A case of Hematometra with a chromosomal abnormality: A case report

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Srilekha A, Sujatha M, Pratibha N, Jyothy A, Venkateshwari A. A Case of Hematometra with a Chromosomal abnormality: A case report. J of Genet Disord and Genet Med 2019;3(1):1-2.

We report a rare case of a 15-yr-old girl presenting primary amenorrhea and acute abdominal pain due to retention of menstrual blood in the uterus. Ultrasonography of pelvis and abdomen of the pro-band showed enlarged uterus with normal left ovary and rudimentary right ovary. The cytogenetic analysis revealed an autosomal translocation between chromosomes 3 and 18 with [46, XX t(3;18) (p 14;q23)] karyotype. To the best of our knowledge, there are no such reports diagnosed with hematometra showing chromosomal translocations till date.

Key Words: *Autosomal translocation; Counselling; Hematometra; Karyotype; Mullerian anomalies*

INTRODUCTION

Hematometra denotes the retention of menstrual blood in the uterine cavity causing acute abdominal pain. Usually, girls of reproductive age with hematometra have obstruction of the female genital outflow tract. The commonest of these cases is hematocolpos or secondary hematocolpometra resulting from congenital malformation like imperforate hymen [1].

Diagnosis is generally made in childhood, or at menarche age with the advent of cyclic pelvic, abdominal pain and primary amenorrhea. Rarely they may have urinary retention, recurrent urinary tract infections and back pain. Complete cervicovaginal agenesis with a functioning endometrium in a bicornuate uterus is an extremely rare mullerian duct malformation.

The palpation on the rectal or vaginal region may reveal a suprapubic mass. Initially, the pain is relieved by oral analgesics, and later if there is persistent pain, a diagnostic laparoscopy will be performed. About one out of 4,000-10,000 cases of hematometra show mullerian anomalies [2]. The occurrence of congenital hematometra with an enlarged uterus is a rare mullerian anomaly which results in rudimentary horn with functioning endometrium.

The general consequences of treatment of these patients are to remove the mullerian structures during the initial operation to avoid post-operative complications. Our case highlights the mullerian duct anomalies with a chromosomal abnormality involving a balanced translocation between chromosomes 3 and 18.

CASE STUDY

The proband is a 15 year old, third born, to a healthy consanguineous couple. There is no history of congenital anomalies or intellectual disabilities in the family, and the pregnancy was remarkable. She was referred to our institute with a complaint of Primary amenorrhea. The clinical examination showed that she was 152 cm in height and weighed 43 kg. She had been experiencing a cyclical abdominal pain for every 15 days for the past one year. She had no history of withdrawal bleeding on taking hormonal pills.

She has poor breast development with the presence of axillary and pubic hair. All routine examinations revealed no obvious abnormalities [TSH, Total bilirubin, Serum creatinine, random blood sugar, blood urea]. Diagnostic laparoscopy revealed an enlarged uterus with signs of hematometra. The left ovary was normal in size, and the right ovary was rudimentary. Ultrasonography of pelvis and abdomen showed hematometra with the right rudimentary horn of uterus and a bilateral mild hydro ureter. A lower 1/3rd of the vagina is present.

Cytogenetic analysis was performed on routine peripheral blood lymphocyte cultures according to the modified method of Moorhead et al. (1960), followed by standard GTG-banding [3,4]. A minimum of 25 metaphases were analyzed by using an Olympus microscope and GenAsi imaging software with a 550 band resolution. Chromosomal nomenclature followed as per ISCN (2016) guidelines for the karyotype analysis [5]. The study revealed a balanced autosomal translocation of chromosomes 3 and 18 with [46, XX, t (3;18)(p14;q23)] karyotype (Figure 1).

