JMSCR Vol||04||Issue||12||Page 14759-14761||December 2016

www.jmscr.igmpublication.org
Impact Factor 5.244
Index Copernicus Value: 83.27
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v4i12.72

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

# 18q Deletion Syndrome – A Case Report

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**ABSTRACT**
*The 18q deletion syndrome is among one of the commonest cytogenetic abnormalities with an incidence of 1 in 40000 live births without any ethnic predilection. Here we a present a with multiple dysmorphic features like micrognathia, large forehead, flat midface, hypertelorism, prominent antihelix, moon face with full cheeks were recognized, flat nasal bridge, thin lips and short neck and overlapping of the fingers. A MRI brain was done which showed enlarged ventricle with abnormal signal intensities in bilateral frontal and periventricular region. High resolution and G-banding chromosome analysis using peripheral lymphocytes was performed due to her many symptoms, which revealed a unique karyotype of 46, XX, del 18q3.1.*

## INTRODUCTION

The 18q deletion syndrome is among one of the commonest cytogenetic abnormalities with an incidence of 1 in 40000 live births without any ethnic predilection [(1)]. The condition is usually characterized by developmental delay, seizures, obesity, abnormal behavior, and minor facial anomalies including ptosis, bilateral epicanthus strabismus, short and slightly down-slanting palpebral fissures, full cheeks(2-6).Other features that are common in 18q deletion syndrome include short stature, weak muscle tone (hypotonia), narrow auditory canals leading to hearing loss, and limb abnormalities such as foot deformities and thumbs that are positioned unusually close to the wrist. Eye movement disorders and other vision problems, genital abnormalities, heart disease, and skin problems may also occur in this disorder. The overall level of mental retardation appears to be mild in patients with deletions distal to 18q21.33 and severe in patients with deletions proximal to 18q21.31. The critical region for the typical 18q-phenotype is a 4.3 Mb region within 18q22.3-q23. The maximum estimated size of the deletion ranges from 7.7 to 29.4 Mb.

## CASE REPORT

The patient was born out of a non consanguineous marriage with a weight of 3,205 g and height of 46.5 cm. Her head and chest circumference were each 33.0 cm. Her mother had one previous normal delivery. Delivery at 39+ weeks, gestation was difficult, and he was diagnosed with the meconium aspiration syndrome. At the time of her birth, her father and mother were 39 and 31 years of age, respectively. Although slight, mild muscle hypotonia of her extremities was observed, signs of birth asphyxia were noted at delivery. Multiple dysmorphic features like micrognathia, large forehead, flat midface, hypertelorism, prominent antihelix, moon face with full cheeks were

recognized, flat nasal bridge, thin lips and short neck and overlapping of the fingers (FIG 1-3).The child had an episode of convulsion on day 12 of life after which a MRI and EEG was planned.MRI brain showed abnormal signal intensities in bilateral frontal and periventricular region with dilated ventricles suggestive of birth asphyxia/ congenital infection (FIG 4,5) and sleep-induced electroencephalogram exhibited multifocal spikes in both hemispheres of the centro-parietal area. A BAER test was done which showed significant conductive abnormality. Echocardiography did not reveal any abnormality. To explore the cause of her facial anomalies, epilepsy and other conditions, we performed several examinations. Blood and urine screening tests showed normal data. High resolution and G-banding chromosome analysis using peripheral lymphocyte was performed, which revealed unique karyotype of 46, XX, del 18q3.1.

**DISCUSSION**

The 18q deletion syndrome is a well-described disorder resulting from a partial deletion of the long arm of chromosome 18. The breakpoints vary greatly between different reports. The clinical features of this syndrome include short stature, hypotonia, hearing impairment, dysmorphic features, foot deformities, low levels of immunoglobulin A and growth hormone deficiency. The incidence of these clinical symptoms in different patients with 18q syndrome is variable.

The patient reported here has a severe form of the 18q syndrome, presenting with most of the characteristic features of this condition. This severe clinical picture is explained by the cytogenetic findings, namely a deletion proximal to 18q21.

