

Cornelia De Lange Syndrome in Iraq

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Abstract

Background: Cornelia de Lange syndrome is a rare syndrome of highly variable phenotype making a spectrum ranging from classic syndrome with many cardinal features to mild condition few cardinal features. Typically patients with classic syndrome had growth and mental retardation and distinctive facial dysmorphism including thick (bushy) and / or long eyebrows commonly with synophrys, short nose with depressed or concave nasal bridge and/or upturned nasal tip, long or smooth or indistinct philtrum, thin upper lip vermilion and/or downturned corners of mouth, and low set ears. The diagnosis of the syndrome is clinical. Ocular abnormalities that can be associated with Cornelia de Lang syndrome squint, nystagmus, refractive errors, and ptosis.

Materials and methods: The occurrence of Cornelia de Lange syndrome has not been reported or well-documented. The first four Iraqi patients (Three boys and one girl) with Cornelia de Lange syndrome are described. The relevant literatures were reviewed with aim of determining the early documentation of the syndrome in the medical literatures.

Results: All the patients were sporadic cases and had growth retardation, severe mental retardation with significant developmental delay, thick eye brows with some degree of synophrys, short nose with depressed or concave nasal bridge, and low set ears. All the patients had normal karyotype. One male patient had all of the classical features including long smooth and indistinct philtrum, thin upper lip vermilion, and downturned corners of mouth. The second male patient had a concave nasal bridge that becomes more obvious during crying, nystagmus and bilateral convergent squint. The third boy had milder dysmorphic features. The fourth patient was a girl who was the second of a twin. She had severe growth retardation and was hypotonic with poor head control. She also had bilateral convergent squint, refractive error, and reduction in visual acuity.

Conclusion: The first four Iraqi patients with Cornelia de Lang syndrome are reported.

Keywords: Cornelia de Lang syndrome, Iraq, ocular abnormalities, nystagmus, squint.

Introduction

Cornelia de Lange syndrome is a rare syndrome of highly variable phenotype making a spectrum ranging from classic syndrome with many cardinal features to mild condition few cardinal features. Typically patients with classic syndrome had growth and mental retardation and distinctive facial dysmorphism including thick (bushy) and / or long eyebrows commonly with synophrys, short nose with depressed or concave nasal bridge and/or upturned nasal tip, long or smooth or indistinct philtrum, thin upper lip vermilion and/or downturned corners of mouth, and low set ears. The diagnosis of the syndrome is clinical [1-6]. Ocular abnormalities that can be associated with Cornelia de Lang syndrome squint, nystagmus, refractive errors, and ptosis. The majority of cases are thought to be caused by spontaneous genetic mutations [7-11].

Materials and Methods

The occurrence of Cornelia de Lange syndrome has not been reported or well-documented. The first four Iraqi patients (Two boys and one girl) with Cornelia de Lange syndrome are described. The relevant literatures were reviewed with aim of determining the early documentation of the syndrome in the medical literatures.

Results

All the patients were sporadic cases and had growth retardation, severe mental retardation with significant developmental delay, thick eye brows with some degree of synophrys, short nose with depressed or concave nasal bridge, and low set ears. All the patients had normal karyotype. The first patient (Figure-1) was a boy who was first seen at the age of ten months and had all of the classical features including long smooth and indistinct philtrum, thin upper lip vermilion, and downturned corners of mouth. The second patient (Figure-2) was a three-year old boy who had a concave nasal bridge that becomes more obvious during crying, nystagmus and bilateral convergent squint. The third boy had milder dysmorphic features with thick eyebrows, hypertichosis, long philtrum, low set ears. (Figure-3). The fourth patient (Figure-4) was a girl who was the second of a twin. She was first seen at about 16 months of age and she had severe growth retardation and was hypotonic with poor head control. She also had bilateral convergent squint, refractive error, and reduction in visual acuity. Her twin sister (Figure-5) had significant developmental delay, but she didn't have the characteristic facial features of Cornelia de Lang syndrome.

Literature review revealed that during four decades (1930s, 1940s, 1950s, 1960s), about 130 cases of Cornelia syndrome were reported in the medical literature.



Figure-1: The first patient was a boy who was first seen at the age of ten months and had all of the classical features including long smooth and indistinct philtrum, thin upper lip vermilion, and downturned corners of mouth.



Figure-3: The third boy had milder dysmorphic features with thick eyebrows, hypertichosis, long philtrum, low set ears.



Figure-2: The second patient was a three-year old boy who had a concave nasal bridge that becomes more obvious during crying, nystagmus, and bilateral convergent squint.



Figure-4: The third patient was a girl who was the second of a twin. She was first seen at about 16 months of age and she had severe growth retardation and was hypotonic with poor head control. She also had bilateral convergent squint.



Figure-5: The twin sister of the third patient had significant developmental delay, but she didn't have the characteristic facial features of Cornelia de Lang syndrome.

Discussion

The syndrome was named after Dutch pediatrician Cornelia Catharina de Lange [Figure-6], who described two patients in 1933, and in 1938, she reported the autopsy findings of her first patient, and described a third patient [1,2]. McArthur RG, Edwards (1967) estimated that 69 cases, including nine autopsies, have been reported in the literature and reported an other 20 patients with the syndrome [4].

Thereafter, about 41 case of the syndrome were reported during the 1960s, including the cases reported by Crisalli and colleagues, 1967; Lee and Kenny, 1967; Marsella, 1967; Huang, et al, 1967 (Two cases); Ganassi, et al, 1968 (Two cases); Stănescu and colleagues, 1968; Beer and colleagues, 1968 (Two cases); Hillman, et al, 1968; Familant, 1968; Calò and colleagues, 1968 (Four cases); Li Moli, 1968 (Two cases); Klosovski, et al (1968); Bartsocas and colleagues, 1968; Kaplan, 1968; Ferenczy and colleagues, 1968; Broholm and colleagues, 1968; Calò and colleagues, 1968; Szczepański (1969); Koike, et al (1969); Pelc and colleagues, 1969; Kondo and colleagues, 1969 (Two cases); Okita and colleagues, 1969; Boon, 1969; Fazekas and Nagy, 1969; Jacoby and Bonham, 1969; France and colleagues, 1969; Cherington and colleagues, 1969; Földioreanu, et al, 1969; Pashayan, et al, 1969 (Three cases); Say and colleagues, 1969; De Tomasi and Catapano, 1969 [12-44].



Figure-6: A sketch of a Dutch pioneer of pediatrics, Cornelia Catharina de Lange

During four decades (1930s, 1940s, 1950s, 1960s), about 130 cases of Cornelia syndrome were reported in the medical literature.

Conclusion

The first three Iraqi patients with Cornelia de Lang syndrome are reported.

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