Case Report Of 48 XXYY Syndrome Associated To Father’s Radioactive Contamination During The Cesium Accident in Goiânia - Goiás, Brazil.

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INTRODUCTION

48,XXYY Syndrome occurs in 1:20,000–1:50,000 male births. It used to be considered as a variant of Klinefelter syndrome, but now it is considered as a distinct clinical and genetic entity with increased risks for congenital malformations, additional medical problems and more complex psychological and neurodevelopmental involvement.

48,XXYY Syndrome results from the fertilization of a normal female oocyte (Xm), with an aneuploid sperm (XpYpYp) produced through nondisjunction events in both meiosis I and meiosis II of spermatogenesis. Literature shows that 100% of the triploid gamete is from paternal origin.

OBJECTIVES

We report a case of 48 XXYY Syndrome, whose father was contaminated by radioactive Cesium 3 years before the proband conception.

Since radiation can induce abnormal chromosome segregation during mitotic division, we hypothesize that the father’s Cesium contamination might be responsible for this rare occurrence.

CASE REPORT

SFAD, male, second child of a non-consanguineous young couple.

At 12 years of age, he searched for genetic testing due to agenesis of cart palate and nasal septum. Karyotype = 48 XXYY.

CONCLUSIONS

Recognition of medical, developmental and psychological problems that are associated to 48 XXYY Syndrome is important for early diagnosis and interventions, as a way to best outcomes.

This is the first reported case of 48 XXYY associated to the Cesium Accident.

REFERENCES


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