In Introduction

A congenital syndrome is the occurrence of multiple physical abnormalities at birth in one or 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 Presentation

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.
6. Asymmetrical ocular abnormalities including:
   - A-Left eye abnormalities:
     1. Microphthalmia.
     2. Convergent squint of the left microphthalmic eye.
     3. Microcornea.
     4. Colobomata of the iris, uvea.
     5. Transient ocular hypopigmentation observed at the age of five months and disappeared before the age of eighteen months.
   - A-Right eye abnormalities:
     1. Colobomata of the optic disc.
     2. 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 and 2) (Table 1).

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

- Intramuscular cerebrolysin in 1 ml daily in the morning, she received 10 doses.
- Intramuscular piracetam 1 ml every other day in the morning, she received 10 doses. Oral citicoline 1 ml (100 mg) 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).

### 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].

<table>
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<td>Frontal prominence</td>
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<td>Asymmetrical ocular abnormalities</td>
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Table 1: The clinical features of the new syndrome.
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.

Acknowledgement

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References


