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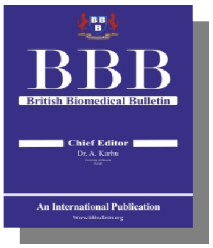

Case Report

Cri-du-chat Syndrome Misdiagnosed as Cerebral Palsy - A Case Report

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ABSTRACT

Cri du chat (CCS) is a rare chromosomal abnormality characterized by the deletion on short arm of chromosome 5.The clinical features include cat cry, microcephaly, dysmorphic facies, severe psychomotor and mental retardation. The diagnosis is usually based on the clinical phenotype with cat cry. Karyotyping and FISH analysis is used to confirm the diagnosis. We are reporting an eleven year old girl with Cri- du- chat syndrome who had been misdiagnosed as a case of cerebral palsy.

Introduction

Cri-du-chat syndrome (CCS) is a rare chromosomal abnormality characterized by deletion of variable length of chromosome 5.This syndrome was first described by Lejeune *et al*. in 1963[1]. The incidence of this condition is estimated between 1:15,000 and 1:50,000. The clinical features include cat cry, microcephaly, dysmorphic facies, severe psychomotor and mental retardation. We are reporting a case of Cri du chat syndrome which has been misdiagnosed as a case of cerebral palsy with seizure disorder.

Case Report

11 year old girl presented with generalized tonic-clonic convulsions to our hospital and on careful history taking, it was known that she had convulsions since birth and was on three anticonvulsants for the same. She was seizure free for the past 3 ½ years prior to admission and the drug compliance was good. She is the first of the siblings born out of a non consanguineous marriage and there is no history of birth asphyxia with developmental delay in all domains since birth. There is a history of medical termination of pregnancy in the second trimester for the second pregnancy due to increased nuchal translucency in anomaly scan and positive triple test. The chromosomal analysis of the abortus was not performed. The youngest sibling is a healthy six year old girl. On examination, she had severe wasting, cat like cry, epicanthal folds, high arched palate, dental malocclusion, micrognathia, low set ears, microcephaly, transverse flexion creases in the palms, severe hypotonia, hyporeflexia and flexor plantar response. There was a short systolic murmur on auscultation. Ultrasonogram of the abdomen revealed a dilated pelvic calyceal system and altered echotexture of liver. Echocardiogram revealed an atrial septal defect of 3 mm, left to right shunt with normal ejection fraction. She had been diagnosed elsewhere as a case of cerebral palsy. A thorough search of literature was instituted and the particular phenotype with cat like cry was suggestive of Cri-du-chat syndrome. Genetic analysis was planned and it proved the diagnosis. The girl was advised rehabilitation, physical therapy and anticonvulsants.

Discussion

The girl had been misdiagnosed as a case of cerebral palsy. The points against the diagnosis of cerebral palsy are hyporeflexia, bilateral flexor plantar response and no history of birth asphyxia. Though the features of the CCS are not specific, the phenotype as a whole (cat cry, hypotonia, severe psychomotor retardation, dental malocclusion, and microcephaly) points towards the diagnosis. The confirmatory test would be the chromosomal analysis and in cases where chromosomal analysis was normal, fluorescent in-situ hybridization (FISH) can be performed[2-5]. Nearly 85% of cases occur from sporadic de novo deletions and the remaining 15% due to unequally segregated parental translocation[6]. The critical region for the high-pitched cry maps to 5p15.3, and the chromosomal region involved in the remaining features maps to 5p15.2[6]. The risk of recurrence is very negligible for the cases of a de novo deletion, which are the most frequent[7]. The clinical features at birth are low weight (mean weight 2614 g), microcephaly, round face (83.5%), large nasal bridge (87.2%), hypertelorism (81.4%), epicanthal folds (90.2%), downward slanting palpebral fissures (56.9%), down-turned corners of the mouth (81.0%), low-set ears (69.8%), micrognathia (96.7%), abnormal dermatoglyphics (transverseflexion creases) (92%) and the typical cry (95.9%)[8,9]. The case report emphasizes two important

issues-one is the fact that all cases of developmental delay are not cerebral palsy and need for karyotyping and chromosomal analysis in cases of global developmental delay with hyporeflexia and facial dysmorphism. By doing a chromosomal analysis, the genetic counselling of the couple for future pregnancies can be done with utmost precision. Fortunately in our case, the couple have completed family, with the youngest sibling being normal, suggests that the case of Cri du chat would have occurred as a result of spontaneous deletion of short arm of chromosome 5, which rarely recurs.

