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Cri-Du-Chat [5p-Syndrome]*

(Report of Two Cases)

Ergül Tunçbilek, M.D. / Kutay Tayşi, M.D.****
Sevim Balcı, M.D. / Kadir Tuğcu, M.D.*****

Cri du chat syndrome was first described in 1963 by Lejeune et al.[1] The characteristic cry in infancy, microcephaly, profound mental retardation and rather typical facial features are the clinical findings of this syndrome. The chromosomal abnormality is a partial deletion of the short arm of chromosome no.5

We would like to present two patients with this syndrome and a review of the literature.

Case 1: S. A. This three and a half year old girl was seen at Hacettepe Children's Medical Center, with complaints of mental and motor retardation. Her mother and father were both healthy, 26 and 27 years of age, respectively. The pregnancy and delivery were normal. The child was born after 40 weeks of gestation, but was very small and hypotonic. She was cyanotic and had a weak, high-pitched cry resembling that of a cat. She could not walk or talk at the time of examination.

Physical examination revealed her height to be 82 cm., weight 9.5 kg. and head circumference 43 cm (<3rd percentile). She was severely retarded. She had hypertelorism and an antimongoloid slant. There were two skin tags at the front of left tragus and a hemangioma 3x3 cm. in size on the left leg (Figure 1-2). The axial triradius were bilaterally distally displaced (t'). The other systems were found normal in physical examination.

Chromosome analysis of the peripheral blood revealed a partial deletion of the short arm of chromosome no. 5 (46, XX, 5p–) Figure 3. Chromosomal analyses of both parents were normal.

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* From Children's Medical Center, Division of Clinical Genetics, Hacettepe University, Ankara.
** Associate Professor of Pediatrics.
*** Resident in Pediatrics.

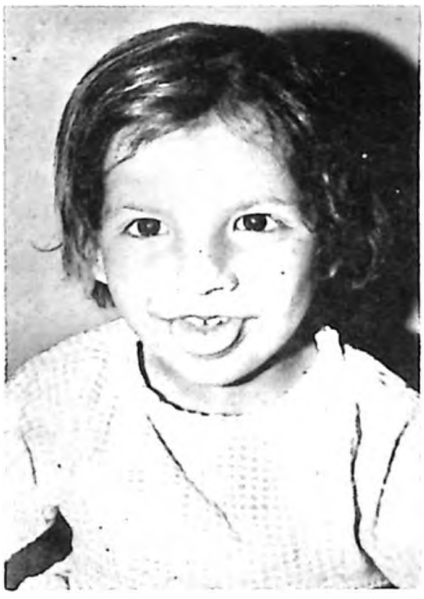

Figure 1
Case 1

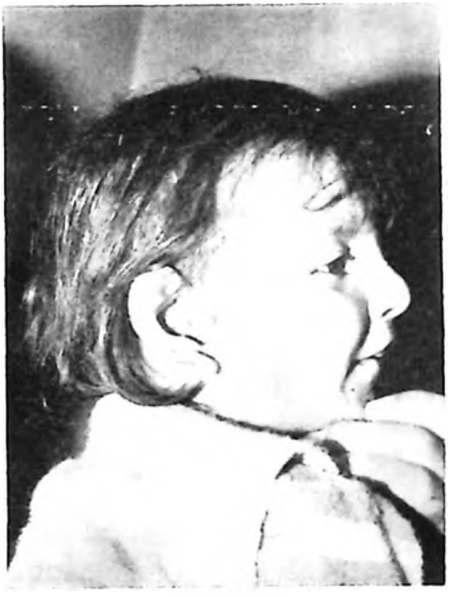

Figure 2
Profile of Case 1. Note the skin tag at the front of the tragus.

