between 5- 8 weeks of fetal life and is characterised by gradual compaction of myocardium, transformation of large intertrabecular spaces into capillaries, and evolution of the coronary circulation [1]. Clinically, patients may be asymptomatic or present with CHF, arrhythmia and embolic events [4-6]. The presenting symptoms include tachypnea, cyanosis, syncope, or failure to thrive, Chin, *et al*. [7] have described facial dysmorphism, including a prominent forehead, low set ears, strabismus, high- arched palate and micrognathia.

LVNC can be diagnosed by two-dimensional echocardiography and color Doppler [7,8]. The criterion for diagnosis is ratio of noncompacted to compacted layer of >2 measured at end systole.

There is no specific therapy. The mainstay of treatment is diuretics, ACE inhibitors and beta- blockers (Carvedilol) to improve the left ventricular systolic function. Anticoagulation (target INR – 2.0-3.0) is recommended when LVEF <40% [3]. Pignatelli, *et al*.[4] recommend oral aspirin and a metabolic cocktail including thiamine, coenzyme Q10, riboflavin and carnitine. Poor prognostic markers predicting death or heart transplant are age at onset, NYHA functional class III–IV, sustained ventricular arrhythmias, ratio of noncompacted to compacted layers, number of affected segments, LVEDD and abnormal lateral mitral tissue Doppler Ea velocity [3,9].

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Angiokeratoma Circumscriptum of the Tongue

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Angiokeratoma circumscriptum is rare cutaneous disorder. It usually presents as multiple,red, blue or black asymptomatic papules on lower extremities. Oral involvement, common in systemic form, is rare in localized forms. We report a case of angiokeratoma circumscriptum of tongue, involving both dorsal and ventral aspects.

Key words: *Angiokeratoma, Tongue*.

Angiokeratoma is the term applied to describe quite distinct clinical conditions that share a clinical presentation with asymptomatic hyperkeratotic cutaneous vascular lesions and a histological combination of superficial dermal vascular ectasia with overlying hyperkeratosis. The following five varieties are generally recognized: (*i*) generalized systemic type angiokeratoma corporis

diffusum of Fabry; (*ii*) Bilateral form occurring on the dorsa of fingers and toes- angiokeratoma of Mibelli; (*iii*) Localized scrotal form-angiokeratoma of Fordyce; (*iv*) Solitary papular angiokeratoma and (*v*) Multiple papular and plaque like -angiokeratoma circumscriptum [1]. All types, though differ clinically, share the same histological features, i.e. hyperkeratosis, acanthosis and dilated capillaries in the papillary dermis that are partly or completely enclosed by the papillomatous epidermis. Angiokeratoma circumscriptum presents as multiple purple papules that later become verrucous plaques. We report a case of 10 year old male, who presented with lesions of angiokeratoma circumscriptum, extending onto both the dorsal and ventral surfaces of tongue.

CASE REPORT

A 10 year old boy presented with multiple, red small raised lesions on the tongue for the last 4 years. The condition started as an asymptomatic single raised lesion on the undersurface of the tongue which gradually increased in number and extended onto the sides and upper surface of the tongue. There was no preceeding history of trauma or bleeding from lesions. Patient denied any history of similar lesions elsewhere on the body. His past medical history was unremarkable.

On examination of the oral cavity, the patient was found to have two raised lesions of sizes 1×1cm and 2×3 cm, on the dorsum of the tongue, anterior to the base, which were extending onto the ventral surface of the tongue. They were studded with multiple grouped, erythematous shining papules some of which had a keratotic top. The masses were non-friable and moderately tender on palpation. They were mobile, firm on palpation, and did not bleed on manipulation. The rest of the cutaneous and systemic examination was normal. A biopsy specimen of a representative tongue lesion showed parakeratosis, acanthosis, papillomatosis with large dilated spaces lined by normal appearing endothelium and filled with erythrocytes and organizing thrombi. On the basis of clinical examination and histopathological findings, a diagnosis of angiokeratoma circumscriptum of the tongue was made.

DISCUSSION

Angiokeratoma circumscriptum is a rare vascular malformation of the papillary dermis manifesting as one or several purple papules and blood filled cystic nodules that gradually become verrucous and coalese into plaques. They may be linear or zosteriform pattern and bleed readily from trauma. Vessels are ectatic histologically and may be thrombosed. The overlying epidermis shows variable degree of hyperkeratosis, papillomatosis and acanthosis. The elongated rete ridges may partially or completely envelop the dilated vessels. Usually, the lesions are present at birth, but in some cases they may occur during childhood as in our case and even in adulthood. Angiokeratoma may be associated with Klippel-Trenaunay, Weber syndrome, Cobb syndrome and other mixed vascular malformations. The lesions are typically situated on the lower leg, foot, thigh or buttock but may occur elsewhere on the skin [5,6].

