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Mental-Growth Retardation, Microphthalmia, Microcornea, Iris and Uvea Colobomata, Transient Ocular Hypopigmentation, and Contralateral Optic Disc Colobomata, and Dilated Third Ventricle: A New Syndrome

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Abstract

There is a large number of dysmorphic mental-growth retardation syndromes associated with various combinations of low set ears, ocular abnormalities such as microphthalmia, microcornea, ocular hypopigmentation, and colobomatas of the iris, uvea, and optic disc. The aim of this paper is to report the novel occurrence of a new dysmorphic mental-growth retardation syndrome associated with asymmetric ocular abnormalities including microphthalmia, microcornea, iris and uvea colobomata, transient ocular hypopigmentation, and contralateral optic disc colobomata, and dilated third ventricle.

Keywords: Mental-growth Retardation, Microphthalmia, Microcornea, Colobomata, Transient Ocular Hypopigmentation

Introduction

A congenital syndrome is the occurrence of multiple physical abnormalities at birth in one ore more than one structure in the body that are caused by single etiologic mechanism which can be a chromosomal disorder, a single gene defect, or an environmental factor.

Physical abnormalities resulting from a cascade of effects related to a single localized anomaly should be more appropriately called a sequence rather than a congenital syndrome. A classical example of a sequence is the potter sequence.

The term dysmorphic syndrome is used in a wider sense to include both congenital syndromes and sequences. Therefore, potter sequence can also be included in the dysmorphic syndromes.

The discipline of dysmorphology has developed remarkably in association with tripling the number of recognizable patterns of malformations during the previous three decades [1, 2].

Various types of ocular colobomatas can be associated with microphthalmia and other ocular abnormalities. They are observed in chromosomal syndromes such as trisomy-13, trisomy-18, triploidy syndrome, and cat-eye syndrome. Microphthalmia and ocular colobomatas can be seen in non-chromosomal syndromes such as the CHARGE association, Joubert syndrome, Aicardi syndrome, Meckel syndrome, Warburg syndrome, Rubinstein-Taybi syndromes, Goldenhar syndrome, Lenz microphthalmia syndromes, and Goltz focal dermal hypoplasia [3-6].

The aim of this paper is to report the novel occurrence of a new dysmorphic mental-growth retardation syndrome associated with asymmetric ocular abnormalities including microphthalmia, microcornea, iris and uvea colobomata, transient ocular hypopigmentation, and contralateral optic disc colobomata, and dilated third ventricle.

Case report

An eighteen-month-old girl who was born to consanguineous parents was seen at the pediatric neuropsychiatry clinic of the Children Teaching Hospital of Baghdad Medical City because of motor developmental delay and ocular abnormalities.

The girl was hypotonic, and had poor head control and was not crawling and was unable to sit on the chair without slipping. She had poor awareness to the environment and was not responding to name, and has not showed any eye contact or social smile nor has started babbling.

She had the following abnormalities (Figure-1):

1-Frontal prominence.
2-Low set ears.
3-Macrostomia.
4-Growth retardation: Her weight was 6.280 Kilograms, and her height was 68 centimeters. 5-Brain abnormalities: dilated third ventricle.
6-Asymmetrical ocular abnormalities including:

A-Left eye abnormalities:

Microphthalmia.
Convergent squint of the left microphthalmic eye.
Microcornea.
Colobomata of the iris, uvea.
Transient ocular hypopigmentation observed at the age of five months and disappeared before the age of eighteen months.

A-Right eye abnormalities:

Colobomata of the optic disc.
Convergent squint of the right eye.
The rest of the physical examination was normal and echocardiography showed normal findings.
Ultrasound of the eye showed normal right eye and small irregular left eye globe confirming microphthalmia.
Karyotype showed normal findings.