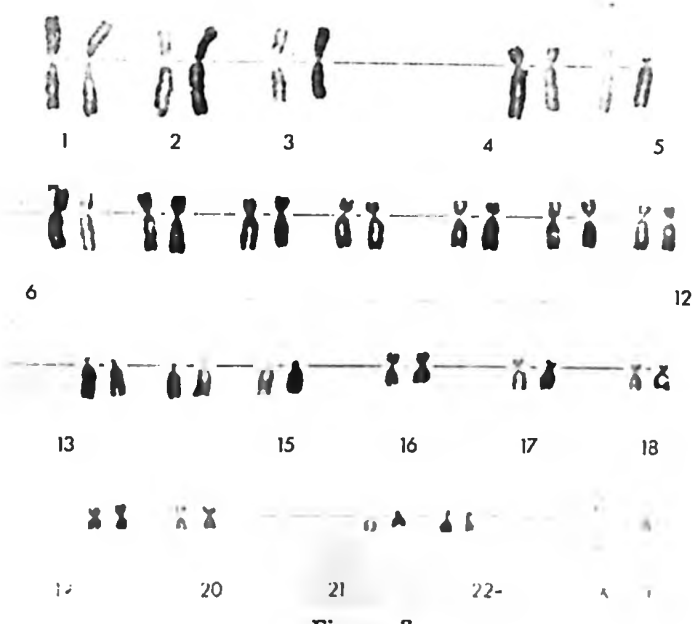

Figure 3
Karyotype of Case 1

Case 2: Ö. A., a 40-- day old boy, was hospitalized with a complaint of gastroenteritis.

The mother and father were both healthy and 24 and 26 years of age, respectively. The pregnancy and delivery were normal. The patient was born after 38 weeks gestation and was cyanotic at that time, birth weight being 1,770 gm.

Physical examination revealed his height to be 44 cm, weight 2 kg. and head circumference 31 cm (< 3rd percentile) Figure 4. His general condition was poor, he was hypotonic and dyspneic. He had hypertelorism and was crying with a cry resembling the mewing of a cat. There were bilateral inguinal hernia. Dermatoglyphic analysis revealed an increased number of whorls, distal axial triradius and bilateral simian creases.

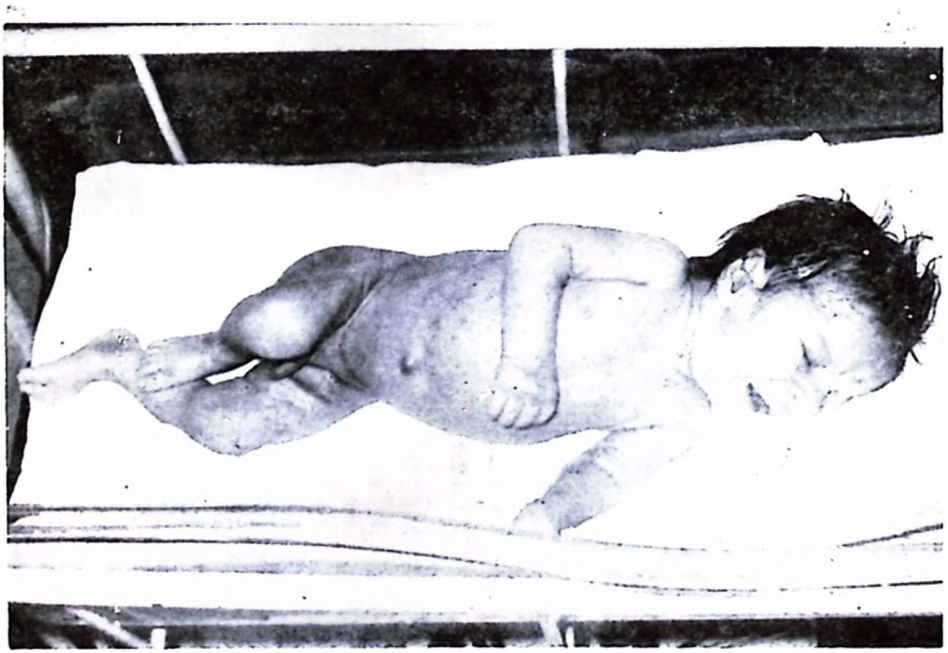

Figure 4
Case II

Intravenous pyelography was normal but an EEG revealed supression of cerebral bioelectric activity.

Chromosome analysis revealed partial deletion of the short arm of chromosome no. 5. The father's chromosome analysis was normal. In

G banded preparations, balanced translocation were observed between chromosomes no. 5 and no. 16 in the mother's karyotypes [46, XX, t (5 p-; 16p+)] (Figure 5).

