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Murray Puretic –Drescher Syndrome: Case Report of a rare syndrome

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Abstract

Puretic-Drescher syndrome is a rare, autosomal-recessive connective tissue disorder. We report a case of 1.5 years old child presented with multiple soft tissue swellings. The histopathology of cutaneous lesions showed classical features. This case is being reported as it is a rare entity.

Keywords: Bilateral nasal septum, Gingival hyperplasia, Hyaline deposition,Nodular skin lesions.

Introduction

Murray Puretic-Drescher syndrome is a rare autosomal recessive hereditary disease. It starts in early infancy with flexural contractures and innumerable skin papules and nodules with osteolytic bone lesions and gingival hyperplasia[1].We here report a child admitted in our hospital showing characteristic clinical and histopathological features.

Case Report

A one and half-year old female child presented to pediatric department of tertiary care hospital for delayed milestones and dermatology referral was sought for skin lesions. The child was mentally retarded. There were multiple soft tissue swellings involving nasal septum bilaterally ,ear pinnae, nape of neck, parietal and occipital prominence, interphalangeal joints of both hands and anal region(Figure 1).

The child was fourth in order of birth, born of non consanguineous marriage. Birth history was uneventful. She started developing swellings along the nasal septum and ear pinnae since the age of one year and three months.These swellings were small to begin with and progressively increased in size and number. Swellings were in the form of multiple, firm, non tender waxy papules and nodules. The movement of interphalangeal joint was painful and restricted.

Her vitals were stable .There was no lymphadenopathy or hepatosplenomegaly. The head circumference was 47 cm (1SD) whereas her weight and height was 7.5 kg(-2 to -3 SD) and 72 cm (-2SD) respectively. She had delayed motor and developmental milestones. Her dentition was delayed, while on examination there was gum hypertrophy. She had normal hearing and vision. There was no other systemic involvement. The complete haemogram, renal function tests, liver function tests, electrolytes and ultrasound of the

abdomen were performed to rule out systemic involvement. These were within normal limits. Radiograph of the chest was normal. Skeletal survey showed large soft-tissue shadows and CT scan of the head showed large soft-tissue shadows (Figure2,Figure3)

At this point of time, differential diagnosis included neurofibromatosis, murray puretic-drescher syndrome, congenital generalized fibromatosis and Winchester syndrome.

Considering the cutaneous involvement, a skin biopsy was performed from nape of the neck. The histopathological report revealed accumulation of hyaline material with fibroblast in the dermis(Figure 4).

The diagnosis of murray puretic-drescher syndrome was made after clinicopathological correlation. The parents were counseled about the progressive nature of the disease and the 25% chance of an affected future child. We could not go ahead with the genetic testing in our patient due to non availability of this facility in our institution.

The parents were offered surgical excision of the few lesions , however in view of high rate of recurrence they deferred surgical management and opted for conservative treatment. Child was advised multivitamins, vitamin d3, active physiotherapy and healthy diet. Parents were advised to follow-up 3 monthly to assess the progression of the disease.

Discussion

Murrey puretic-drescher/inherited systemic hyalinosis is a rare, autosomal recessive and hereditary disease with distinct clinical and histopathological features [3]. There is paucity of available literature with only < 70 cases reported worldwide and a few case reports from India [2-6].

The mutant gene has been mapped to 4q21[7].Various abnormalities in biosynthesis of glycosaminoglycans and collagen III- VI has been defined.Mutations in the capillary morphogenesis factor-2 gene has also been described[8]. The disease commonly affects children in the age group of 2-5 years. The symptoms include gingival hypertrophy, papules distributed around the nose, the ears, in the genital area and on the thighs, joint contractures, cutaneous pressure necrosis and ulceration[9,10].

Infantile systemic hyalinosis is a severe form of hyaline fibromatosis syndrome with widespread visceral involvement and invariably fatal outcome[11].

Close differentials of the condition due to the cutaneous nodules were neurofibromatosis, congenital generalized fibromatosis while isolated gingival hyperplasia, Winchester syndrome, and lipoid proteinosis may also be considered as differential diagnoses[11]. Treatment is predominantly symptomatic and palliative. Early surgical excision may help to prevent the appearance of new lesions although recurrence has been documented after excision[9]. Intralesional steroid injection and oral D penicilamine are other modalities[2].

Conclusion

Murray Puretic-Drescher syndrome is a rare autosomal recessive disease. Clinical suspicion prompts histopathological examination of the lesions followed by medical and surgical intervention as a part of palliative care. As of now there is no specific treatment for this disorder however genetic and preconception counseling should be done to prevent the disease.

