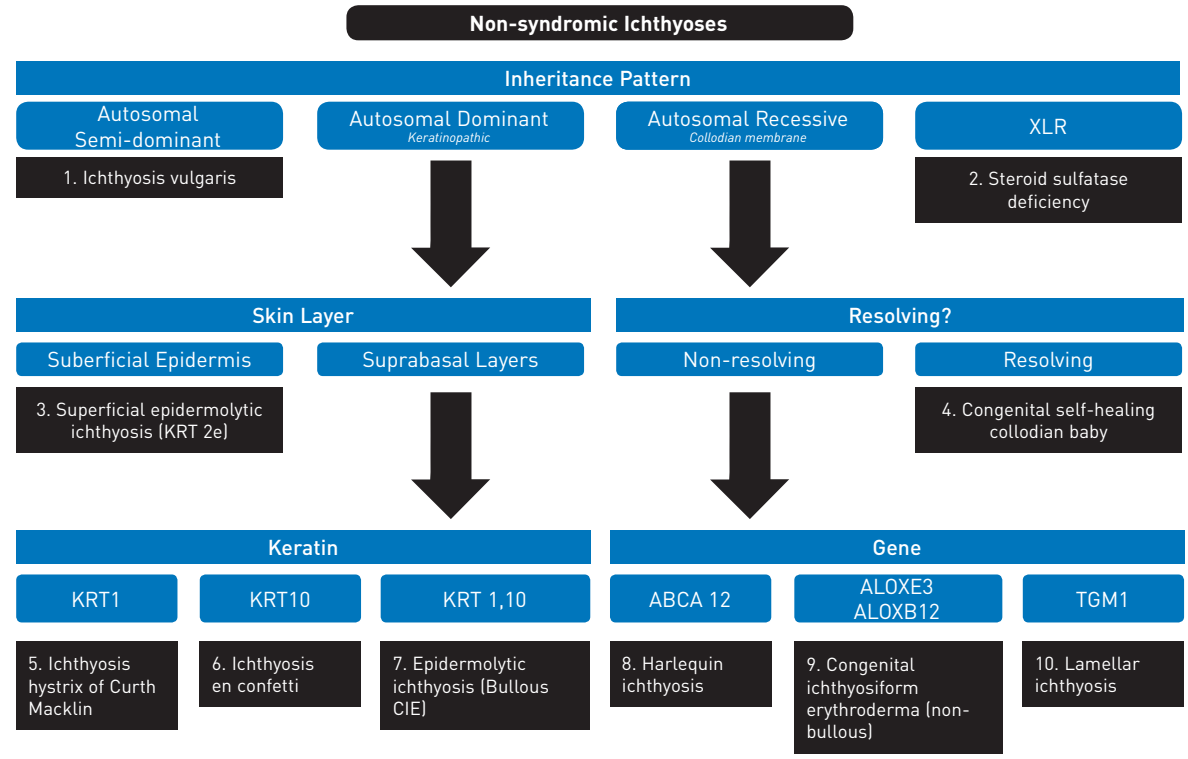

Disease	Gene/Onset	Unique Qualities
1. Ichthyosis vulgaris	Gene: FLG Onset: Infancy or childhood	* Spares flexures * Hyperlinearity of palms/soles * Keratosis pilaris/Atopic diasthesis * Fine adherent scale on extremities and trunk
2. Steroid sulfatase deficiency	Gene: STS or arylsulfatase C deficiency Onset: Infancy	* If continuous gene deletion of STS occurs = Kallman syndrome * "Dirty" brown scale * Spares flexures * Comma-shaped corneas (only finding found in female carriers) * Prolonged labor secondary to placental sulfatase deficiency * Decreased serum estriol in pregnancy * Increased risk of testicular cancer * cryptorchidism
3. Superficial epidermolytic ichthyosis *Aka Ichthyosis bullosa of Siemens*	Gene: KRT2E Onset: Birth	* Erythroderma + superficial blistering at birth * Mauserung phenomenon – German for molting; superficial shedding of skin with palms/soles spared
4. Congenital self-healing collodion baby	Gene: TGM1, ALOXE3, ALOXB12 Onset: Birth	* After resolution, skin appears normal

Non-syndromic ichthyoses *(continued)*

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Disease	Gene/Onset	Unique Qualities
5. Ichthyosis hystrix of Curth Macklin *Aka Ichthyosis hystrix*	Gene: KRT1 Onset: Early childhood	* Diffuse or striate PPK * "porcupine quill"-like verrucous yellow-brown scaling, esp on hand and feet * Digital constriction/pseudoainhum PPK
6. Ichthyosis en confetti *Aka Congenital reticular ichthyiosiform erythro-derma or Ichthyosis variegata*	Gene: KRT10 Onset: Birth	* At birth, erythroderma * After birth, confetti-like areas of scaling * Joint contractures * Hypoplasia of mammillae * Dorsal acral hypertrichosis
7. Epidermolytic ichthyosis *Aka Bullous congenital ichthyosiform erythroderma*	Gene: KRT1, KRT10 Onset: Birth	* Erythroderma with blisters and erosions at birth * After birth, hyperkeratosis + cobblestone appearance with predominance over joints * Retinol exacerbates skin fragility * Malodorous with frequent skin infections * Gait and posture abnormalities * Somatic mosaicism (extensive epidermal nevus aka ichthyosis hystrix) vs gonadal mosaicism (offspring with full expression of disease)
8. Harlequin ichthyosis	Gene: ABCA12 Onset: Birth	* Extreme eclabium and ectropion * Early initiation of systemic retinoids reduces mortality * Death is secondary to sepsis and respiratory insufficiency * Ear deformities
9. Congenital ichthyosiform erythroderma (non-bullous)	Gene: ALOXE3, ALOX12B Onset: Birth	* After collodion membrane resolves, develop white fine scale in generalized distribution (Flexures involved) - like "snow flakes" * Erythroderma as background (hence not bullous) * +/- palm/soles * Heat intolerance and hypohydrosis * +/- scarring alopecia and ectropion
10. Lamellar ichthyosis	Gene: TGM1 Onset: Birth	* Thick plate-like brown scale with significant flexure involvement * Ectropion/eclabium * Heat intolerance * Scarring alopecia

References:

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2. Paller, A. S. Mancini, A.J. (2016) *Hurwitz Clinical Pediatric Dermatology*. [E-reader version]. Retrieved from https://expertconsult.inkling.com: Elsevier

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