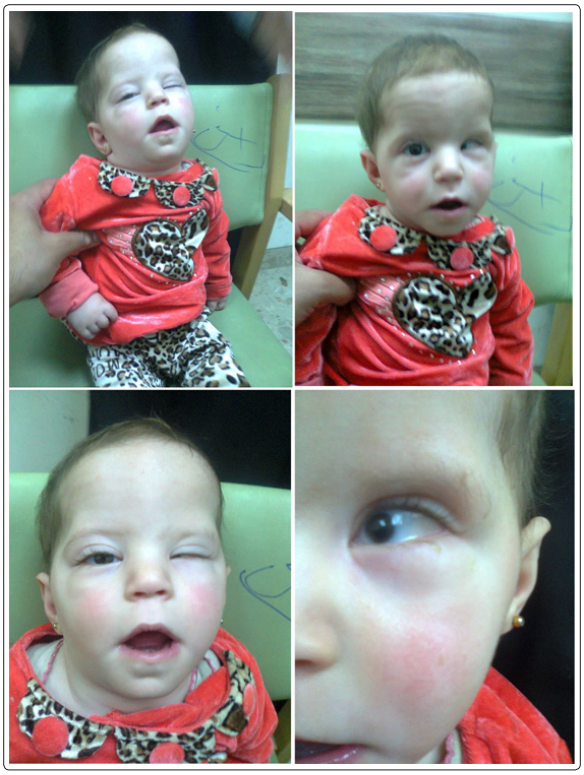


Figure 1: The girl at the age of eighteen months. She had low set ears and was hypotonia with poor head control and was unable to sit on the chair without slipping. She had prominent forehead, microphthalmia of the left eye, convergent squint of the microphthalmic eye and macrostomia

Figures 2: The girl at the age of nineteen months. She had bilateral convergent squints and was still hypotonic with poor head control and psychomotor retardation

Table 1: The clinical features of the new syndrome

Sporadic occurrence
Consanguineous parents
Hypotonia
Psychomotor retardation
Growth retardation
Cranio-facial dysmorphic features
Frontal prominence Low set ears Macrostomia
Brain abnormalities: dilated third ventricle
Asymmetrical ocular abnormalities
Left eye abnormalities: Microphthalmia Convergent squint of the left microphthalmic eye Microcornea Colobomata of the iris and uvea Transient ocular hypopigmentation observed at the age of five months and disappeared before the age of eighteen months
Right eye abnormalities: Colobomata of the optic disc Convergent squint of the right eye

The girl was treated with intramuscular cerebrolysin, intramuscular piracetam, and oral citicoline based on our published experiences with use of these agent in childhood neurologic and psychiatric disorders [7-11]. She received:

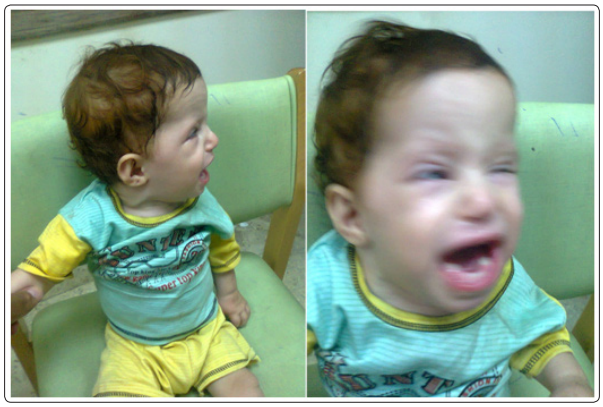
Intramuscular cerebrolysin 1ml daily in the morning, she received 10 doses. Intramuscular piracetam 1ml every other day in the morning, she received 10 doses.

Oral citicoline 1ml (100mg) daily in the morning for one month.

After treatment, the mother reported that the mother started crawling, babbling, smiling socially and responding to her name.

At the clinic, she had obvious lessening of psychomotor retardation. she with better head control and good alertness to the environment, and it was possible that she can see with her right eye.

Figure-3 show the girl at the age of twenty-one months after receiving a course of medical therapies.



Figures 3: The girl at the age of twenty one months after receiving a course of medical therapies. She had better head control and good alertness to the environment

Discussion

The increasing number of congenital syndromes demanded the evolution of approaches for their clinical recognition and diagnosis. It is generally recommended to make a list of the anomalies in the patient that are likely to be more specific, followed by listing the possible syndromes. Finally, the most probable diagnosis can be reached by narrowing of the diagnostic possibilities depending on the combination of anomalies the patient have [1, 2, 6].

There is a large number of dysmorphic mental-growth retardation syndromes associated with various combinations of low set ears, ocular abnormalities such as microphthalmia, microcornea, and ocular hypopigmentation, colobomata of the iris, uvea, and optic disc.

However, in this paper a patient with novel association of mental-growth retardation, low set ears and asymmetric ocular abnormalities is reported.

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