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Legend Figure

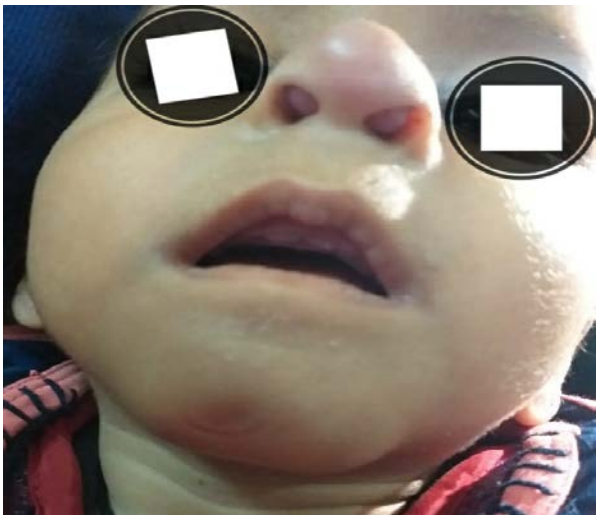

Figure1a: shows clinical presentation of the child in the form of multiple soft tissue swellings involving bilateral nasal septum.

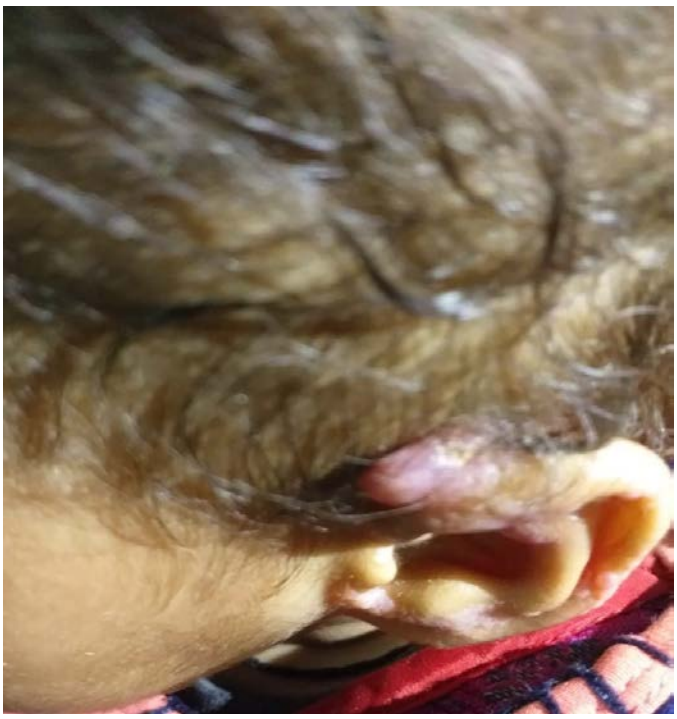

Figure1b: multiple, asymptomatic, non tender, pink to skin coloured papules clustered over the pinna of the ear.

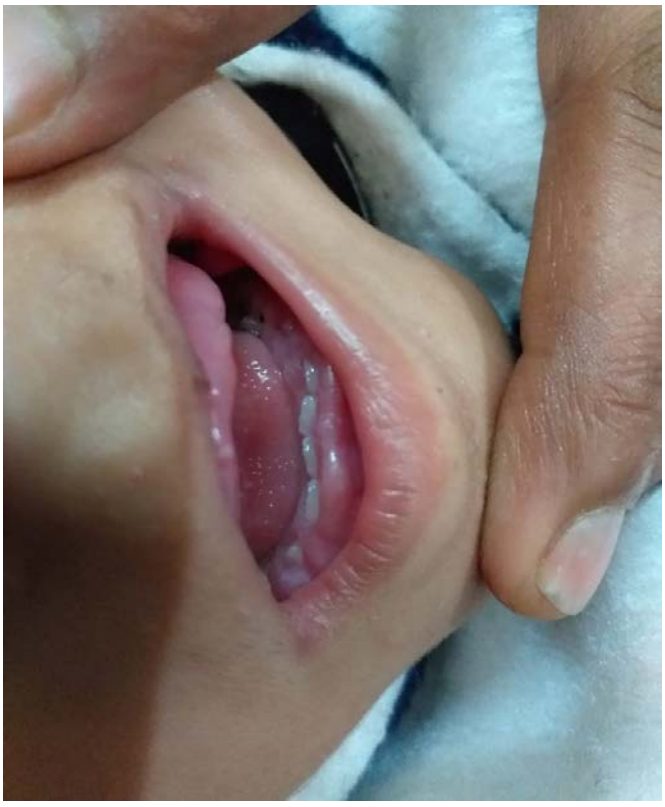

Figure1c: Dental examination of the child showed gingival hypertrophy.

