# Short Communication

# A case of monilethrix

Sir, monilethrix is a rare hair shaft disorder with structural defect resulting in increased fragility. It is usually transmitted in an autosomal dominant pattern. Affected individuals usually have normal appearing hair at birth, which is replaced by short, fragile and brittle hair. Perifollicular erythema and follicular hyperkeratosis are commonly observed.[1]

A three-year-old boy presented with scanty, short, brittle scalp hair. His father gave history that the child's hair was short and scanty since infancy. The father of the child complained that his hair were very fragile and didn't grow long. There was no similar history in family and the other siblings were unaffected. Examination showed diffuse hair loss with short and brittle hairs over scalp. Follicular keratotic papules were seen over the entire scalp but more prominent on occipital region (**Figures 1** and **2**). The development milestones of the child were appropriate for age. Rest of the systemic examination was within normal limits.

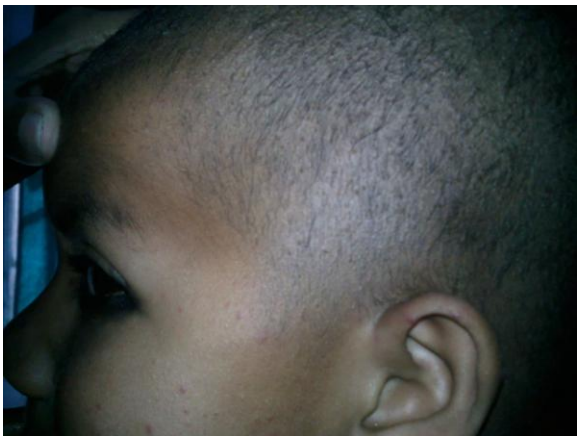

**Figure 1** Clinical photograph showing scanty, short scalp hair.

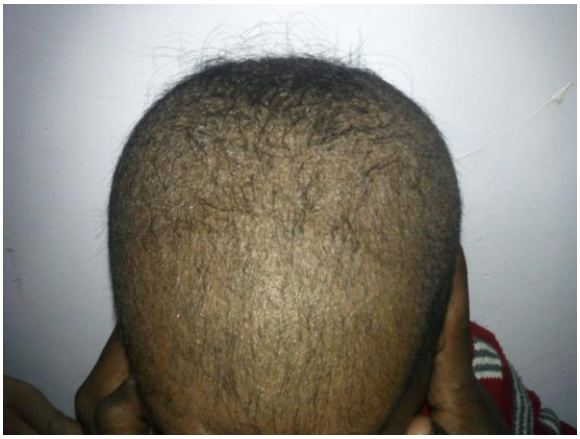

**Figure 2** Clinical photograph showing scanty, short hair and keratotic papules on the scalp.

Monilethrix was first described by Walter Smith in 1879 who called it 'a rare nodose condition of the hair' for which Radcliffe Crocker subsequently suggested the term monilethrix.[2] The word is derived from monile (Latin), which means necklace, and thrix (Greek), which means hair indicating the resemblance of the hair to a string of beads or a necklace. Hairs usually break at internodal area where cortex is defective and medulla is absent. Autosomal dominant transmission has been demonstrated in numerous large pedigrees.[3] Monilethrix shows considerable variation in age of onset, severity, and course. Light microscopy of hair shows a beaded appearance because of alternate zones of spindle like thickening and thinning placed about 0.7□1 mm apart. Some cases improve spontaneously after puberty though most persist throughout life. Avoidance of chemical and mechanical trauma may be helpful.[1] Oral etretinate has been used with limited success.[4] This case shows the variable expression as none of the family members were affected.

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# Case report of harlequin ichthyosis who had similar history in two earlier siblings

Harlequin ichthyosis is the most severe form of congenital ichthyosis. It is a rare autosomal recessive disorder. The skin development is altered *in utero* due to defective lipid metabolism in lamellar granules of the keratinocytes. Lack of desquamation and massive accumulation of scales lead to the characteristic morphological features.[1] We report a case of harlequin ichthyosis that had a history of similar affection in two earlier siblings and two siblings are absolutely normal.

