Incontinentia Pigmenti Achromians

INCONTINENTIA PIGMENTI ACHROMIANS

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SUMMARY

Incontinentia pigmenti achromians is a neurocutaneous disorder with swirled hypopigmentation following lines of Blaschko. It has been described as a distinct multisystem defect. We report a 16-year-old boy with incontinentia pigmenti achromians. There were bandlike and swirls of hypopigmentation on his trunk and extremities which had been recognized when he was 1-year-old. There was also malformed auricle of his right ear. A biopsy specimen showed decreased numbers of melanocytes. EEG showed irregular waves in the parietooccipital area. Magnetic resonance showed cystic dilatations in the right frontal and parietal periventricular white matter. However there was not any abnormal neurological symptoms.

Key words: hypopigmentation, neurocutaneous

ÖZET

Incontinentia pigmenti achromians Blaschko çizgilerini takip eden girdapsı hipopigmentasyon ile karakterize neurokutan bir sayrılıktır. Farklı bir multisistem defekti şeklinde tanımlanmıştır. Incontinentia pigmenti achromians'lı 16 yaşında bir erkek olgu sunulmaktadır. Olgunun gövde ve ekstremitelerinde, 1 yaşında başladığı tanımlanan, band tarzında ve girdaplar oluşturan hipopigmentasyon belirlenmiştir. Aynı zamanda sağ kulakta malformasyon saptanmıştır. Deri biyopsisinde melanositlerin sayısında azalma gözlenmiştir. EEG'de parietooksipital alanda irregüler dalgalar saptanmıştır. Magnetik rezonans görüntülemede sağ frontal ve parietal periventrikuler beyaz maddede kistik dilatasyonlar saptanmıştır. Bununla birlikte anormal nörolojik simptom belirlenmemiştir.

Anahtar sözcükler: Hipopigmentasyon, neurokutan

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Incontinentia pigmenti achromians (IPA) is a rare neurocutaneous syndrome characterized by whorls, streaks and/or patches of hypopigmentation typically distributed along lines of Blaschko (1-3). Ito described, in 1951, a variant of systematized depigmented nevus and proposed the term "incontinentia pigmenti achromians", because the configuration of the hypopigmentation was similar to that of the hyperpigmentation seen in incontinentia pigmenti Bloch-Sulzberger (4-6). However, in the dermis, no incontinent melanin is seen (7). Jelinek et al, indicated that the disease should be called as hypomelanosis of Ito (8). Although Ito's original report described a pure cutaneous disease, subsequent reports included patients with abnormal features of mainly central nervous, musculoskeletal, ocular and other systems (9). We report a case of IPA with an abnormal head magnetic resonance (MR) who is neurologically normal.

CASE REPORT

A 16-year-old boy referred to our hospital with the complaint of hypopigmentation on his trunk and extremities which began from his legs when he was 1-year-old. There were not any signs of inflammation,

vesiculous and verrucous lesions at the beginning. The lesions became more pronounced as he grew older. His medical history and psychomotor development was normal. He was the product of uncomplicated pregnancy. There was not any family history of similar disturbances and there was not any consanguinity of his parents.

Dermatologic examination revealed bilateral hypopigmentation mainly arranged in streaks on his extremities (Figure 1) with swirls of hypopigmentation on his trunk (Figure 2) along Blaschko lines. Involvement of the right side was more prominent than the left. The margins are blurred. Hair, teeth and nails were found normal. Malformation of the right auricle (Figure 3) was detected. Systemic examination including neurologic, musculoskeletal, cardiovascular systems and eyes were normal.

Laboratory examination of his routine urinalysis, hematological and biochemical analysis were normal. Also, hormone investigations, IgE and tumor markers were normal. Blood standard chromosomal analysis did not show any anomaly. Ultrasonography, chest X-ray, EKG, vertabrae X-ray investigations were normal. EEG showed sharp-wave activities on the parietooccipital area. Head MR investigation was performed on cranial T2 weighted images, there were high signal foci in the parietal periventricular white matter. Those round, linear foci were hypointense similar with cerebrospinal fluid signal on T1 weighted images. These findings confirmed the diagnosis of dilated perivascular spaces in the parietal white matter.

The histopathological investigation of the biopsy material taken from the hypopigmented area revealed decreased number of melanocytes and melanin in only focal areas of the basal layer.

