

A Rare Association of Monosomy 18 with Translocation 13p 11/18 with Cholelithiasis

Abstract

Monosomy 18p is a rare chromosomal disorder in which all or part of the short arm (p) of chromosome 18 is deleted (monosomic). The disorder is typically characterized by short stature, variable degrees of mental retardation, speech delays, malformations of the skull and facial (craniofacial) region, and/or additional physical abnormalities. We report monosomy 18 with translocation 13p 11/18 with Cholelithiasis in a 8 year old girl who presented with acute right hypochondriac pain with fever. This association has never been reported in this age group. This case report highlights this rare association which a clinician should keep in mind while dealing with this disorder.

Keywords: Monosomy; Dysmorphic; Cholelithiasis; Microcephaly; Holoprosencephaly

Case Report

Volume 4 Issue 4 - 2016

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Received: March 6, 2016 | **Published:** May 05, 2016

Introduction

Monosomy 18p is a rare chromosomal disorder in which all or part of the short arm (p) of chromosome 18 is deleted. It has been first described by the French geneticist Jean de Grouchy in 1963 [1]. This disorder includes short stature, mental retardation, speech delays, malformations of the skull and facial (craniofacial) region, and/or additional physical abnormalities like unusually small head (microcephaly), a broad flat nose, large protruding ears, widely spaced eyes and in some case with holoprosencephaly. We report monosomy 18 with translocation 13p 11/18 with Cholelithiasis in a 8 year old girl who presented with acute right hypochondriac pain with fever. This association has never been reported in this age group. This case report highlights this rare association which a clinician should keep in mind while dealing with this disorder and it increases the horizon of symptom complex.

Case Details

A 8 year old girl born to non consanguineous marriage reported to the outpatient department (OPD) with history of acute pain abdomen of 3 days duration, speech delay, global developmental delay and squint since birth. She was born at 37 weeks of gestation by emergency Lower segment caesarean section LSCS (Indication fetal distress) with birth weight 2.25 kg, length 45cm and head circumference 34cm. The baby cried immediately after birth and was started on breast feeds within 4hrs of birth. There were no antenatal risk factors. The initial milestones was delayed (cognitive more than motor). There was history of recurrent lower respiratory tract infection since 2yrs of age which required frequent hospital admission and antibiotics. She developed seizure at 5yrs of age which was managed with anticonvulsant. The MRI (Brain) done at this stage revealed small foci of hyperintensity in T2 weighted image in frontal and parietal region. The EEG was normal. There was no family history of

any chromosomal disorder. She was the eldest sibling and other siblings were 3 and 5 years old normal for age. Presently she was brought with history of acute abdomen pain on right upper abdomen which was colicky associated with fever with chills and vomiting. The clinical evaluation revealed weight 21kg (5th P) and height 120cm (3rd P) and head circumference 51cm. The Dysmorphic features were doliocephalic head, hypertelorism, Squint, flat bridge of nose, high arched palate, wide spaced nipple, prominent forehead and microcornea on right side and everted umbilicus (Figure 1A & 1B). The vital parameter revealed temperature 102 degree Fahrenheit with pulse 110/min, respiratory rate 30/min, blood pressure 110/70 mm Hg. The abdominal examination revealed marked tenderness over right hypochondriac region and other system was essentially normal. She was immediately admitted and was started on IV fluids and antibiotics and sample was sent for complete blood count, biochemistry and was planned for sonography of abdomen. The investigation revealed polymorpho leucocytosis with normal biochemistry with positive CRP. The thyroid profile was normal. The ultrasonography revealed multiple Gall bladder calculi with largest measuring 18mmx 19mm (Figure 2). The gall bladder was distended and Common bile duct (CBD) was normal. The lipid profile was normal. The Chest radiograph was normal. She was managed with antibiotics and IV fluids and she responded well to conservative management. The Karyotyping (Figure 3) was done in view of dysmorphism and developmental delay which revealed monosomy 18 with translocation 13p 11/18 which was later confirmed by FISH. The hearing assessment test was normal. The Eye evaluation revealed concomitant squint. The Intelligent quotient test revealed score of 60. She was planned for speech therapy and was kept under follow up.

Discussion

Deletion of the short arm of chromosome 18, del (18p), is now a well established chromosomal aberration. The female to

male ratio is 3:2 with incidence around 1 in 40000 live birth. The most frequent abnormalities consist of mild to moderate growth deficiency, mental retardation, microcephaly, ptosis, epicanthal folds, low nasal bridge, hypertelorism and large protruding ears. Holoprosencephaly and clinodactyly of the fifth finger is observed in about 10% and 20% of the cases respectively. Recent evidences have suggested an association with growth hormone deficiency that responded well to hormone supplementation [2]. Mental retardation is mild to severe with an average intellectual quotient (IQ) between 45 and 50. There is a significant discrepancy between verbal and non-verbal performance, verbal performance being more severely affected [2-4]. Most cases of deletion 18p are supposed to originate from de novo deletions, which accounts for 85% of cases [5]. The disorder is usually diagnosed or confirmed after birth (postnatally) based upon a thorough clinical evaluation, detection of characteristic physical findings, and chromosomal analysis. The treatment of Monosomy 18p is multidisciplinary including pediatrician, neurologist, speech therapist and pathologist. The specific surgical procedures performed will depend upon the severity and location of the anatomical abnormalities, their associated symptoms, and other factors. Early intervention may be important in ensuring that affected children reach their potential. Genetic counseling will also be of benefit for affected individuals and their families.



Figure 1A

Figure 1B

Figure 1: 1A AND 1B Dysmorphic features showing doliocephalic head, hypertelorism, Squint, flat bridge of nose, high arched palate, prominent forehead and microcornea.



Figure 2: Ultrasonography of abdomen showing gall bladder stone with post acoustic shadow.

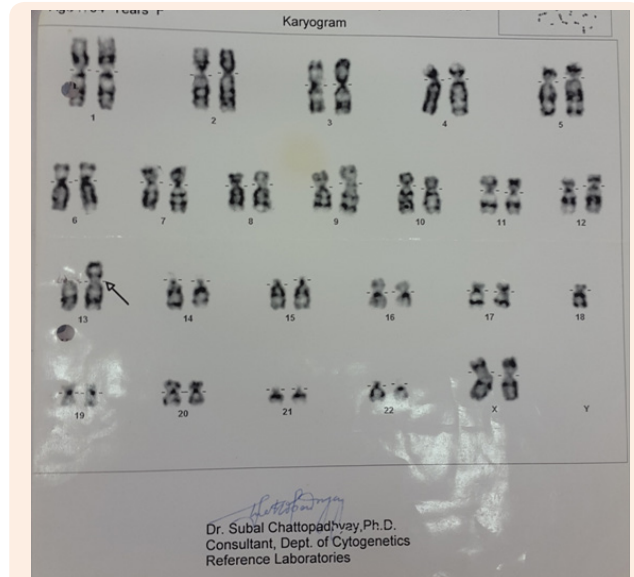


Figure 3: Karyotype showing monosomy 18 with translocation 13p 11/18.

In our case the association of Cholelithiasis with monosomy 18 with translocation 13p 11/18 has never been reported in the literature in this age group. The exact cause of cholelithiasis with this disorder requires research and further analysis. This case report highlights this rare association which a clinician should keep in mind while dealing with this disorder.

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