**A** 35-year-old, G5P4+0 (previous 2 sections) presented through emergency department at 35 weeks with complaints of labor pains and underwent emergency caesarean section and delivered male baby who cried immediately after birth. His anthropometric measurements were birth weight - 2.0 kg, crown-heel length 40 cm, head / chest circumference - 32/29 cm. On examination the skin of the baby was leathery white with large shiny plaques separated by deep erythematous fissures along with flexion contractures of arms, legs and digits. He also had severe ectropion, rudimentary pinna, sparse hairline at the back of the scalp, nasal hypoplasia and eclabium with a fixed, open mouth. Fingers and toes were hypoplastic and tips were gangrenous. All natural orifices were patent. External genitalia were male with bilateral undescended testes. Umbilical cord was fragile. The vital parameters were within normal limits except mild tachypnea (68/min). With these findings clinically diagnosis of harlequin ichthyosis was made. On interrogation, mother gave past history of similar affection in the first and the forth issues who died within few hours after birth while the second and third are absolutely normal. Baby was advised admission in NNU but parents refused admission and taken him to home where he expired on the way.

Harlequin babies are born with dense plaque-like scales forming a massive horny shell around their body. These scales or plaques measure up to 4-5 cm on a side and are usually diamond or triangular shaped. 'Splits' between these scales reveal erythematous moist fissures, whose bright red color is in sharp contrast to the lighter colored scales. This shell impedes movement, and the fissures cause the protective skin barrier to be compromised, thus leaving the fetus susceptible to dehydration and infection. Because of the scaling of the skin, the limbs are often deformed to the extent that movement is restricted, and may be undersized and incompletely formed. The scales may sometimes also constrict blood flow to the limbs resulting in swelling or in severe cases, gangrene.[2]

Abnormalities of skin is linked to the abnormal structure and function of lamellar granules originating from the Golgi apparatus of keratin-producing cells in the horny outer layer of the epidermis, whose function is to secrete lipids that maintain the skin barrier. Harlequin fetuses either do not have these granules, or have defective ones, which results in massive loss of water between the layers of skin. The lack of a class of enzymes known as hydrolases further prevents shedding of skin cells, thus causing overgrowth of the horny layer of skin.[3]

The inheritance is thought to be autosomal recessive. It has recently been shown that the vast majority of affected individuals are homozygous for mutations in the ABCA12 gene, which cause a deficiency of the epidermal lipid transporter and result in hyperkeratosis and abnormal barrier function.[4,5] Ultrasonography can make a prenatal diagnosis and should be used particularly in those families with history of a Harlequin baby.[6]

Our patient had the typical phenotypic features of a harlequin fetus. Though USG in expert hands during the antenatal period is diagnostic of the condition the family but our patient belong to rural background and her USG were done but not up till marked. Because of the shock and previous bad experience family was not ready to get baby admitted in NNU and taken him to home where he died on the way.

In general, harlequin fetuses do not survive for long. death is often due to dehydration, systemic infection or impaired respiration.

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# Keratolytic acanthoma of the penile shaft

Epidermolytic acanthoma (EA), histologically characterized by the presence of "epidermolytic hyperkeratosis" (EH), is a rare acquired, benign cutaneous disorder. Usually it occurs as a solitary papillomatous lesion in early adulthood. The exact etiology is not known. Few cases have been reported in the literature. we report a case of EA with unusual presentation over the male external genitalia.

*Case report* A 40-year-old male was referred from Community Health Centre to our Dermatology Department with history of

