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Case Report

Unraveling a mysterious skin eruption in a case of Down syndrome

Sanjana Saikumar, T. P. Thankappan, P. K. Balachandran, Asha Thankappan*, Shabna Shihabudeen, Antony P. Kanichai

Department of Dermatology, Pushpagiri Institute of Medical Sciences and Research Centre, Thiruvalla, Kerala, India

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***Correspondence:**
Dr. Asha Thankappan,
E-mail: ashat80@gmail.com

ABSTRACT

Collagenoma, also known as collagen naevus, is a connective tissue hamartoma which manifests as a single or localized group of fibrous dermal papules and plaques on the skin, and is characterized by increase in dermal collagen with normal or reduced elastin on histopathology. Cases have been reported in Down syndrome, as isolated or localized lesions, and were presumed to be due to altered free radical metabolism by the enzyme superoxide dismutase, its gene being located on chromosome 21. However, eruptive collagenoma in Down syndrome patients are rarely reported. We present a 22-year-old male Down syndrome patient with asymptomatic eruption of skin lesions on trunk and upper extremities since the past 18 months. He was offered surgical and laser therapy, but at his request, we kept him on regular follow-up.

Keywords: Collagenoma, Connective tissue naevus, Down syndrome

INTRODUCTION

Collagenoma, also known as collagen naevus, is a connective tissue hamartoma which manifests as a single or localized group of fibrous dermal papules and plaques on the skin, and is characterized by increase in dermal collagen with normal or reduced elastin on histopathology. They can either be a part of inherited Familial cutaneous collagenoma or or present as shagreen patches of tuberous sclerosus, or even appear as eruptive or isolated lesions.[1] Only few cases of eruptive collagenoma in Down's syndrome have been reported in literature.[2]

CASE REPORT

A 22-year-old male Down syndrome patient came with asymptomatic eruption of skin lesions on trunk and upper extremities since the past 18 months, of insidious onset (Figure 1).

The lesions were slowly progressive in terms of size and number. Prodromal features were absent. There was no antecedent history of trauma or other skin lesions or any obvious inciting factors over the lesioned skin.

He had antenatal history of an emergency caesarean birth at 24 weeks due to maternal bleeding complications, and a history of tongue-tie from birth, but no other congenital deformities. He was diagnosed with Down syndrome, at the age of 5, when he was consulted with a paediatrician for delayed gross motor milestones. Our patient had completed vocational higher secondary schooling with average scholastics. No history of seizures/regression of intellect/congenital heart disease. He had no history of similar lesions in the past, or in his family members. He was not on long-term medications.

On physical examination, he was obese, with a body mass index (BMI) of 35. Down syndrome features such as low-set ears, up-slanting eyes, large tongue, high-arched palate, widely spaced nipples, plantar crease, sandal-gap and wide-stance gait were present.

However, axillary and pubic hair growth appeared normal. His vitals were stable and his system examination revealed no abnormalities, apart from speech impediment due to ankyloglossia.

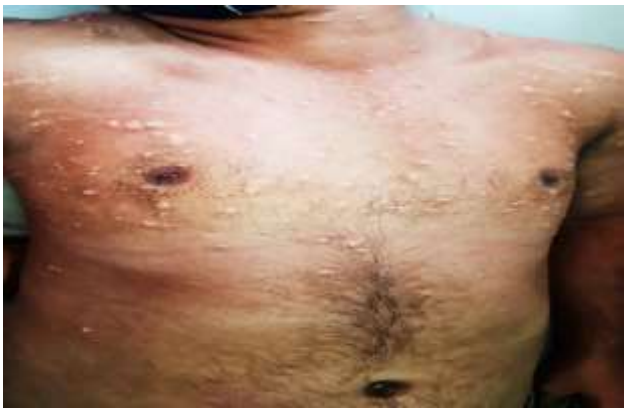

Figure 1: Asymptomatic pale- to skin-coloured soft to firm papules and plaques over trunk and upper limbs.

Dermatological examination revealed multiple well-defined and irregularly shaped pale- to skin-coloured soft-to-firm papules and plaques over the face, front and back of trunk, abdomen and both upper limbs up to midarm (Figure 2). Their sizes varied from 0.2 cm diameter to 6×2 cm.

