Rare disease

Becker's nevus syndrome with quadriparesis

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Summary

A 12-year-old girl presented to Manipal Teaching Hospital with quadriparesis of 8 months' duration. Examination revealed a hyperpigmented patch over the chest wall with overlying hypertrichosis, musculoskeletal anomalies, upper limb asymmetry and ipsilateral breast hypoplasia. MRI scan revealed cranio-vertebral junction anomaly and spina bifida occulta at the cervical spine level. Histopathological examination of the skin revealed findings consistent with Becker's nevus. Based on the patient's clinical presentation and investigations, a diagnosis of Becker's nevus syndrome was made. However, she was managed conservatively as surgical intervention was not suitable in her case. The authors review Becker's nevus syndrome and its clinical manifestations below.

BACKGROUND

Becker's nevus syndrome is a rare entity characterised by the presence of Becker's nevus,¹ a well-circumscribed hyperpigmented patch with overlying hypertrichosis which affects a particular area, most commonly the thorax and scapular region, and associated abnormalities which consist of one or more of the following: ipsilateral breast hypoplasia,²⁻³ ipsilateral hypoplasia of the shoulder and limb asymmetry.⁴ Other musculoskeletal defects and vertebral defects such as spina bifida occulta can also be present.

After Becker's description of the first two patients with "acquired concurrent melanosis and hypertrichosis", a total of 275 cases of Becker's nevus have been documented, of which only 60 have been reported in the literature with associated underlying abnormalities, with a female:male ratio of 1.5:1.0.⁵

We present a case report of congenital Becker's nevus syndrome with typical Becker's nevus and underlying cranio-vertebral junction anomaly, musculoskeletal abnormality, ipsilateral hypoplasia of the breast and limb asymmetry with quadriparesis.

CASE PRESENTATION

A 12-year-old girl presented with a hyperpigmented patch and hypertrichosis over the upper limbs and chest wall, which was more pronounced over the right arm. A limited range of movement of the neck had been noticed by her mother since birth and she had a 6-month history of weakness of both the upper and lower limbs. The weakness followed an injury sustained after a trivial fall 8 months previously when she had fractured her right humerus and cervical spine. Her medical history was unremarkable. There was no significant family history of skin or systemic disorders and there was no mental retardation or developmental delay.

Her physical examination revealed a well-defined, hyperpigmented patch over the entire right forearm and arm extending along the chest wall and entire scapular region to the left arm and forearm. It was segmental and non-pruritic. Hypertrichosis was seen covering most of the right forearm and arm and right scapular region (figure 1A,B). Chest examination revealed pectus excavatum and hypoplasia of the right breast bud which measured 2 cm while the left

Figure 1 (A,B) Well-defined hyperpigmented patch over the scapular region extending over the anterior chest wall with overlying hypertrichosis. (B) Pectus excavatum and limb asymmetry.

Figure 2 MRI scan of the spine showing deformed posterior elements of the upper cervical vertebrae. Compression of the cord by the odontoid process of C2 and spina bifida occulta at second cervical spine level can be seen.

breast bud measured 3 cm. On examination of her limbs, her right arm was observed hanging by her side and rotated medially; the forearm was extended and pronated due to Erb's palsy. The right arm and shoulder girdle, however, revealed increased bulk compared to the left side. Her higher mental functions were intact, pupillary reactions were normal, and the cranial nerves and sensory system were intact. Motor examination revealed hypotonia in the right upper limb. Power in the right upper limb and both lower limbs was 1/5, while in the left upper limb it was 3/5. Bilateral plantar reflex showed an extensor response. Ankle clonus was present in both lower limbs. Other systemic examinations were normal.

INVESTIGATIONS

Serum follicle-stimulating hormone, thyroid-stimulating hormone, testosterone, oestrogen, progesterone and prolactin were found to be within normal limits.

Histopathological examination of the skin revealed epidermis lined by keratinised stratified squamous epithelium with slight acanthosis. There was hyperpigmentation of the basal layer with increased melanocytes. Melanophages were seen in the upper dermis. Pilar structures were increased in number. The dermis showed irregularly arranged thick bundles of smooth muscle. Findings were consistent with Becker's nevus syndrome.

CT scan of the brain was normal. Chest x-ray showed bony wall deformity, pectus excavatum, and ribs aligned more vertically than normal and fused to the sternum. Soft tissues were normal.

MRI of the cervical spine revealed scoliosis towards the right side and lordosis of C1–C7 vertebrae. Posterior elements of the upper cervical vertebrae were deformed and the odontoid process of C2 was compressing the spinal cord. Spina bifida occulta was seen at the level of C2 spine (figure 2).

DIFFERENTIAL DIAGNOSIS

The differential diagnosis was McCune Albright's syndrome.

TREATMENT

The management plan in our case was very limited as the patient had presented with the vertebral anomaly after quadriparesis had already set in. However, if the abnormality had been detected prior to the onset of quadriparesis, suitable surgical management would have been transoral odontoidectomy and posterior fusion. However, this surgery could not be performed in our case as it did not guarantee an improvement in prognosis and could have caused deterioration in the patient's condition. We therefore resorted to conservative management which included applying a neck brace for support and regular physiotherapy as well as counselling.

OUTCOME AND FOLLOW-UP

Our patient does not have a good prognosis as she is young and her condition is expected to worsen as her spine grows. Also the manifestation of quadriparesis restricted treatment options. After careful consideration we decided that spinal surgery would only cause deterioration, so we managed her with a brace and regular physiotherapy.

She returns for follow-up each month when we check for any improvement, but unfortunately there has been none so far.

DISCUSSION

Becker's nevus syndrome is a rare phenotype. It is of interest because since one of its entities, Becker's nevus, was defined by Becker in 1949, several cases have been reported with underlying anomalies which suggests that the nevus could be one symptom of an unknown structural anomaly.

Its aetiology remains obscure. However in 1984, Person and Longcope detected an increase in the levels of androgen receptors in the skin of Becker's nevus.⁶ An excess of androgen receptors in the region counteracts the effect of oestrogen. However, this cannot explain the underlying musculoskeletal anomalies seen in the syndrome. Happle and Koopman suggested that Becker's nevus syndrome might represent a paradigmatic pattern of inheritance which means that the disorder manifests when postzygotic mutation occurs.⁷

A timely diagnosis can be made by performing a detailed clinical examination at birth. In suspected cases, the first choice of investigation is histopathological examination of the affected skin. A CT scan and MRI are essential to detect any vertebral defects and soft tissue or musculoskeletal abnormalities. Diagnostic investigation for Becker's nevus should focus on androgen receptor status in skin biopsy. Frequently, the most disabling symptoms are obscure and require a trigger to become apparent, as in our case. The suggested surgical procedure in patients with such vertebral defects with no disability is transoral odontoidectomy, which involves removal of the odontoid process and cervical spine segments and posterior fusion. Another problematic feature of this syndrome is the cosmetic appearance of the nevus, for which ablative resurfacing laser therapy has been introduced.⁸ Patients with isolated Becker's nevus can be reassured about the benign nature of the disease. However, those with underlying anomalies will have to be treated according to the diagnosed defects.

Learning points

- ▶ Becker's nevus syndrome presents with different symptoms except for the one common clinical sign, Becker's nevus.
- ▶ When Becker's nevus is detected, it is very important to rule out any underlying anomalies.
- ▶ The cranio-vertebral anomaly was the main difficulty in our case, which if detected earlier could have been managed, thus avoiding the development of quadriplegia.

Competing interests None.

Patient consent Obtained.

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