Popliteal pterygium syndrome: A rare syndrome

Sir,
This report describes the clinical features of an adolescent female who presented to us for popliteal webbing and hence difficulty in walking and operated cleft lip and palate with lower-lip pits. She was diagnosed as popliteal pterygium syndrome (PPS) which is a rare autosomal dominant congenital disorder described by Trelat in 1869 that includes orofacial, cutaneous, musculoskeletal and genital anomalies.\(^1,2\) This syndrome has some features in common with Van der Woude syndrome, also inherited as an autosomal dominant condition.\(^3\) The minimum diagnostic criteria for PPS are any three of the following: cleft lip/palate, popliteal pterygium, paramedian lower-lip pits/sinuses and genital and toenail abnormalities.\(^4\) Lower-lip pits, which may resemble a depression or furrow, represent the opening of a tract leading from a mucous gland embedded in the lip.\(^5\) Lip pits may be divided into three types according to their location: commissural, midline upper lip and lower lip.\(^5\) The genital anomalies include cryptorchidism and bifid scrotum in males and hypoplastic labia majora and uterus in females. Various hypotheses include a primary microvascular abnormality with associated oedema leading to disturbance of epithelial tissues resulting in adhesion formation,\(^6\) excessive epithelial growth leading to fusion and secondary mesenchymal involvement,\(^7\) a primary collagen defect\(^8\) or a loss of programmed cell death.\(^9\)

We present this case of a 14-year-old girl who had difficulty in walking for 12 years. She was operated for cleft lip and palate at 2 years of age. Her two paternal uncles had similar features of this syndrome, but her parents were healthy and unrelated. There was no significant antenatal history. Her developmental and menstrual histories were normal. She had extensive popliteal webs, extending from the ischial tuberosities to the Achilles tendons as a fibrous cord, and was unable to extend her legs fully with tight Achilles tendon with ankle in equinus [Figures 1 and 2]. The genitalia were involved with hypoplasia of the labia majora. She had a surgical scar of a repaired cleft lip and palate and the lower-lip pits were present [Figure 3]. Magnetic resonance imaging of the right distal thigh and leg revealed diffuse fibrofatty changes in the muscles representing mild fatty atrophic changes and associated
PPS is a unique anomaly and hence underreported. A careful examination of the patient may identify lip pits in addition to the popliteal web. Meticulous...
physical examination of the family members and genetic counselling is also required for these patients.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest
There are no conflicts of interest.

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