Congenital Ichthyosiform Disorders

A Unique Presentation of Possible Ichthyosis Follicularis, Atrichia, and Photophobia (IFAP) Syndrome

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Case Presentation

- The infant was born at 36-1/2 weeks to a 20 year-old African American mother by vaginal delivery.
- Family history: IBD, trisomy 18, possible cardio-facio-cutaneous syndrome, congenital heart defects, and developmental delays.
- Neonatal period: apnea, severe gastroesophageal reflux and feeding difficulties, neurologic abnormalities, skeletal abnormalities, and abnormal eye findings.
Case Presentation

- Physical findings: dysmorphic features including coarse facies, microphthalmia, hypertelorism, small palpebral fissures, thick low-set ears, high sloping forehead, macroglossia, transverse palmar creases, and short broad fingers.
- Cutaneous findings: multiple keratotic spicules on the face and scrotum, thickened doughy skin, prominent follicles and hypotrichosis.
Figure 1. Cutaneous abnormalities and dysmorphic craniofacial features present at birth.
Figure 2. Cutaneous appearance at 3 months of age. *(Courtesy of Moise L. Levy, MD)*
Case Presentation

- Ophthalmologic exam: chorioretinal dystrophy with tapetoretinal degeneration and blepharophimosis.
- Neurological exam: absent septum pellucidum and myotonic seizures.
- Audiologic exam: possible hearing loss with ABR testing.
- Skeletal abnormalities: brachydactyly, platyspondyly, metaphyseal cupping, mildly dysplastic talus and calcaneus, shortened ribs, and hypoplastic sacrum.
Specialized Studies

- Karyotype: 46, XY
- Skin biopsy: follicular ichthyosis
- Urine mucopolysaccharide screen: normal
- Arylsulfatase A and steroid sulfatase activity: normal
- Sterol quantification: mildly increased 8(9)-cholestenol level
- Genetic testing for GJB2 and GJB6 gene mutations: normal
Congenital Ichthyosiform Disorders

• These are rare genetic syndromes characterized by cutaneous scaling from defective cornification and multiple other congenital anomalies.
• Each ichthyosiform disorder is related to a structural defect of specific proteins encoded in the mutant gene.
Ichthyosis follicularis, atrichia, photophobia (IFAP) syndrome

• This is an X-linked syndrome with a classic triad of ichthyosis follicularis, atrichia, and photophobia.
• Skin findings frequently described as ‘thorn-like projections’.
• A patient may also have short stature, mental retardation, seizures, enamel dysplasia, and recurrent respiratory infections.
• Etiology is from mutations in MBTPS2 gene encoding an intramembranous zinc metalloprotease.
• Diagnosis is based on exclusion of other likely diagnoses such as KID syndrome, chondrodysplasia punctata type 2, and multiple sulfatase deficiency.
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References