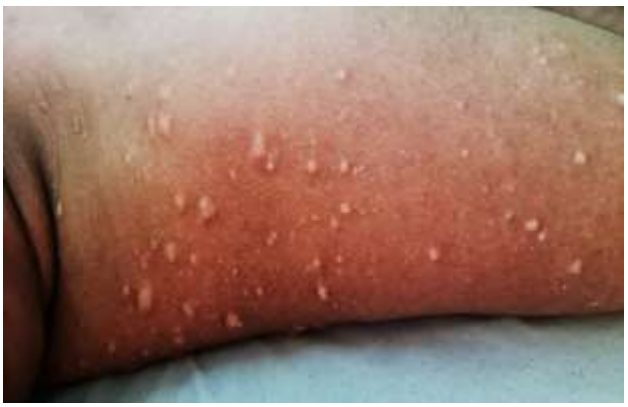

Figure 2: Lesions over the inner aspect of left arm.

A few lesions over the back of trunk showed open comedo (Figure 3). He also had a soft pedunculated erythematous nodule of around 1.5 cm diameter over his left lower lid, and a small non-tender firm erythematous plaque of 0.6 cm diameter and a small hyperpigmented macule <0.2 cm diameter on either sides of his forehead respectively (Figure 4).

A skin biopsy was taken from a lesion on his back, and sent for histopathological examination. On haematoxylin and eosin (H and E) staining, Epidermis showed mild hyperkeratosis, loss of rete ridges and focal basal layer vacuolar degeneration (Figure 5). Dermis appeared homogenously collagenized with focal areas of thickened and degenerated collagen (Figure 6). Mild dermal perivascular and periadnexal lymphocytic infiltrate was present (Figure 5).

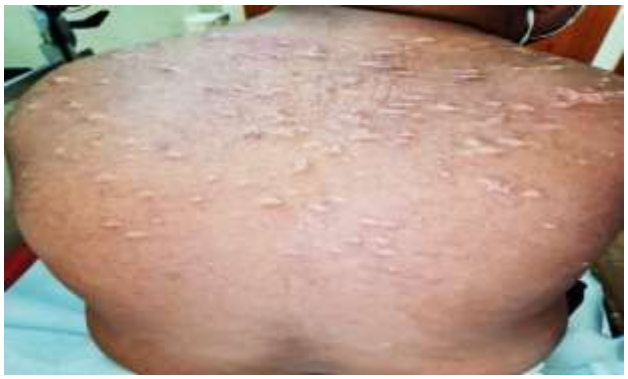

Figure 3: Lesions over the back of trunk, some showing open comedo.

Figure 4: A pedunculated erythematous nodule over left lower lid, an erythematous plaque over left side of forehead, and a hyperpigmented macule over the right side.

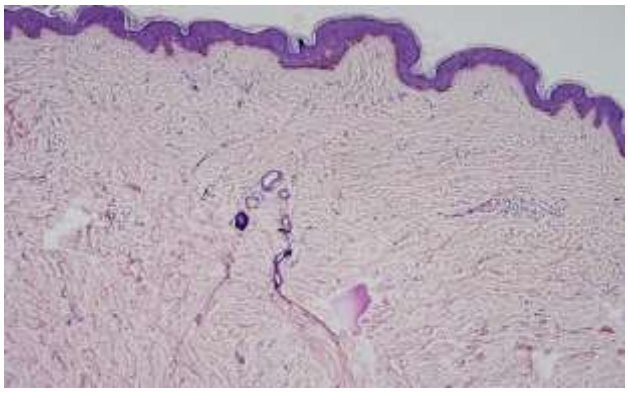

Figure 5: H and E staining, 4X magnification, showing focal basal vacuolar degeneration, irregular rete ridges, homogenously collagenised dermis and perivascular inflammatory infiltrate.

Special staining for elastin was done using Verhoeff van Gieson staining, and it showed sparse to absent staining in dermis (Figure 7), absent normal distribution and

fragmentation of elastin fibres in dermis (Figure 8). These findings led to a histopathological diagnosis of a collagenoma. Further evaluation revealed a normal chest radiograph, echocardiography and MRI brain, with normal TSH levels.

