THE INCIDENCE, MANAGEMENT AND EARLY OUTCOMES OF CONGENITAL ICHthyosis

Abstract
The term ichthyosis refers to a group of incurable genetic conditions, the most severe of which is harlequin ichthyosis with a neonatal mortality rate of up to 44%. The appearance of HI at birth can be so shocking that some health professionals express reluctance to provide ongoing care. Less severe cases present as a collodion membrane, which can improve with time and even resolve, but more often evolves into a life-long ichthyosis which significantly impairs quality of life for sufferers and their families. There is no proven correct treatment, so the neonatal management of these babies varies from admission to the neonatal intensive care unit with multiple interventions to care delivered in a more normal setting.

The aim of the study is to establish the number of new cases and early death rates in the cohort of babies born in the UK and Ireland between 1st November 2018 and 31st October 2020. We will record any key interventions, and outcomes. Ultimately, we aim to improve care for children and families with severe ichthyosis, reducing misconceptions about the disease, variation in practice and avoidable deaths.

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Website
www.rcpch.ac.uk/bpsu/ichthyosis

Coverage
United Kingdom and Republic of Ireland

Duration
November 2018 to November 2020 (13 months of surveillance) with a 12-month follow-up

Research Questions
- Determine the birth incidence in the UK and other participating countries and report the distribution by sex, ethnic group and parental consanguinity.
- Describe variation in patient journey, including circumstances of birth, transfer to NICU or specialist centre, investigations, management and specialist consultations.
- Describe variation in other maternal and fetal factors relevant to outcome including family history, parity, gestational age and condition at birth.
- Describe variation in outcome including final diagnosis, morbidity, mortality, age at discharge home and other milestones.
Case definition

Any suspected cases of severe congenital ichthyosis in live newborn or still-born babies. This includes babies with collodion membrane (a shiny film covering the skin) or harlequin ichthyosis (thick scales encasing the babies’ body).

Collodion membrane and harlequin ichthyosis both comprise a tight layer of abnormal skin, present at birth, affecting the whole skin surface.

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<tr>
<th></th>
<th>Collodion membrane</th>
<th>Harlequin ichthyosis</th>
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<tbody>
<tr>
<td><strong>Consistency</strong></td>
<td>Thin and moderately flexible, like a coat of lacquer or collodion</td>
<td>Thick and rigid, like plastic</td>
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<td><strong>Pattern of cracking</strong></td>
<td>Mostly smooth at birth but cracks progressively with neonatal movements, particularly across major joints e.g. in the groins</td>
<td>Broad irregular cracks already present at birth, in a pattern likened to a harlequin suit, reflecting growth of the fetus within a rigid casing.</td>
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<td><strong>Restriction of limb movements</strong></td>
<td>Minimal, although digits may be temporarily fixed in flexion</td>
<td>Significant restriction with fingers and toes firmly fixed in flexion.</td>
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<td><strong>Facial distortion</strong></td>
<td>Facial features are mildly distorted by the tight membrane. There is usually some degree of ectropion and eclabium,</td>
<td>Facial features are significantly distorted by the tight membrane: severe ectropion with complete eversion of the upper lids, marked eclabium, flattened nose, occluded nares</td>
</tr>
<tr>
<td><strong>Retroaural folds</strong></td>
<td>Present</td>
<td>Usually absent</td>
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Reporting instructions

Please report any child seen in the last month who meets the case definition in the UK or the Republic of Ireland.

Methods

Each paediatrician reporting a child who meets the above case definition of severe congenital ichthyosis in live new-born or still-born babies will be sent a clinical questionnaire by the study team, which explores demographic and clinical information about the affected neonate. An initial questionnaire will be completed at 30 days of age and the second at 12 months of age (that is 11 months after the first).

Throughout the study, all patient data will be dealt with in strict confidence, and families will not be contacted directly by the study team at any stage.

Ethics approval

The study has been approved by West Midlands - Black Country Research Ethics Committee (reference: 18/WM/0211); HRA Confidentiality Advisory Group (reference: 18/CAG/0105); and Public Benefit and Privacy Panel for Health and Social Care (PBPP) approval in Scotland (PBPP reference: 1819-0098).

Support group

Ichthyosis Support Group

Funding

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References