

Case Report

Cornelia De Lange Syndrome with generalized pustular psoriasis: a rare coexistence

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Abstract Cornelia De Lange Syndrome is a rare disorder of unknown etiology, characterized by peculiar facial appearance. Patients are small at birth and remain so as compared to children of the same age. Their ability to learn is delayed. All patients present delayed or limited speech development. A case of this rare disorder is reported here, which has a classical presentation, but unusual association with generalized pustular psoriasis not reported earlier.

Key Words Cornelia De Lange Syndrome, generalized pustular psoriasis

Introduction

Cornelia De Lange Syndrome is also known as Amsterdam Dwarf. It is a multiple congenital anomaly syndrome characterized by a distinctive facial appearance, prenatal and postnatal growth deficiency, feeding difficulties, psychomotor delay, behavioral problems and associated malformations mainly involving the upper extremities. Cornelia De Lange first described it as a distinct syndrome in 1933,¹ although Brachmann had described a child with similar features in 1916.²

Etiology and pathogenesis are not clearly known. Most cases are sporadic. It may be transmitted in an autosomal dominant pattern. Some features of the syndrome have occurred in relatives. Incidence is 1 per 50,000 to 100,000 live births. There is no predilection for any race or age.^{3,4}

Case Report

A 17-year-old girl presented to the Department of Dermatology, Unit II, Mayo Hospital, Lahore with generalized erythematous scaly plaques studded with pustules for 1 month (**Figure 1& 2**). Clinically a diagnosis of generalized pustular psoriasis was made which was later confirmed on histopathology. On history she was born after an uneventful pregnancy to a consanguineous couple. She was under weight at birth. She had a feeble and low pitched cry. The patient showed delayed milestones since birth. She started walking at the age of 3 years and talking at 6 years. There is history of recurrent respiratory tract infections. She developed pulmonary tuberculosis 5 years back, for which she took antituberculous therapy for 9 months and was cured completely.

General physical examination of the patient reveals a short statured, thin lean young girl with a weight of only 25 kg. The patient has a mask like expression less face with thick and confluent eyebrows (synophrys) and a

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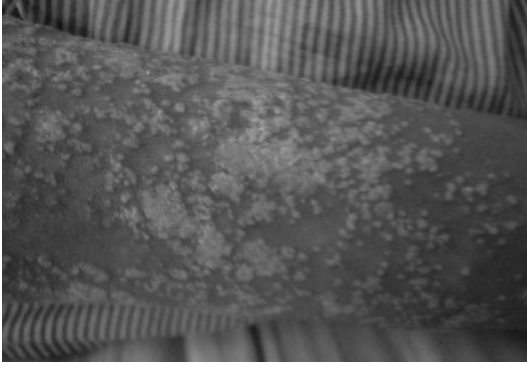


Figure 1 Multiple pustules on the forearm of patient coalescing to lakes of pus.

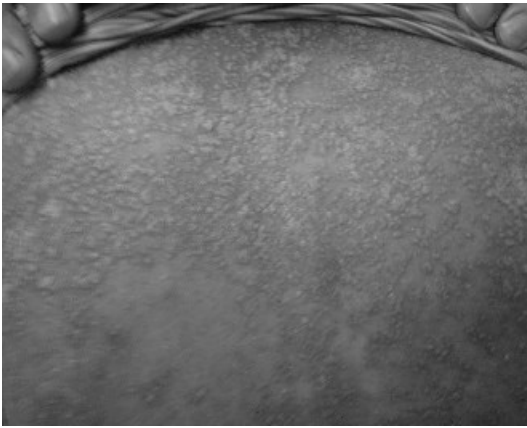


Figure 2 Erythematous plaque studded with multiple pustules on the upper back.

low hairline on the forehead and neck (**Figure 3**). She has hypertrichosis and long eye lashes (**Figure 4**). Her head size is small, lips are thin and especially the upper lip is long and thin. The nipples and genitalia were hypoplastic.

Ophthalmological examination reveals bilateral cataract. Examination of the central nervous system shows mental retardation (IQ of 54%). Cardiovascular system examination is unremarkable. Echocardiography was performed to rule out congenital heart disease and was found to be normal. The patient was given topical diluted steroids, oral antibiotics and investigated for systemic therapy.

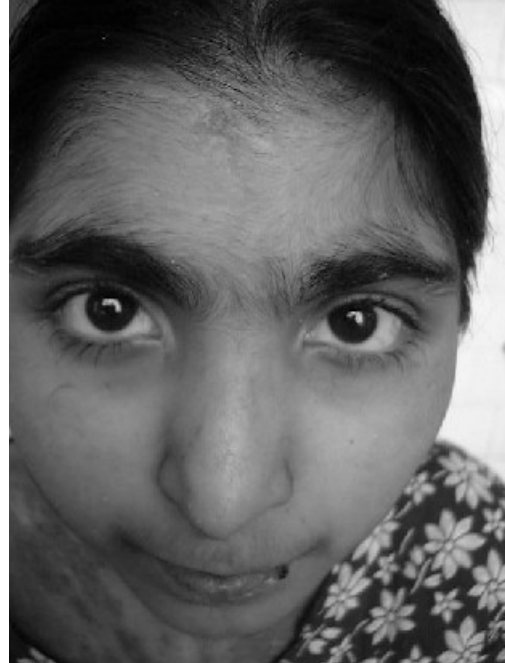


Figure 3 Synophrys an important feature of Cornelia De Lange syndrome.



Figure 4 Long eyelashes an important feature of Cornelia de Lange syndrome.

Discussion

Low birth weight, delayed milestones, recurrent infections, physical and mental retardation, expressionless face, hypertrichosis and synophrys in our patient led to the diagnosis of Cornelia De Lange syndrome.

Unlike our patient, not all cases develop classical features of the syndrome, many of

them presenting as forme fruste. The presence of generalized pustular psoriasis is in our case is a unique finding which is never mentioned in the literature before. We believe that it is a coincidental finding.

Occasional familial cases are reported but most are sporadic. The pattern of inheritance is probably autosomal dominant.⁵ Some features of the syndrome have occurred in the relatives.^{3,4} This syndrome has also been described in twins.⁶

There is a high incidence of chromosomal abnormalities, but these are not consistent.⁷ A phenotype similar to that of Cornelia De Lange syndrome may be observed in patients with a duplication of band q26-27 of chromosome 3. Molecular studies of genes mapped to this region of chromosome arm 3q have failed to identify mutations in patients with Cornelia De Lange syndrome.⁸ The dup (3q) syndrome simulates Cornelia De Lange syndrome but is probably fundamentally distinct.^{9,10}

The features like depressed bridge of nose, anteverted nostrils, widely spaced teeth, webbing and hyper extensibility of digits, bluish tinge around the skin of eyes and nose are not present in our case but the classical features of this syndrome did exist. However, some mildly affected individuals do not have a classical presentation.¹¹

Prenatal diagnosis is made after careful evaluation of Cornelia De Lange syndrome abnormalities on prenatal ultrasonography. These include growth retardation, limb defects, diaphragmatic hernia, hypo plastic forearms, underdeveloped hands and typical facial defects.¹³ We did the genetic counseling and referred her to

ophthalmologist for bilateral cataract surgery. She was given topical diluted steroids on psoriatic lesions, investigated for systemic methotrexate therapy and put her on 7.5 mg methotrexate per week. In our case life expectancy is normal because there are no major malformations.

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