Carcinogenesis in a Congenital Deformed Fetus - A Case Report

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Abstract: USG report of a 27+3 week old female fetus in an 18 year old primi, who had undergone CT scan during her first trimester showed a Meromelia fetus. Cytogenetic studies from the cord blood cells of the medically terminated fetus revealed 48% aberrant metaphases. The congenital deformity of the fetus could have occurred due to effects of ionizing radiation. But even more interesting were the aberrant chromosome which are the indicators of the future fatal childhood cancer that would have gone unnoticed without the chromosomal studies. The case is reported to stress the need for prenatal cytogenetic studies in an event of CT of a fetus, done either knowingly or unknowingly during the first trimester.

Keywords: CT scan, Meromelia, cytogenetics, aberrant chromosomes, prenatal diagnosis

1. Introduction

Organogenesis occurs predominantly between second and fifteen weeks gestation, a period when the fetus is most susceptible to the teratogenic and carcinogenic effects of ionizing radiation. CT at dose of >10 mSv (500 times a routine chest X-ray) during first trimester may result in birth defects and future fatal childhood cancer [1, 2]. The present study report one such combined findings in a medically terminated deformed female fetus which may provide a positive contribution to the understanding of the importance of prenatal cytogenetic study on first trimester CT scan exposed fetuses and also to help to develop a more effective measures of patient safety in future.

2. Case Presentation

An eighteen year old primi met with a bike accident injuring her hip. Unknowing of her 5+week pregnancy, she underwent an emergency pelvic CT at a dose of approximately 5mSv, as an accurate diagnosis was at stake. During the antenatal checkup she was advised for a medical termination of the pregnancy after the USG report of her 27+3 week old female fetus showed Meromelia. Cytogenetic study was done using heparinized umbilical cord blood cells following standard Hungerford method [3]. Cytogenetic studies of the fetus revealed 48% aberrant metaphases with polyploidy, chromosomal breaks, fragments and gaps

3. Discussion

Meromelia is a birth defect of limbs caused by genetic or teratogenic factors. The present condition of the fetus with deformed extremity of the hands, could be due to effects of ionizing radiation [4]. Published reports support an association between in utero irradiation and the increased risk of childhood cancer, yet the data still remain controversial [5]. In general, Stochastic or Nonstochastic, in utero irradiation results in cellular or multicellular injury at the DNA and chromosome level leading to cancer [2]. The present interesting cytogenetic findings of the meromelia fetus are the aberrant chromosomes, which are the biomarkers for carcinogenesis, clearly indicate the future fatal childhood cancer that would have gone unnoticed without the chromosomal studies.

Figure 1: 27+3 week old Meromelia fetus
4. Conclusion

Thus, in an event of CT of a fetus, done during the first trimester either knowingly or unknowingly, prenatal diagnosis and genetic counseling should be made mandatory. Such a safety measure would provide an eager mother to have her unborn child to be born anatomically and genetically a normal individual.

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References


Authors Profile

A.N.Uma received her M.Sc and M.Phil in Genetics from Dr.ARML PGIBMS, Madras University in 1992 and 1993 respectively. Has also received B.A and M.A in English from Annamalai University and B.Ed in Science and English from IGNOU University. In 2006 she joined Genetic Lab, Mahatma Gandhi Medical College & Research Institute, Sri Balaji Vidyapeeth, Puducherry. She has is a faculty and helps the clinicians in the clinical diagnosis of patients pertaining to genetic defects. She has completed her Ph.D thesis in Oral Cancer from the Department of Pathology.

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