Oral involvement in angiokeratomas is most commonly a component of angiokeratoma corporis diffusum which is associated with several inherited lysosomal disorders [6] It is rare in other types of angiokeratomas.

Till date there are three reported cases of angiokeratoma circumscriptum solely localized to the oral cavity in pediatric patients [2-4]. Kumar, *et al*. reported the

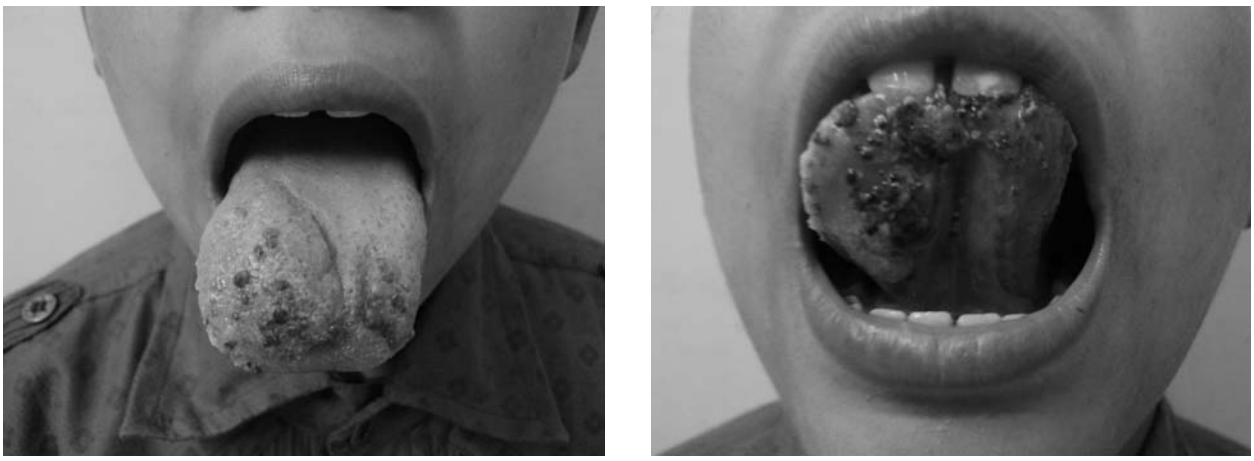

FIG.1 *Raised lesions of angiokeratoma circumscriptum on the (a) dorsum of the tongue and (b) extending onto the ventral surface of the tongue.*

case of a 16 year old boy with histologically defined angiokeratoma circumscriptum on the ventral aspect of the tongue [3]. Another 12 year old boy with angiokeratoma circumscriptum isolated to the ventral tongue was reported [2]. The third patient was a 6-year old male who presented with a 2 year history of recurrent mass on the dorsal tongue [4]. Our case of lingual angiokeratoma circumscriptum involved both the dorsal and ventral surfaces of the tongue. Another interesting point to be noted is that all four patients including ours were male patients.

The pathogenesis of angiokeratomas is still unknown. It has been reported to develop overlying an arteriovenous fistula and in areas of lymphangioma circumscriptum after local injuries [7,8]. Angikeratomas may be treated with complete surgical excision, cryotherapy and laser ablation including copper vapour, potassium tritanyl phosphate, and argon lasers.

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Aldosterone Synthase Deficiency Type II with Hypospadias

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Aldosterone synthase deficiency (ASD) type II was diagnosed in a 3 week old boy with severe dehydration. Elevated plasma renin activity, low-normal aldosterone, increased levels for 18-OH corticosterone (18-OHB) and 18-OH-deoxycorticosterone were measured. Sequencing revealed a homozygous mutation for c554C>T in exon 3 (p.T185I) (*CYP11B2)*. Hypospadias has so far not been reported in ASD.

Key words: *Aldosterone synthase deficiency type II, Hypospadias.*

Two types of aldosterone synthase deficiency (ASD) are described at the hormonal level: type 1 (ASD1), with undetectable aldosterone levels, while the levels of 18-hydroxy-11-deoxycorticosterone (18-OHDOC) are increased, levels of 18-OHB are reduced, and the ratio B/18-OHB is increased. Type 2 ASD (ASD2) is characterized by low aldosterone levels, increased 18-OHB and 18-OHDOC levels, as well as an increased 18-OHB/aldosterone ratio.

Clinical signs for ASD are failure to thrive, vomiting, and severe dehydration [1,2]. Hyperkalemia, hyponatremia, metabolic acidosis, elevated plasma renin activity and low or undetectable aldosterone levels are the main laboratory characteristics of ASD [1, 3]. Both types of ASD have not been described to have genital anomalies. Herein we describe a one year old boy with ASD2 and penile hypospadias.

CASE REPORT

The boy was born after uneventfull pregnancy and delivered at 39 wk gestation with a birth weight of 3400 g, and a length of 51 cm. The parents were young,