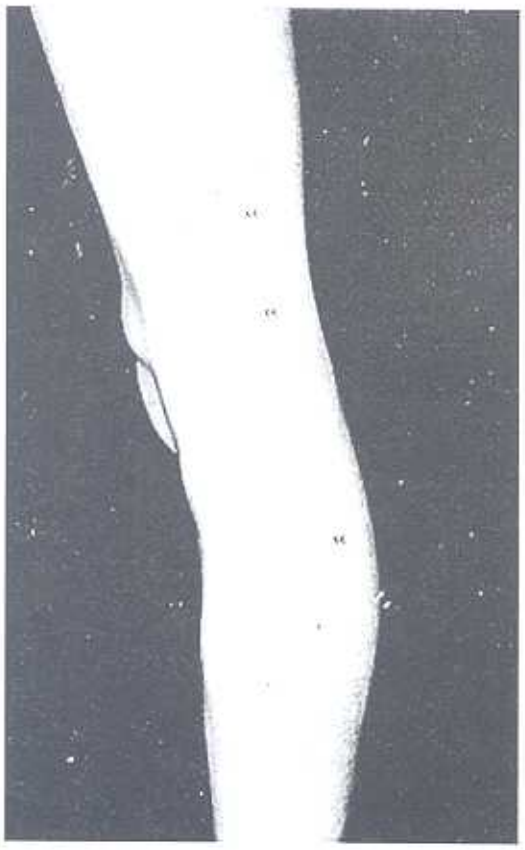

Figure 1. Hypopigmentation arranged in streaks

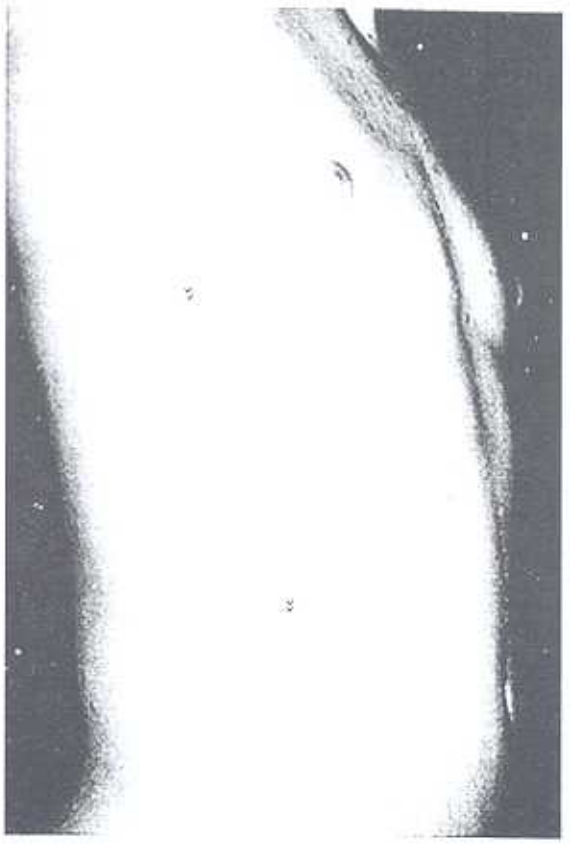

Figure 2. Swirls of hypopigmentation

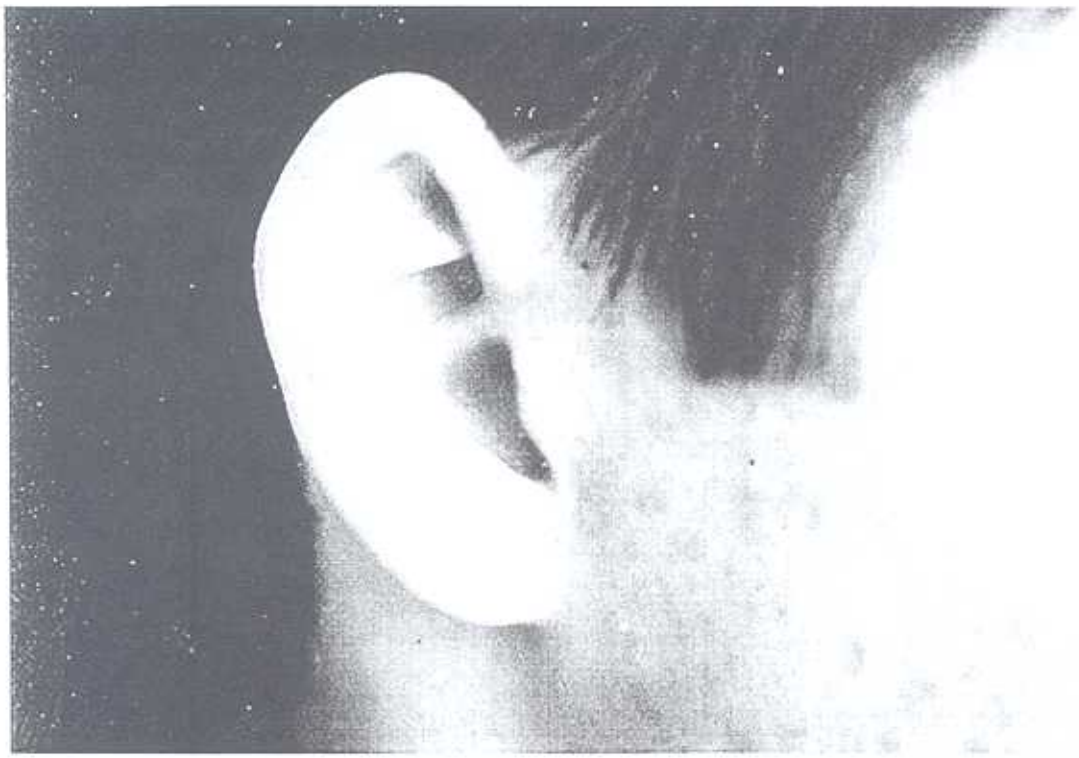

Figure 3. Malformation of the right auricle

DISCUSSION

IPA may be associated with various abnormal systemic features, predominantly neurologic. So, it is referred as a neurocutaneous syndrome (4,7,9,10). Clinically the lesions were described as the negative image of incontinentia pigmenti (10,11). Whirls and streaks of hypopigmentation often in parallel array and along Blaschko lines were detected in unilateral or bilateral distribution (5,7,10). Particularly the trunk and extremities are involved. The margins of the lesions are serrated and blurred or sharply outlined (10). In our patient, whorls and streaks of lesions are distributed bilaterally and the margins are blurred.

The lesions are usually present at birth, however may become prominent usually in the first year of life, like in our patient (4,7,11). But, appearence after 5 years of age was also observed. They may progress or regress minimally but are most often stable (11). It is seen more common in females and only few males are present (1,10).

Histologically decreased number of melanocytes with defective function is detected. Melanogenesis is reduced (11). A decrease in the melanin content of the basal layer was described (5,7).

Although the majority of IPA cases are sporadic, there have been only few reported familial cases (5,7,10,12). Chromosomal mosaicism has been shown as a common finding in IPA (5,7,10-13). It has been stated that in IPA mosaicism of different types of aneuploidy is frequently present and most commonly involves the X chromosome (1). In addition to mosaicism, chromosomal translocations have been described in several patients (7).

Ruiz-Maldonado et al. proposed diagnostic criteria for IPA. According to them in addition to whorls and streaks in more than two dermatomes, one major (CNS and musculoskeletal anomalies) and two minor (other congenital anomalies) criteria was required. However the original cases of Ito would not have met these criteria (10). There did not appear any abnor-

mality except for hypopigmentation in Ito's first case (4). Also, in our case in addition to classic cutaneous findings, only head MR and ear abnormalities are detected.

