(EP07.05) A case of double trisomy (T21 and T18)
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Introduction

- It is estimated that as much as 60% of first trimester pregnancy losses are chromosomally abnormal and that single trisomies comprise the majority of these.
- In comparison, the occurrence of double and triple aneuploidy in the same individual is a relatively rare phenomenon, and estimated to occur in only 0.21 to 2.8% and 0.7% respectively of karyotyped spontaneous abortions.
- Pregnancy losses with double and triple trisomies are reported to occur at an earlier gestational age and at an older maternal age than single trisomic losses (mean gestational age for pregnancy losses with a single trisomy was 12.3 weeks, double and triple trisomy pregnancies were lost at an earlier gestational age, 8.6 weeks; the mean maternal age for the double and triple trisomies was 38.8, in comparison to the single trisomies in which mean maternal age was 37.3 years).
- Various combinations of double trisomy in more than 50 cases have been described in the literature. The common double trisomies were the combinations of 48,XXY, +21 and 48,XXX,+18; 20 and 16 cases were reported, respectively.
- There was only one case report published by Grosse and Schwanzit regarding a live born female with double trisomy of the autosomes 18 and 21 (48,XX,+18,+21), exhibiting the clinical features of mongolism.
- We describe a case of double trisomy of the autosomes 18 and 21 (48,XX,+18,+21) noted from the cytogenetic studies of products of conception.

Case

A 42 year old lady G1P0 was referred to the fetal medicine unit at approximately 11 weeks gestation with cystic hygroma. She had non-invasive prenatal test (NIPT) which showed high risk for T13 and T18. Unfortunately, she had spontaneous miscarriage at 11+3/40, and subsequently had evacuation of retained products of conception. The cytogenetic studies of the products of conceptions showed 48,XX,+18,+21.

Discussion

- Some cases of double trisomies involving chromosomes 8, 13, 18, 21, X and Y have been observed in liveborns, suggesting that lethality of the abnormality depends on which chromosomes are involved in the aneuploidy.
- Despite the fact that maternal meiosis I non-disjunction seems to be the major cause of the whole single trisomy cases, chromosome-specific patterns do exist and a possible mitotic origin must also be considered.
- Little is known about biological mechanisms underlying double trisomy. Cytogenetic analysis Molecular techniques could be useful in diagnosing not only single but multiple aneuploidy and determining its origin. This will improve our knowledge about mechanisms underlying human aneuploidy, and enable appropriate genetic counselling.

Conclusion

- Recurrence risk following a single trisomic pregnancy is approximately 1%. Currently, the recurrence risk following double trisomy is unknown.
- Case control studies are needed to determine recurrence risk following double or triple trisomic pregnancies.
- A significantly greater recurrence risk than that for single trisomy would have implications for genetic counselling.

References