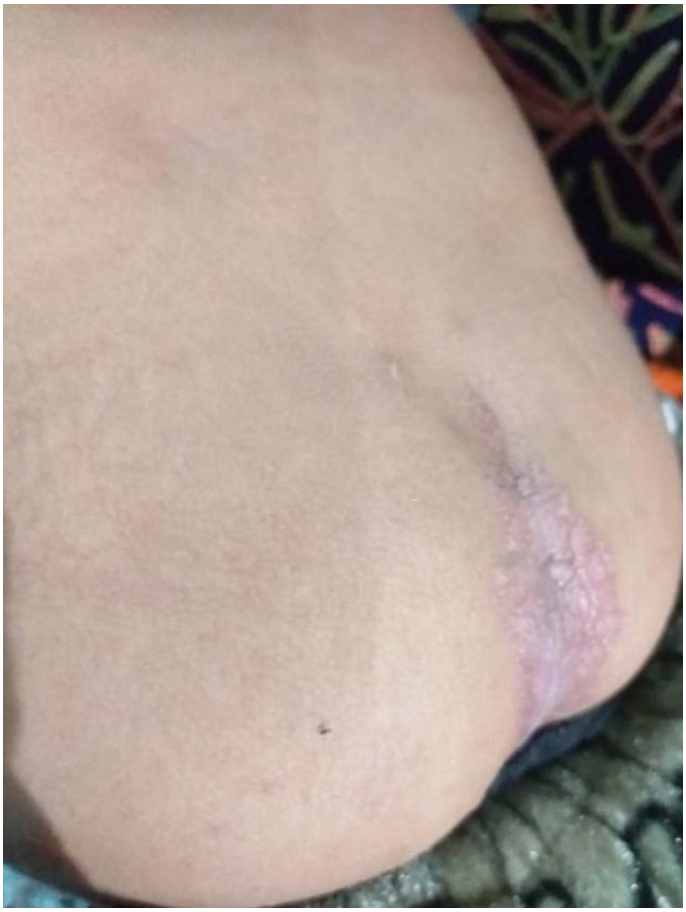

Figure1d: A pink fleshy linear plaque was seen in natal cleft and lower back.

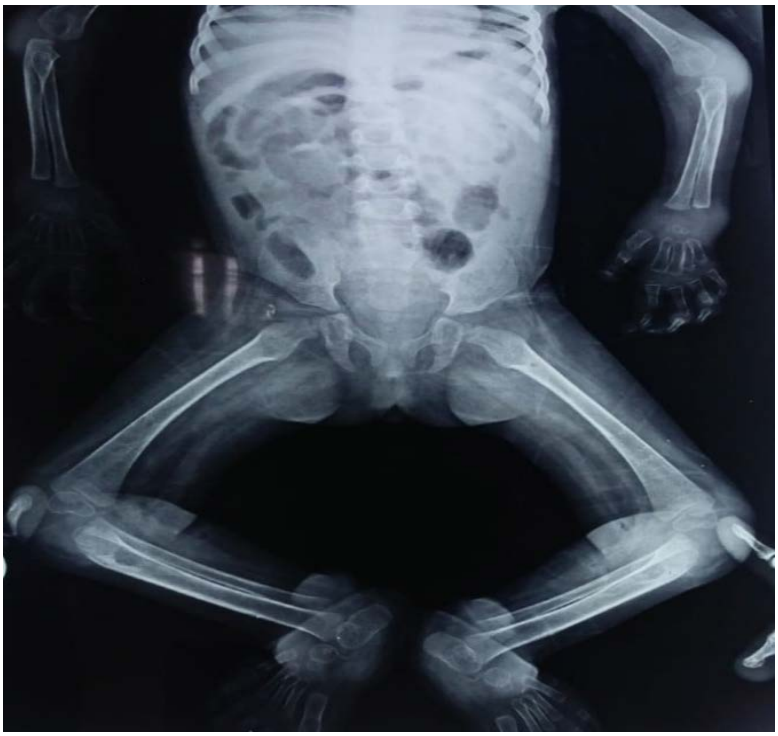

Figure 2: X-Ray shows multiple disseminated soft tissue nodular lesions involving major joints of both the limbs.