IPA must be distinguished mainly from incontinentia pigmenti and nevus depigmentosus. IPA lacks early cutaneous vesicular or verrucous changes like seen in incontinentia pigmenti and depigmentation occurs much later in incontinentia pigmenti, if at all. Also, basal layer degeneration, pigment incontinence and dermal inflammation of incontinentia pigmenti is not observed in IPA (1,10). Nevus depigmentosus is usually congenital, hypomelanosis is permanent and usually stable, whereas IPA may appear years after birth and hypomelanosis tends to fade (4,10). In IPA an initial increase in the extent of involvement can be followed by a gradual repigmentation (7). Moreover, nevus depigmentosus is localized and unilateral, but IPA is systematized and often a marker for underlying congenital defects (10). In contrast to IPA, neurologic and ectodermal changes are very rare in patients with nevus depigmentosus (4,10).

There is a high incidence of systemic abnormalities in IPA (11). Abnormalities of central nervous system, eyes, musculoskeletal system (including ear malformations), hair, teeth, or internal organs was found in more than 75%, mental retardation in over 60% and seizures in over 50% (8,10). In fewer than 10%, the cutaneous findings are the only apparent abnormality (10). It was observed that male patients were generally not more severely affected by congenital anomalies than female patients, like seen in our patient (4).

Central nervous system (CNS) involvement in IPA has been reported in 50-60% of patients. These abnormalities included mainly mental retardation, seizures, hypotonia, nystagmus, ataxia and language disabilities (3,14). Gray matter heterotopia, diffuse and focal cerebral or cerebellar atrophy, hemispheric asymmetry, porencephaly, periventricular cystic lesions, Moya-Moya syndrome, agenesis of the corpus callosum and defection of white matter involvement especially in cases with neurologic manifestations have been reported with CT or MR (14). Also, various neurologic abnormalities have been detected in all of the patients reported to have abnormal CNS imaging studies. However, Fryburg et al reported an IPA case with an abnormal head MRI (enlargement of he third and lateral ventricles, several small cyst-like lesions and other white matter abnormalities) who was neurologically normal and they stated that abnormal brain MRI may not predict a poor or abnormal neurologic outcome in IPA (3).

In our case, there were only dilated perivascular spaces on MR and neurologic abnormalities were not detected. Routine head MR investigation should be made in all IPA patients, including neurologically symptom free ones.

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