Eruptive Vellus Hair Cysts in Association with Hypomelanosis of Ito with Turner's Syndrome

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A 24-year Korean woman presented with bizarre pigmentary skin changes. Eruptive vellus hair cysts(EVHC) were observed in conjunction with Hypomelanosis of Ito(HI). Also, the karyotype of 45XO with typical clinical symptoms of Turner's syndrome was detected in this patient, which has been rarely reported in HI as a chromosomal defect. EVHC may manifest as a rare skin manifestation of HI.

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Key Words : Hypomelanosis of Ito, 45XO, Eruptive vellus hair cysts

Hypomelanosis of Ito (HI) is characterized by hypopigmented skin areas of abnormal patterns along the Blaschko line with neurologic and musculoskeletal abnormalities. HI is reported to be associated with many kinds of chromosomal abnormalities[1]. Commonly these include mosaic trisomy 18, diploidy/triploidy, mosaicism for sex chromosome aneuploidy, and tetrasomy 12q, and the majority were found to be mosaic for aneuploidy or unbalanced translocations. All of these indicate that HI is a multisystem disorder frequently associated with chromosomal defects[2]. In the case reported here, a novel skin manifestation of eruptive vellus hair cysts (EVHC) was observed in conjunction with HI. Also, the karyotype of 45 XO with typical clinical symptoms of Turner's syndrome was detected in this patient, which has been rarely reported in HI as a chromosomal defect. EVHC may manifest as a rare skin manifestation of HI. However, the possibility of casual association of EVHC in HI should be ruled out.

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CASE REPORT

A 24-year-old Korean woman presented with bizarre pigmentary changes. Asymptomatic sharply demarcated hypopigmented areas were on the inner and flexor aspects of both legs and arms, which were arranged in parallel streaks (Fig. 1). There were similar lesions on the trunk but they were less linear, shorter scattered patches. They had started to develop on her legs when she was 3 years old, and gradually progressed to the arms and trunk. The DOPA stain of the specimen from a hypopigmented skin lesion showed decreased melanocytes in the basal layer. She also had multiple yellowish, 1-2mm follicular papules on both upper extremities(Fig. 2A). A biopsy specimen from a yellowish papule on her right forearm showed that it was a cyst in the dermis filled with keratin and vellus hairs, compatible with eruptive vellus hair cysts(Fig. 2B).

An examination revealed webbed neck, redundant skin, low posterior hairline, low-set ears, wide-spaced and underdeveloped breasts, and lymphedema on the legs. She was of short stature (144cm height) for a Korean woman. She had an increased number of nevi, especially on her face. Ocular examination disclosed mild strabismus,

myopic astigmatism and iridal nevus on the right eye. She had mildly abnormal auricles. X-ray study revealed kyphoscoliosis of the thoracic spine and generalized osteopenic changes. She also had Genu Valgum. Repeated chromosomal analyses upon peripheral lymphocytes showed only 45 XO, without known mosaicism. In her past history, motor development was retarded, sitting at 12 months and walking and talking at three years of age. There was no family history of psychomotor retardation. Her four brothers were normal. She was diagnosed as Turner's syndrome by the characteristic clinical symptoms and chromosomal defect. She had been receiving estrogen therapy in the department of Gynecology due to amenorrhea and lack of secondary sex characteristics.

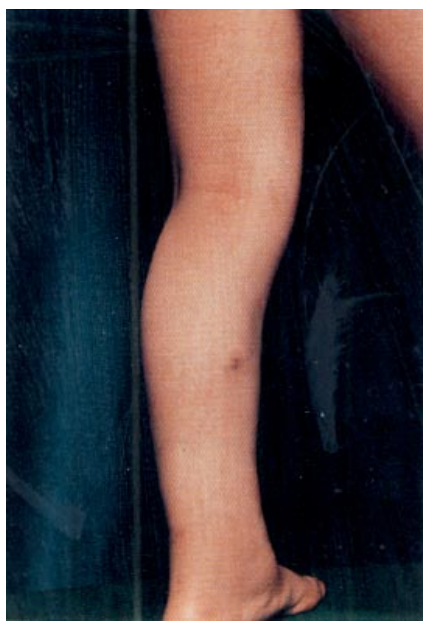

Fig. 1. Sharply demarcated hypopigmented areas as streaks or linear patterns on the leg.

