Development of a disease severity score for newborns with collodion membrane

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Background: Collodion membrane in the neonate may be the initial presentation of a number of different conditions. There is a lack of data correlating the extent of clinical involvement to the underlying disease and prognosis.

Objective: We sought to identify features predictive of the final outcome and complications in a cohort of patients with collodion membrane, using a disease severity score.

Methods: This was a retrospective cohort study of newborns with collodion membrane at a tertiary care institution over a period of 31 years. We designed and applied a 0- to 15-point severity score and correlated the results with the final diagnoses and complications. Data on demographics, membrane shedding, and treatment were collected.

Results: We identified 29 cases. Congenital ichthyosiform erythroderma and lamellar ichthyosis were the most common final diagnoses with 7 of 29 cases (24%) each; 3 patients were given the diagnosis of a syndromic ichthyosis. The classic nonsyndromic ichthyoses had higher average score results (7.33) than the syndromic ichthyoses (2.0) and other presentations (4.0), (P = .0097). Patients with major complications had higher, but nonsignificant, average severity scores (P = .64).

Limitations: The retrospective design and small number of patients with a syndromic ichthyosis are limitations.

Conclusions: Prospective studies are required to validate the proposed disease severity score. (J Am Acad Dermatol 2014;70:506-11.)

Key words: collodion baby; collodion membrane; congenital ichthyosis; disorders of cornification; neonatal diseases; nonsyndromic ichthyosis; syndromic ichthyosis.

Collodion membrane describes the congenital and transient keratinized membrane that precedes a disorder of cornification in the majority of cases.[1-6] A parchment-like membrane wraps the neonate's body and may cause ectropion, eclabium, flattening of the ears and nostrils, restricted extension of the digits and extremities, and limitation

Abbreviations used:

ARCI:	autosomal recessive congenital ichthyosis
CIE:	congenital ichthyosiform erythroderma
HI:	harlequin ichthyosis
LI:	lamellar ichthyosis
RXLI:	recessive X-linked ichthyosis
UCI:	unspecified congenital ichthyosis

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of chest movement.[1-7] It spares the mucosal surfaces and stops at the edge of natural orifices and at the cutaneous level of the umbilical cord; hairs usually penetrate the collodion membrane and nails are exceptionally covered.[1,2] The membrane gradually peels off during the first 4 weeks, but may not completely disappear until the third month of life.[1,2,4,6]

The collodion membrane phenotype is related to several underlying disorders and molecular abnormalities. The most common are the autosomal recessive congenital ichthyoses (ARCI).[1-19] Infrequently reported associations include epidermolytic ichthyosis,[4] hypohidrotic ectodermal dysplasia,[20] ichthyosis vulgaris,[1,7] Netherton syndrome[1] (although this association has been questioned),[21] Sjögren–Larsson syndrome,[4] koraxitrachitic syndrome,[22] and palmoplantar keratosis with leukokeratosis anogenitalis[23] (there are no identified gene abnormalities for the last 2 syndromes). Harlequin ichthyosis (HI) is recognized as a severe form of collodion membrane in the revised nomenclature and classification of inherited ichthyoses published in 2010.[11] Other reported associations include hypothyroidism and asymmetric crying face caused by congenital hypoplasia or agenesis of the depressor anguli oris muscle.[24-28]

Although the collodion membrane phenotype is well recognized, there is no consensus about the relevance of the severity of the initial presentation as a predictor of the final outcome. Many consider that the clinical findings do not predict a particular diagnosis or outcome.[2,4,8] Anecdotally, patients presenting with limited collodion membrane are less likely to develop one of the classic ichthyosis phenotypes. The purpose of our study was to explore the significance of the extent of the collodion membrane and its associated features as predictors of the underlying disease, severity, and complications.

CAPSULE SUMMARY

- Collodion membrane is a neonatal presentation of both syndromic and nonsyndromic ichthyoses.
- This study proposes a severity score to correlate the extent of the collodion membrane with the underlying diagnosis.
- Validation of the score is needed to confirm its value.

METHODS

A retrospective cohort study was performed involving patients who presented with collodion membrane at our institution from January 1, 1981, to December 31, 2011. The study was approved by the hospital's research ethics board. We collected medical record data on demographics, clinical characteristics, complications, date of membrane shedding, laboratory investigations (including molecular and genetic testing), treatment, and final clinical outcome. We included both patients with the classic collodion baby phenotype and infants with HI, presenting as severe collodion membrane with an armor-like appearance at birth and evolving to congenital ichthyosiform erythroderma (CIE).[11,29]

We designed a disease severity score (range: 0-15) including 15 clinical items where 0 represented absence and 1 represented presence of the item (Table I). We counted involvement of the upper extremities, fingers, lower extremities, and toes separately because the degree of limitation could indicate variable clinical severity. Hypernatremia was included as it reflects the overall loss of epidermal barrier function in the regulation of fluid balance. The score was applied to the cohort of patients and the results were analyzed.

Statistical analysis

Demographic and clinical data were summarized and reported as means with SD and frequencies. An analysis of variance was performed to compare the severity score among the groups. A P value of .05 was used to indicate statistical significance. Analysis was performed using software (Minitab, Version 14.0, Minitab Inc, State College, PA).

RESULTS

We identified 29 patients, of whom 17 of 29 (59%) were male. The mean age at diagnosis was 1.9 days (SD 4.17). South Asian ethnicity (Pakistani, Indian, and Sri Lankan descent) was found in 9 of 29 patients (31%), 4 of 29 (14%) were African Canadian, 3 of 29 (10%) were Caucasian, 2 of 29 (7%) were Asian (Chinese and Filipino descent, respectively), and 1 of 29 (3%) was of Turkish descent. The ethnicity of 10 patients was unknown. The final diagnoses are listed in Table II. CIE and lamellar ichthyosis (LI) accounted for 48% (14 of 29 patients) of cases. Syndromic ichthyoses, including a case of recessive X-linked ichthyosis (RXLI) (syndromic and nonsyndromic presentations) with a contiguous gene syndrome (steroid sulfatase deficiency, near complete absence of the corpus callosum, underdeveloped optic nerves, gastroesophageal reflux, undescended testicles, kyphoscoliosis, short stature, X-linked chondrodysplasia punctata type 1, dyschondrosteosis, and possible Kallmann