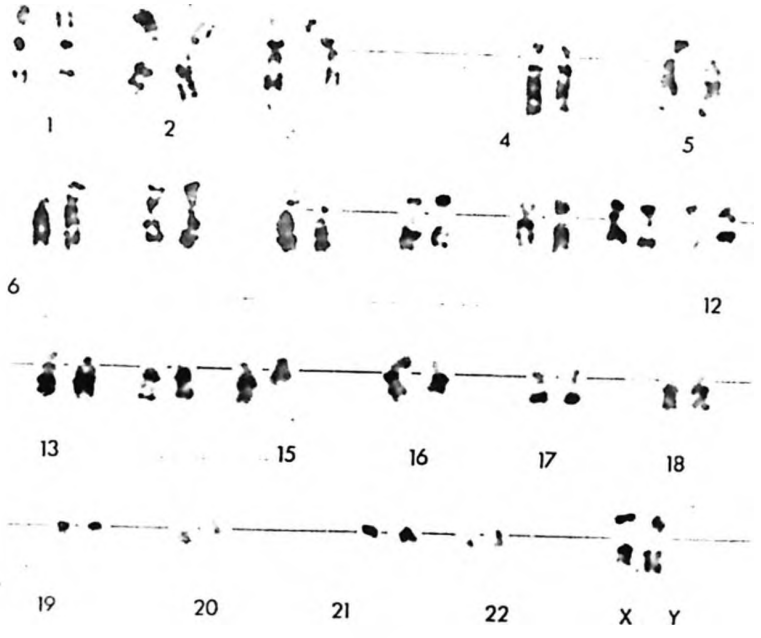

Figure 5
Giemsa banded karyotype of the Case II 46, XX, t (5p–; 16 p+)

Discussion

The most striking clinical feature of this syndrome is the characteristic cry resembling the mewing of a cat hence the name, "cat cry syndrome." The mewing cry becomes less pronounced with the advance of the age of the patient, thus making the diagnosis more difficult in older patients. The cause of this cry is not known exactly. Malacia of the cartilages, smallness, and subglottic malformations of the larynx have been observed but laryngoscopic findings can be normal despite a high pitched voice. After careful acoustic analyses Schroeder et al[5] concluded that the cat cry is caused by an organic or functional cerebral lesion and not by a malformation of the voice-forming organs. Micheau and Schlumberger[6] found minimal changes in the vestibule of the larynx as the only anomaly and concluded that a malformation of the nerve center or pathways controlling phonation must be responsible for the characteristic cry.

Mental retardation and microcephaly are the leading signs in addition to the cat cry. The mental retardation is usually severe and can amount to the degree of idiocy. The pathology of the brain is not typical.

Intrauterine growth retardation is common. More than half the patients had birth weights of 2,500 gm. or less, although the gestation time was normal in most.

Usually muscular hypotonia is noted but hypertonocity has also been observed. Our first patient was hypotonic only during the newborn period.

Although there is not a typical face in this syndrome, facial asymmetry, round face, hypertelorism, epichanthal fold, oblique palpebral fissures, low set and or poorly formed ears are frequently encountered. Congenital heart disease, variable in type, can be diagnosed in nearly 30 percent of the cases. Genitourinary tract abnormalities such as absent kidney and spleen, cryptorchidism or inguinal hernia are rarely seen anomalies. Cleft lip and cleft palate, myopia, optic atrophy, preauricular skin tag, bifid uvula, dental malocclusion, hemivertebra, scoliosis and premature graying of hair are also occasional abnormalities seen in this syndrome.[7, 8]

Simian crease can be observed in more than half of the patients. Distal axial triradius is the most commonly observed dermatoglyphic finding in this syndrome.

Clinical findings of our patients and comparison with those in the literature are shown in Table I.

TABLE I

CLINICAL FINDINGS OF OUR PATIENTS AND COMPARISON WITH THOSE OF THE LITERATURE

	1. case	2. case	Literature[8]
Low birth weight	+	+	72 %
Cat-like cry	+	+	100 %
Mental deficiency	+	?	100 %
Hypotonia	+	+	78 %
Microcephaly	+	+	100 %
Round face	+	+	68 %
Hypertelorism	+	+	94 %
Epicantic folds	—	—	85 %
Antimongoloid slant	—	+	81 %
Strabismus	—	—	61 %
Facial asymmetria, low-set easrs	—	—	58 %
Congenital heart disease	—	—	30 %
Simian creases	+	—	81 %
Distal axial triradius	+	+	40 %

Patients with the cat cry syndrome have a deletion of the short arm of chromosome no. 5 (46,5p–). This chromosomal abnormality has appeared as a fresh phenomenon in the majority of the cases. Our Case 1 was a fresh mutation, similar to most of the patients with this syndrome. The mother of Case 2 had a balanced translocation between chromosomes no. 5 and no. 16 [46, XX, t (5p– ;16+)]. Approximately 10-15 % of the cases have a balanced translocation parent with an increased risk of recurrence.[8]

Summary

Two cases with cri du chat syndrome are presented. The characteristic cry resembling the mewing of a cat was the most prominent finding in both patients during the newborn period. In addition to this, low birth weight, microcephaly, round face, hypertelorism, hypotonia and distal axial triradius were observed.

One of the patients is considered to be the result of a fresh mutation and the mother of the second case had a balanced translocation between chromosomes no 5. and no. 16.

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