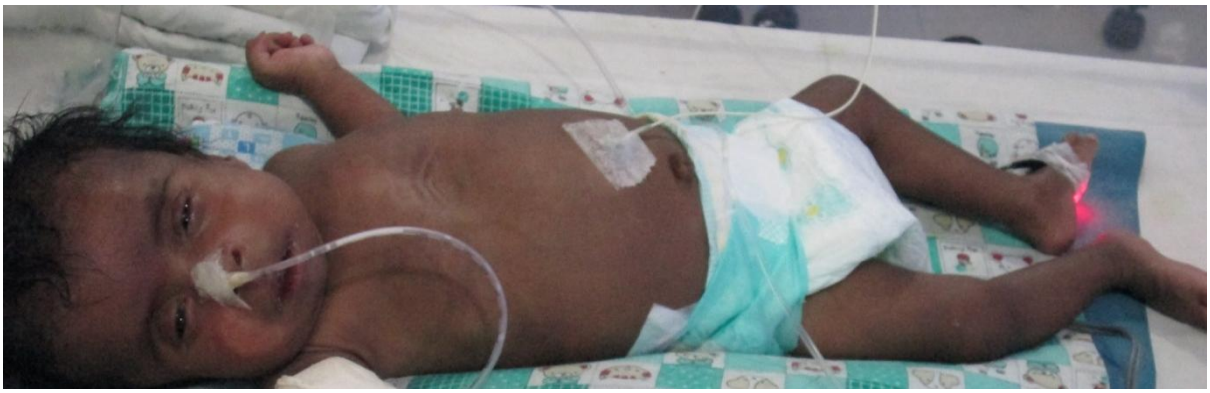

**FIGURE 1**


**FIGURE 2**

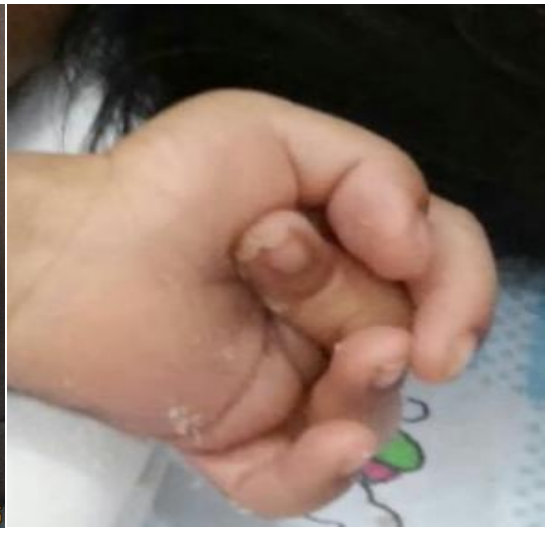

**FIGURE 3**

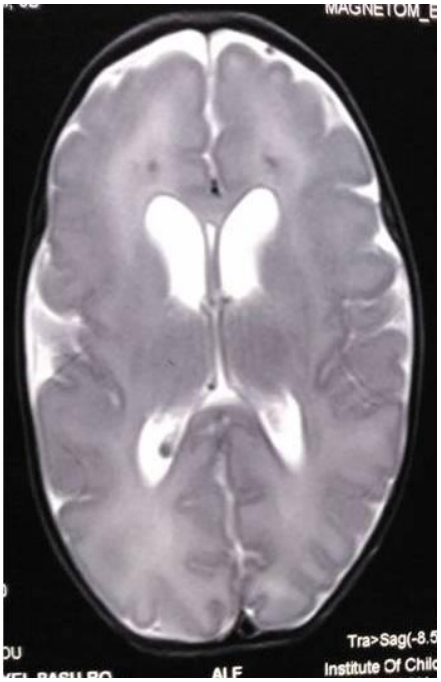


**FIGURE 4**

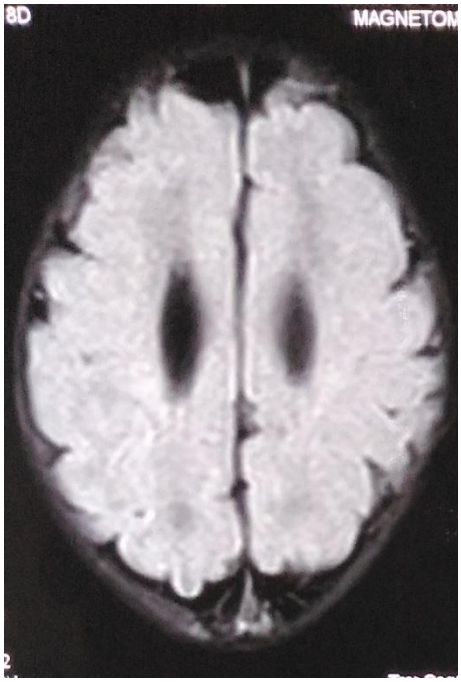


**FIGURE 5**

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