DISCUSSION

HI is a heterogeneous neurocutaneous disorder that has been associated with systemic manifestations including neurological, ophthalmologic, dental, and musculoskeletal abnormalities in individuals with hypopigmentation or depigmentation distributed along the Blaschko line[1]. As a cutaneous manifestation, incontinentia pigmenti achromians is found as the negative image of incontinentia pigmenti, which appears as bizarre shaped hypopigmented lesions, such as streaks, whorls, eddies, and guttate patterns. Ruiz-Maldonado et al. [3] proposed the diagnostic criteria for HI based upon the presence of hypopigmented whorls and streaks in more than two dermatomes with one major (central nervous system or musculoskeletal anomaly) and two minor (other congenital anomalies) criteria for definite diagnosis. Our case fulfills the diagnostic criteria by the presence of marginal mental retardation, kyphoscoliosis and chromosomal abnormality as well as typical skin lesions of streak patterned hypopigmented patches.

The majority of HI cases are known to have chromosomal abnormalities including mosaic and nonmosaic structural abnormalities, aneuploidy,

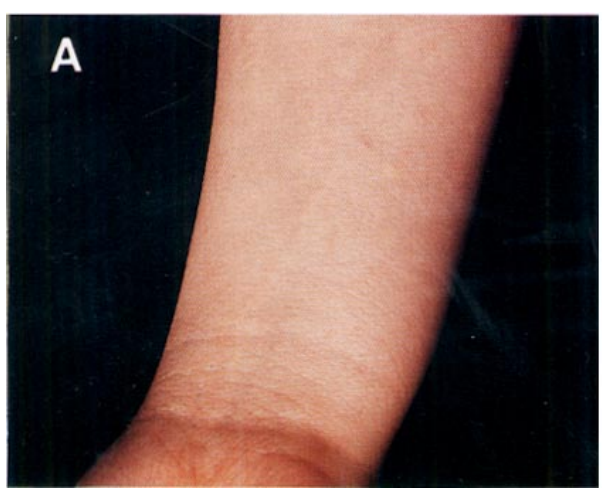

Fig. 2A. Multiple yellowish, follicular papules on the forearm.

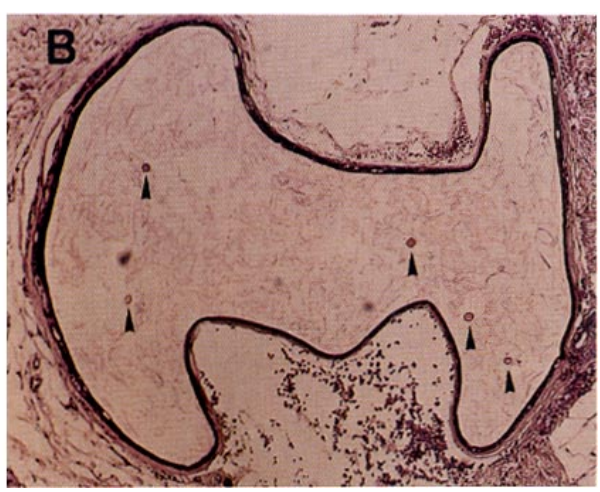

Fig. 2B. A cyst in the dermis filled with keratin and vellus hairs(arrow heads).

polyploidy, and chimerism [4]. Therefore, HI is regarded as a cutaneous symptom of the underlying chromosomal defects. However, some reports on HI have stated that there was no evidence of chromosomal abnormalities. However, in these cases, there is a possibility that multiple tissues were not karyotyped and that genetic aberrations were not recognized. In a review of 70 published cases of HI, 42 individuals (60%) had chromosomal abnormalities in blood and skin samples[2]. Most of the HI patients were mosaic for aneuploidy or unbalanced translocations. The more common alterations included mosaic trisomy (18), diploidy/triploidy, mosaicism for sex chromosome aneuploidy and tetrasomy 12p[2]. X-chromosome abnormalities have been found in 60% of the cases of HI, but the deletion of the X-chromosome is rarely reported [5]. In this case, we found only the abnormal X-chromosome deletion of karyotype 45 XO in 3 samplings of lymphocytes (15 microscopic fields for karyotyping).

Interestingly, she had eruptive vellus hair cysts on her upper extremities, which had not been reported previously in association with HI. Eruptive vellus hair cysts are regarded as a developmental abnormality of vellus hair follicles, manifesting with yellowish to reddish brown, small papules on the chest and proximal extremities. They usually occur sporadically, but are sometimes inherited as an autosomal dominant trait[6,7]. Eruptive vellus hair cysts share a clinical resemblance with steatocystoma multiplex, which is also inherited in an autosomal dominant pattern, by similarity in age of onset, predilection of sites, clinical appearance, and mode of inheritance. Therefore, the term multiple pilosebaceous cysts' was proposed for both conditions because they were regarded as a part of the same disease spectrum [8]. We could not find a family history of vellus hair cysts in this case, however, there is a possibility that the gene for the cystic lesions is aberrantly expressed with the HI gene defects. Further studies are warranted to evaluate genetic relationship between HI and EVHC/steatocystoma multiplex in future.

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