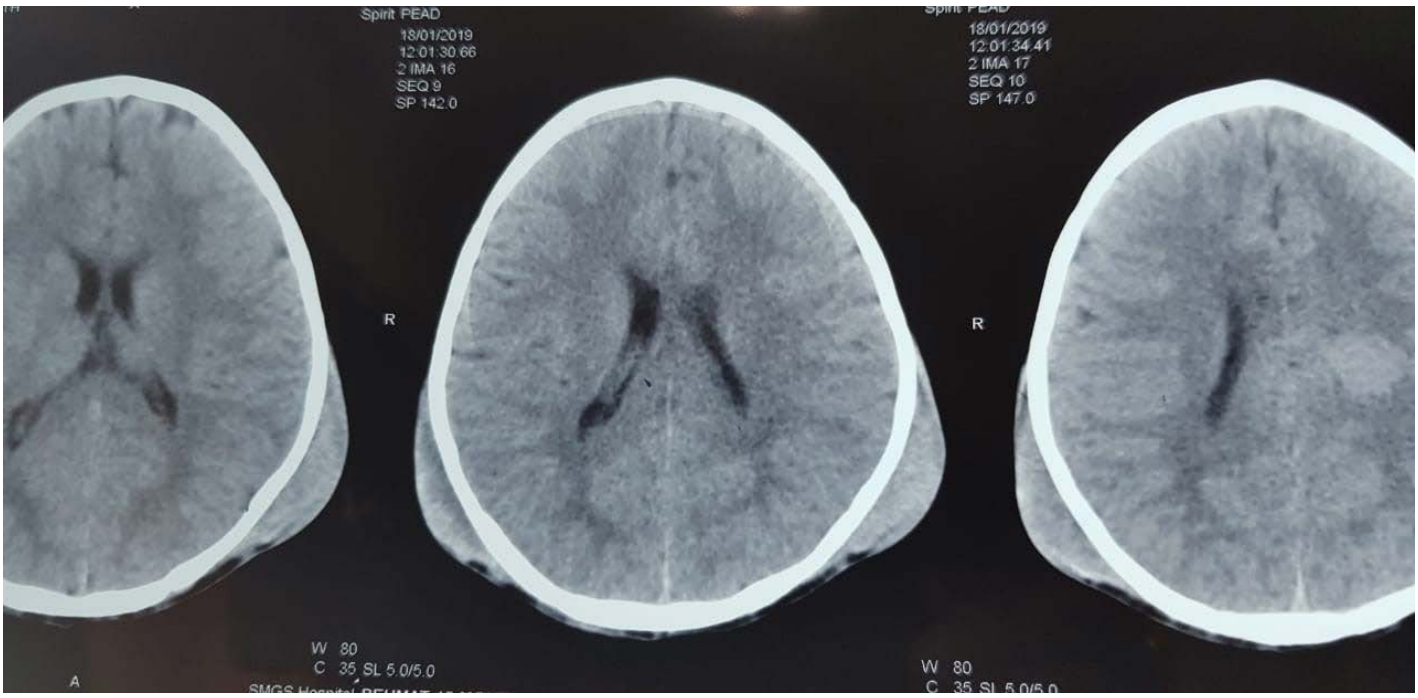

Figure 3: CT Scan shows symmetrical soft tissue scalp swellings in bilateral parietal region.

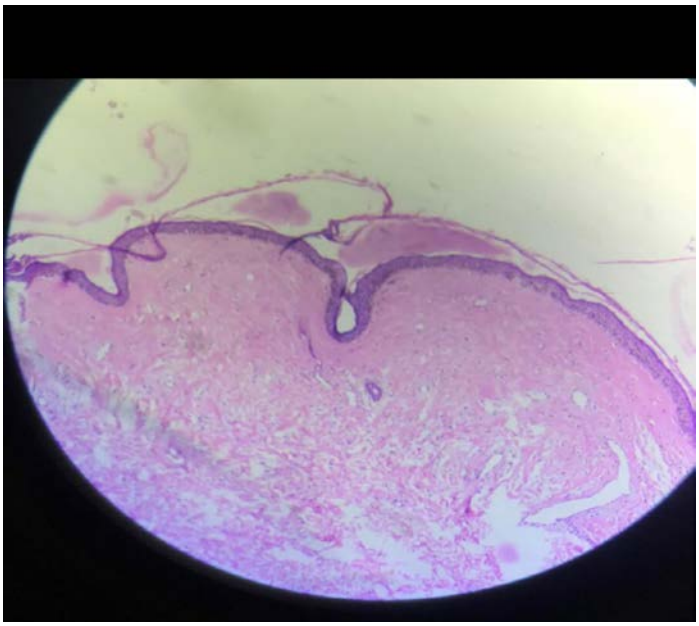

Figure 4: Histopathology of the skin lesions showed atrophic epidermis. Dermis shows presence of amorphous eosinophilic material with sparse uniform spindle cells embedded in it without any necrosis and atypia.(10x)