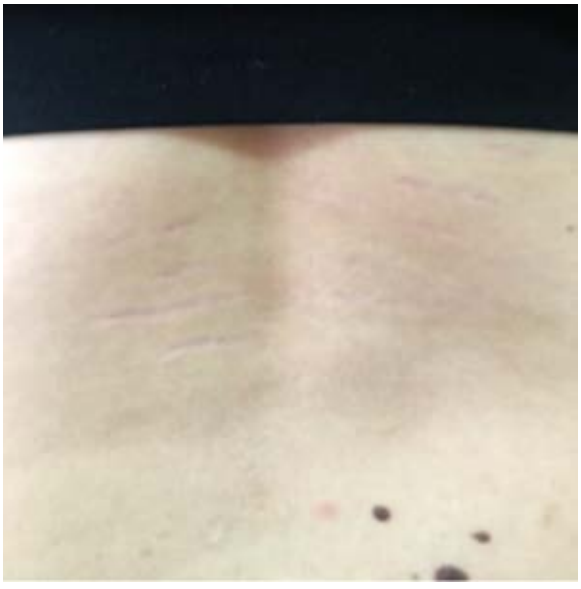
A case of Marfan syndrome presenting with transverse striae of the back

Marfan syndrome is an autosomal dominant inherited genetic disorder resulting from fibrilin gene mutation.[1] The connective tissue associated heart, eye, skeletal, lung, and central nervous system is affected.[1] Differential diagnosis of Marfan syndrome should be considered overgrowth syndromes such as homocystinuria, Sotos syndrome, Beckwith-Wiedeman, neurofibromatosis, Weaver, Proteus and fragile X syndrome.There are rare case reports of Marfan syndrome with transverse striae of the back in the literature.

A girl from non consanguineous family was referred to pediatric endocrine department because of tall stature, joint pain for six months and transverse striae of the back. It was learned from family history that two uncles had tall stature and very long extremities. A 12-years-old girl was born with 4000 gr by caesarean section. On physical examination; height: 182.3 cm (>97p), height SDS: 3.92, weight: 67.1 kg (75-90p), weight SDS: 1.75, body mass index: 20.19, body mass index SDS: 0,54, arm span: 186 cm (>97p), arm span SDS: +4.5(2). On physical examination, the patient with the long-facial appearance had high-archedpalate, long limbs and legs, purple-guinea-colored transverse striae of back (*Figure 1*), arachnodactyly, genu recurvatum, joint laxity, hypermobility, pesplanus, scoliosis and pectus excavatum. The Beighton score: 7/9 for hypermobility and Tanner 4 for puberty were detected. Full blood count, biochemical analysis, thyroid function and pubertal hormone tests were found to be normal in terms of tall stature. Karyotype analysis revealed a normal 46, XX karyotype. Serum levels of IGF-1 and IGFBP-3 ranged from 0 to +1 SDS. Echocardiography revealed mitral valve prolapse. The eye examination was normal in terms of lens subluxation.

FIGURE 1. *Purple-guinea-colored transverse striae of back in our patient*



In this case, Marfan syndrome presenting with transverse striae of the back was reported. Physiological striae atrophicae of adolescence is typically associated with horizontal, linear red streaks (striae rubra) in the lumbar area.[3] Feldman and Smith reported a 15-year-old boy with 12 purple linear markings in the lumbar area.[4] Hormonal (excessive cortisol level) and genetic factors cause linear red streaks. Linear striae was described in a number of conditions such as obesity, pregnancy (striae gravidarum), prolonged use of systemic or topical corticosteroids, excessive use of marijuana, Cushing syndrome, and Marfan syndrome.[5] When our patient was evaluated for Marfan syndrome clinical criteria (the Ghent nosology), Marfan syndrome was diagnosed with systemic score of 10 (a score of ≥ 7 is considered).[1] Marfan syndrome should be considered in cases of tall stature, long-facial appearance, mitral valve prolapse, scoliosis, hypermobility, arachnodactyly, pesplanus, and pectus excavatum. Marfan syndrome should be assessed and followed up with a multidisciplinary approach. Early diagnosis and appropriate treatment will prevent the development of complications.

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References

1. Loeys BL, Dietz HC, Braverman AC, et al. The revised Ghent nosology for the Marfan syndrome. *J Med Genet* 2010;47(7):476-85.
2. Lee TS, Chao T, Tang RB, et al. A longitudinal study of growth patterns in schoolchildren in one Taipei District. II: Sitting height, arm span, body mass index and skinfold thickness. *J Chin Med Assoc* 2005;68(1):16-20.
3. Mishriki YY. Asymptomatic 'streaks' in a healthy young man. Adolescent lumbar striae. *Postgrad Med* 2000;107(4):237-40.
4. Feldman K, Smith WG. Idiopathic striae atrophicae of puberty. *CMAJ* 2007;176(7):929-31.
5. Atwal GS, Manku LK, Griffiths CE, et al. Striae gravidarum in primiparae. *Br J Dermatol* 2006;155(5):965-9.