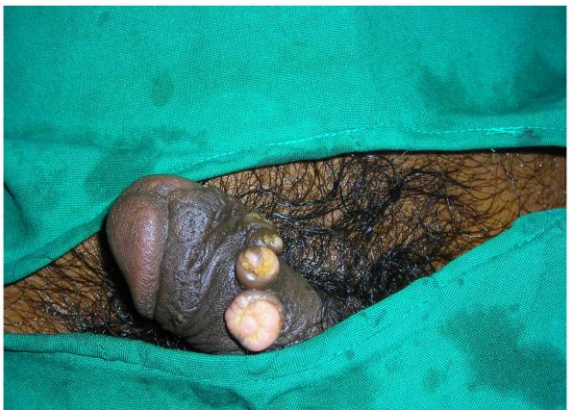

**Figure 1** Discrete, contiguous, skin-coloured, verrucous nodules of the penile shaft.

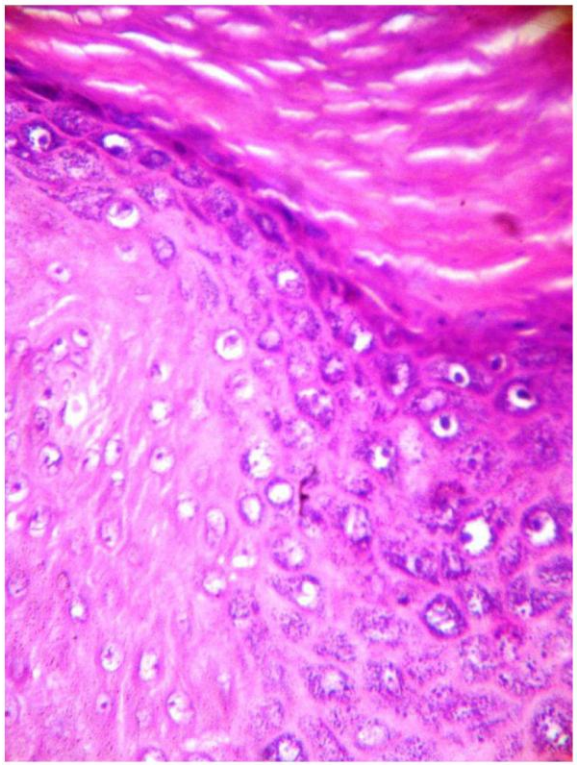

**Figure 2** Perinuclear clear cell change in the upper stratum spinosum and granular layer with keratohyalin granules (H & E x 400).

asymptomatic growths over the external genitalia for evaluation and to rule out the possibility of sexually transmitted disease. There was no history of extramarital sexual exposure. His family history was noncontributary.

Cutaneous examination revealed four discrete, contiguous skin-coloured verrucous nodules of various sizes over the ventral aspect of the penile shaft (**Figure 1**). The rest of the skin and systemic examination were normal.

The differential diagnosis considered were condyloma acuminata and verrucae of the penis. Routine hematological and biochemical tests were normal. Blood VDRL, TPHA, ELISA for HIV 1 and 2, HBsAg and anti-HCV antibodies were negative. Patient was referred to the surgeon for excision and the biopsy material was sent for histopathological examination.

Apart from compact hyperkeratosis and focal acanthosis, light microscopic examination of the skin biopsy specimen revealed the characteristic perinuclear clear cell changes in the upper stratum spinosum and granular layer (**Figure 2**). Based on the clinicopathological correlation final diagnosis of epidermolytic acanthosis was arrived.

Epidermolytic acanthoma (EA) is a rare, acquired benign tumor of the epidermis, mainly characterized histopathologically by a prominent epidermolytic degeneration of the keratinocytes (Epidermolytic Hyperkeratosis). It was first described in 1970 by Shapiro and Baraf *et al.*[1] It usually appears at or after middle age, and has been reported in various locations including the face, abdomen,[2] trunk, extremities[2] and external genitalia.[3,4]

EA usually present as asymptomatic tumor less than 1 cm in diameter with verrucous surface. Lesion can present in either an isolated solitary, localized or disseminated form with more predilection for the genitoscrotal area.

Regarding pathogenesis of EA, many hypothesis including mutation in K-1 and K-10 gene has

been postulated,[5] but none validated. Many attempts failed to demonstrate human papilloma virus DNA from the biopsy material of the EA.[6]

In majority of reports, EA of the external genitalia are solitary and rarely seen. We report the first case, to our knowledge, of a multiple EA over the penile shaft. EA confined to the external genitalia is an under diagnosed entity and it should be considered as a differential diagnosis in asymptomatic, warty, discrete nodular lesions over the external genitalia.

Various treatment options have been proposed including excision, cryotherapy, and electrocauterization. Our patient effectively under went local excision and the postoperative period was uneventful.

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