

## Case Report

### Cri-du-chat syndrome

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**C**ri-du-chat syndrome (CDCS) is a rare chromosomal disorder, refers to a unique combination of physical and mental characteristics associated with a loss of genetic material on the distal short arm of the fifth chromosome.<sup>1</sup> The incidence of CDCS is between 1:25,000 to 1:50,000 births. The prevalence among individuals with mental retardation is about 1.5 in 1000.<sup>2</sup> A significant female predominance exists in affected newborns, with a male-to-female ratio of 0.72.<sup>2</sup>

Subtle dysmorphism with neonatal complications and a high-pitched cry typically initiate diagnostic evaluation by cytogenetic studies.<sup>2,3</sup> Currently, there is no cure for cri-du-chat syndrome. The most successful approach in the management of children with CDCS requires a multidisciplinary team approach.<sup>4</sup> The case presented below will remind us how to reveal, suspect and diagnose Cri-Du-Chat syndrome, a rare case in pediatric.

#### Case

D, a female neonate, was admitted to perinatology Dr M Djamil Hospital in February 24<sup>th</sup> 2005 with the chief complaint of low birth weight. The baby was born by cesarean section due to premature rupture of the membrane (19 hours) with opalescent amniotic fluid. The baby was vigorous and pink. There was no consanguinity's history, congenital anomaly, or mental retardation in family. She was a second child and the mother never used hormonal therapy. The first pregnancy was normal.

On physical examination the baby looked active, had high tone and weak cry like cat. Heart rate was 146/minute, respiration rate was 46/minute, temperature was 36.7°C, body weight was 1800 gram and length 44 cm. There was dysmorphism of the face, i.e. epicanthal folds, hypertelorism, micrognathia and low set ears. We found microcephally and no bulging of fontanel. Heart, lung, abdomen and genitalia examinations were normal. On extremities there was overlapping of fingers, rocker bottom feet and no simian crease. Estimation of gestational age was 34-35 weeks, appropriate for gestational age.

Laboratory findings showed hemoglobin 13.2 g/dl, leukocyte 18,100/ $\mu$ l and blood sugar 122 mg/dl. Chromosomal analysis showed deletion of short arm's (p) fifth chromosome with karyotype 46 XX, del 5 (5 p- syndrome / Cri-du-Chat Syndrome). During nursery, the baby was fine and was breastfed. She was discharged on day 6<sup>th</sup> nursery with body weight of 1800 gram.

She never had respiratory problem and feeding difficulty during observation for 14 months. Stridor vanished when she was at 13 months old. Her cat like cry disappeared at 11 months old. Her development was good enough, she could smile and follows thing with her eyes

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at 2 months old, lay face downward at 3 months, laughing at 6 months, hold thing with two fingers at 10 months and bubbling at 14 months. Her weight at 14 months old was 6 kg with average increase of body weight about 300 grams/month and growth velocity of 26 cm/year. Her head was still microcephalic but dysmorphism of her face decreased.

## Comments

There are a number of genetic mishaps which can result in a child being born with cri-du-chat syndrome and all involve a missing or deleted part of the short arm of one of the pair of number five chromosomes. It is called a deletion syndrome because part of the short arm is missing or deleted. That missing piece must contain a certain region of the short-arm for cri-du-chat syndrome to result.<sup>2,4</sup> A partial deletion of the short arm of chromosome 5 is responsible for the characteristic phenotype.<sup>2</sup> Approximately 80% are caused by a spontaneous deletion in the child, 10-13% by an error in a number five chromosome in either parents and the remaining 7-10% result from rare genetic anomalies.<sup>5,6</sup>

In most cases, this abnormality happens spontaneously, without family history. Parental translocation were present in slightly more than 10% of the families, so we still need chromosomal analysis of the parents to rule out this possibility. If a parent have structural rearrangements, the risk to have other CDCS child is substantially high, but the recurrence risk for de novo case is 1% or less.<sup>3,5</sup> Unfortunately, her parents didn't want to do this examination for economic reason and they didn't plan to have more children now. Genetic counseling for her is also needed because she can deliver viable affected offspring, with an estimated recurrence risk of 50%.

During infancy, the diagnosis of cri-du-chat syndrome is strongly suspected if the characteristic cat-like cry is heard.<sup>7</sup> If a child has this unusual cry or other features seen in cri-du-chat syndrome, chromosome testing should be performed. Chromosome analysis provides the definitive diagnosis of cri-du-chat syndrome.<sup>6</sup> This patient was diagnosed as Cri-du-Chat Syndrome based on a cat like cry, dysmorphism of face and chromosomal analysis that showed deletion of short arm's (p) fifth chromosome.

Individuals with cri-du-chat have a 10% mortality during infancy due to complications associated with congenital heart defects, hypotonic, and feeding difficulties.<sup>2</sup> Currently, death occurs in 6-8% of the overall population affected with the syndrome. Pneumonia, aspiration, congenital heart defects, and respiratory distress syndrome are the most common causes of death.<sup>2</sup> We didn't find neonatal complications in her such as respiratory distress, jaundice and feeding difficulty during nursery at hospital. During observation for 14 months, we didn't find feeding and respiratory problems.

Cat like cry disappeared when she was at 11 months old, stridor vanished at 13 months and dysmorphism of her face decreased. Baccichetti *et al*<sup>8</sup> pointed out that with advancing age the clinical picture of the CDCS become less striking. Her weight increased about 300 gram/months and at 14 months achieved 6 kg, but she was still microcephalic. It is known that growth rate of children with cri-du-chat syndrome may be normal in the early months but frequently slows somewhat during toddler and schools years resulting in growth parameters that are below average.

Until recently, little was known about the cognitive function of patients. Recent literature indicates that many children can develop some language and motor skills. These children attain developmental and social skills at 5 to 6 years old, although their linguistic abilities are seldom as advanced. Older, home-reared children are usually ambulatory, able to communicate verbally or through gestural sign language, and are independent in self-care skills.<sup>2,4</sup>

Currently, there is no cure for cri-du-chat syndrome. Treatment consists of supportive care and developmental therapy.<sup>5</sup> Early intervention programs, using a variety of therapies and educational strategies, focused on enhancing physical, intellectual, sensory and social development and have been shown to greatly improve the future outlook for the child.<sup>2,4</sup> Early intervention programs should include physiotherapy, speech therapy, occupational therapy, sensory integration (if available) and behavioural management (if necessary).<sup>2,5</sup>

Fortunately, both of her parents are educated and also gives her more attention. With early and consistent educational intervention, as well as physical and language therapy and support from her family, we hope that she will be capable to reach their fullest potential and can lead meaningful life.

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