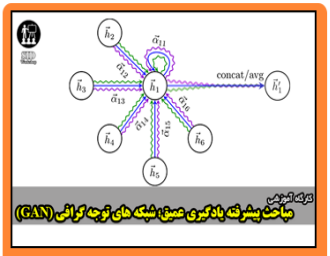
کارگاه های آموزشی مرکز اطلاعات علمی جهاد دانشگاهی

کارگاه آنلاین مکالمه روزمره انگلیسی

کارگاه آنلاین آموزش استفاده از وب آو ساینس



مباحث پیشرفته یادگیری عمیق؛
شبکه های توجه گرافی
(Graph Attention Networks)

Harlequin ichthyosis (or harlequin fetus)

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Dear Editor

The patient was a female neonate born prematurely on 36 weeks of gestation by normal vaginal delivery. During pregnancy, mother had no history of fever, drug use or x-ray exposure. The mother was gravida 2 and had no history of abortion. Her first baby was well.

On physical examination, the skin was markedly thickened, hard and hyperkeratotic with deep crevices running transversely and vertically. She had ectropion and the globe was abnormal. The ears and nose were underdeveloped, flattened and distorted and the lips were everted and gaping (fish-mouth). Extreme inelasticity of the skin with flexion deformity of all joints was noted. The hand and foot were hard and waxy in appearance, with poorly developed distal digits (Figure 1,2). She had respiratory distress, was ill and sucked poorly.

Harlequin ichthyosis (or harlequin fetus) is a rare but very severe disorder of keratinization. Inheritance is likely autosomal recessive, but new autosomal dominant mutations are possible [1]. The genetic abnormality in harlequin ichthyosis has been identified as a mutation in the lipid-transporter gene *ABCA12* on chromosome 2. In harlequin ichthyosis, the *ABCA12* -mediated transfer of lipid to lamellar granules is absent [2].

The skin of the affected infants is markedly thickened, hard (armor-like), and hyperkeratotic, with deep crevices running transversely and vertically (Figure 1). The fissures are most prominent over areas of flexion. Rigidity of the skin around the eyes results in marked ectropion, although the globe is usually normal; however, our patient had ectropion with an abnormal globe (Figure 2). The ears and nose are underdeveloped, flattened, and distorted, and the lips are everted and gaping, producing a "fish-mouth" deformity. The nails and hair are hypoplastic or absent. Extreme inelasticity of the skin is associated with flexion deformity of all joints. The hands and feet are ischemic, hard, and waxy in appearance, with poorly developed distal digits [3]. Affected neonates have respiratory difficulty, suck poorly, and are subject to severe cutaneous infection [4].

Common histopathological abnormalities include hyperkeratosis, accumulation of lipid droplets within corneocytes, and absence of normal lamellar granules [1,5].

Prenatal diagnosis has been accomplished by fetoscopy, fetal skin biopsy, and microscopic examination of cells from amniotic fluid taken at the 17th and 21st weeks of gestation [6]. We confirmed diagnosis in this report by ultrasound in the prenatal period and clinical findings during the neonatal period. Suspicion in this case was aroused only with findings of a persistently open mouth

Figure 1. Thickened, hard (armor-like), and hyperkeratotic skin with deep crevices running transversely and vertically.

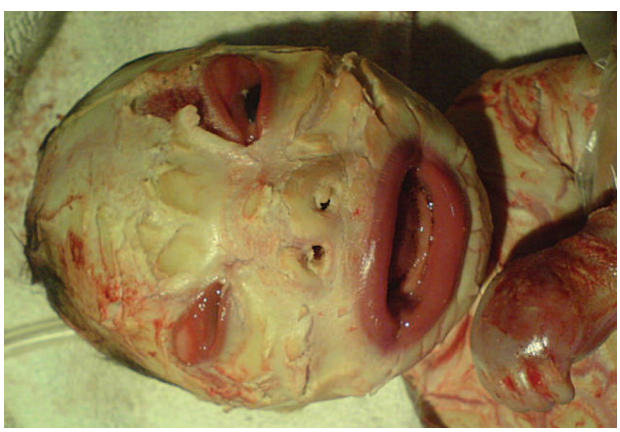

Figure 2. Rigidity of the skin around the eyes with marked ectropion

and fixed flexion deformities on two dimensional sonography whereas three dimensional sonography may disclose facial features more commonly associated with the condition, echogenic amniotic fluid, short foot length and fixed flexion deformity.

Initial treatment includes high fluid intake to avoid dehydration from transepidermal water loss and use of a humidified heated incubator, emulsifying ointments, careful attention to hygiene, and oral retinoids [4]. Unfortunately, our patient died because of sepsis.

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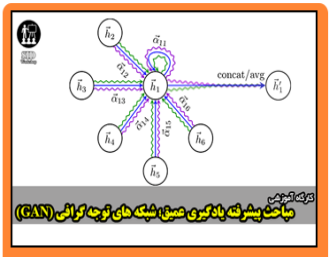

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