Disclaimer

None.

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None.

Competing interest

None.

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774.

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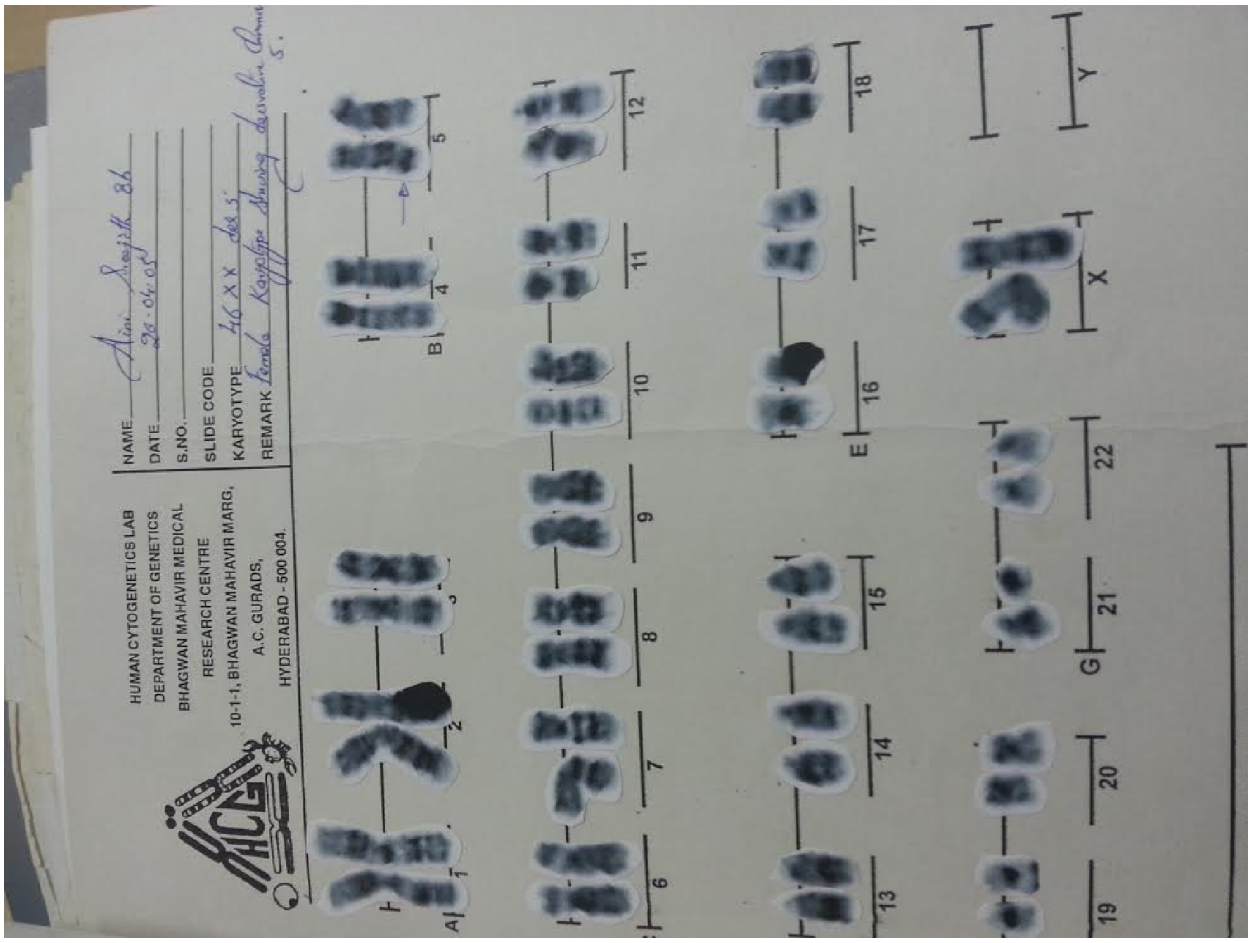

Figure 1. Karyptyping clinching the diagnosis of criduchat syndrome

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