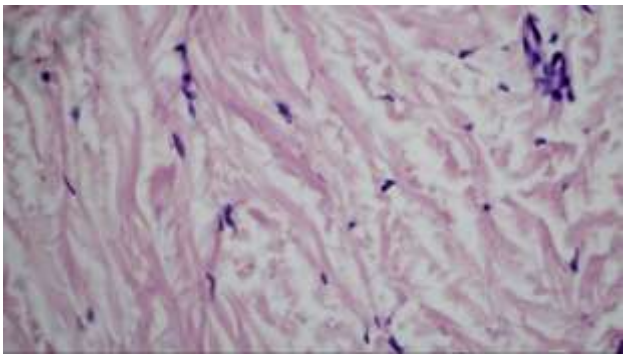

Figure 6: H and E staining, 40X magnification showing focal areas of thickened and degenerated collagen.

Figure 7: Verhoeff van Gieson staining, 4X magnification, showing sparse to absent elastin staining in dermis.

We offered surgical and laser therapy options for removal of lesions but at his request, we decided to keep our patient under regular follow-up instead.

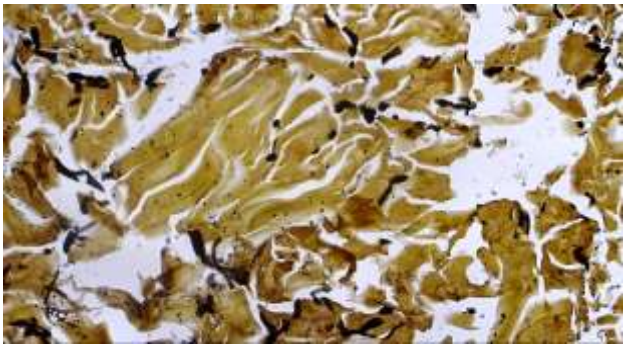

Figure 8: Verhoeff van Gieson staining, 40X magnification, showing absent normal distribution and fragmentation of elastin fibres.

DISCUSSION

Collagenoma or collagen naevus manifests as a single, or localized asymptomatic, firm, flesh-coloured group of fibrous dermal papules, plaques and nodules, of 0.5 to 5.0 cm diameter over the trunk and upper part of the arms. Some lesions may have a peau d'orange appearance.[3]

According to Uitto et al, collagenoma belongs to Lipschutz type of connective tissue naevi.

These are rare hamartomas of the skin, which show increased dermal collagen.[4]

Collagenoma have been categorized clinically into Familial cutaneous collagenoma, zosteriform/linear collagenoma, plantar collagenoma/cerebriform collagenoma and solitary collagenoma.[5] As the name suggests, Familial collagenoma is a syndromic variant, associated with an autosomal dominant mutation of the LEMD3 gene, which codes a nuclear envelope protein component of the inner nuclear membrane (INM).[6] It is involved in controlling the two nuclear signaling pathways, the transforming growth factor beta (TGF-β) pathway and the bone morphogenic protein (BMP) pathway.[7] Therefore, familial collagenoma can sometimes, be associated with Buschke-Olendroff syndrome, where the patient presents with additional signs of osteopoikilosis.[8]

Eruptive collagenoma may be found as sporadic cases or acquired disease. It usually presents as multiple skin lesions appearing after puberty, with numbers ranging from several to more than a 100.[9]

Rapid growing eruptive collagenoma may be associated with multiple endocrine neoplasia (MEN 1) syndrome and pregnancy.[9]

For sporadic cases, only skin biopsy for histopathological examination, and special staining for elastin are necessary. If any of the other variants are suspected, the patient may need to undergo further investigations such as gene analysis for proteus syndrome (plantar/cerebriform collagenoma), Bushke-Olendroff syndrome (familial collagenoma) and screening can be done to rule out tuberous sclerosus in doubtful cases through routine blood work, chest radiography, electrocardiography (ECG), abdominal ultrasonography, fundoscopy, and MRI brain and spinal cord.[7,9]

Cases have been reported in Down syndrome, as isolated or localized lesions, and were presumed to be due to altered free radical metabolism by the enzyme superoxide dismutase, its gene being located on chromosome 21.[3,10] However, cases of eruptive collagenoma are very rare in Down syndrome. Treatment options are limited, and include local excision and laser ablation for isolated lesions.[9]

CONCLUSION

We report a rare case of eruptive collagenoma in an adult male patient with Down syndrome. He was advised surgical and laser therapy, but at his request, we kept him on regular follow-up.

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