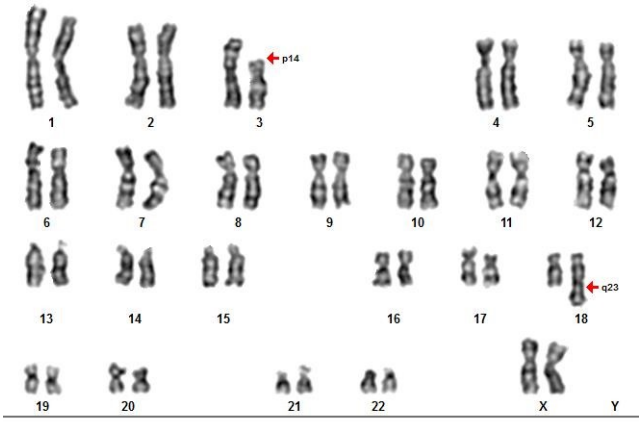

Figure 1) Karyotype of the proband showing the breakpoints on chromosomes 3 and 18

The breakpoint was at band 14 in the short arm of chromosomes 3 and band 23 in the long arm of chromosome 18.

DISCUSSION

Reporting autosomal translocations leading to primary amenorrhea is rare. The present case represents mullerian agenesis, which is among the differential diagnosis of cyclical abdominal pain that responds poorly to

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Received date: September 19, 2019; Accepted date: September 30, 2019; Published date: October 07, 2019

analgesics. The translocation of chromosomes 3 and 18 is the first report with hematometra leading to primary amenorrhea. The breakpoints included are chromosome 3p14 and 18q23. 3p14.2, which are fragile proximal region of chromosomes reported in many cases with deletion of the region leading to abnormal digestive tract cancers, intellectual disabilities and delayed milestones [6,7].

3p14.1 contains the FOXP1 gene that plays a significant role in neurological and language deficits [8]. In literature, up to date, a specific clinical phenotype has not yet been delineated for short stature, failure to thrive, facial dysmorphism, congenital heart disease, urogenital abnormalities, neurological problems, hearing loss, and global developmental delay, thereby suggesting that 3p14 deletion gives rise to congenital anomaly syndromes [9].3p14.2 has a common fragile site (3p14.2, contains the common fragile site, FRA3B, a hereditary renal carcinoma-associated 3;8 translocation and the candidate tumour suppressor gene, FHIT). The region3p14.2-p14.1 also contains genes FEZF2, CADPS, SYNPR, ATXN7, PRICKLE, and MAGI1 which are known to have a role in neurodevelopment. Patients with developmental disorders, autistic features, and/or global developmental delay showed several microdeletions and microduplications mapped to 3p [10].

A deletion on 18q23 causes various abnormalities such as growth retardation, hearing loss, and mental retardation. A case report, reported by Strathdee et al. (1997) on mental retardation could correlate the 18q23 deletion with hearing loss and midfacial hypoplasia and limb abnormalities [11]. A fetus with multiple congenital anomalies, atypical lyssencephaly, corpus callosum agenesis, cerebellar hypoplasia, cleft palate, ventricular septal defects, and hypoplastic aortic arch inherited with 18q23 duplication [12]. In the present case, we observed the mullerian duct anomalies such as enlarged uterus with rudimentary right ovary showing a balanced autosomal translocation of chromosomes 3 and 18 with [46, XX, t(3;18)(p14;q23)] karyotype.

CONCLUSION

As there is a significant role of chromosomal anomalies leading to primary amenorrhea, females encountering blocked uterus should follow a cytogenetic evaluation by the age of 15. The surgery can be selective by removing the mullerian structures during initial operation to avoid other gynecological and obstetrical complications. Although technical advances favor reconstructive surgery for cervicovaginal agenesis, it must be remembered to analyze cytogenetic abnormalities at the earliest.

ACKNOWLEDGEMENTS

Authors would like to thanks the staff of cell biology for the technical assistance.

ETHICAL PUBLICATION STATEMENT

We confirm that we have read the journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

CONFLICTS OF INTEREST

None of the authors has any conflict of interest to disclose.

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