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

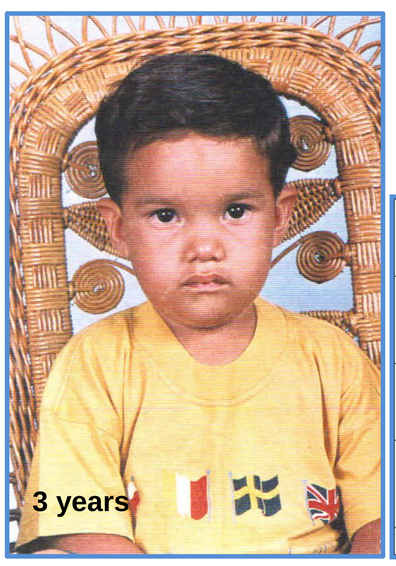
We report a case of 48 XXYY Syndrome, whose father was contaminated by radioactive Cesium 3 years before the proband conception.

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

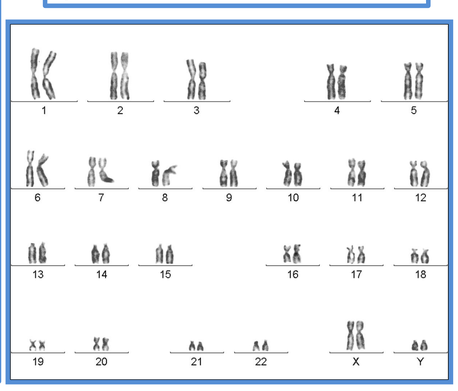
CASE REPORT

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

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



Typical facies:
ocular hypertelorism
epicanthal folds
prominent elbows



CASE REPORT

At 13 years of age, he was referred to the Pediatric Endocrinologist service, presenting with: tall stature (165 at 13 years, 201 final height), eunuchoid body habitus, *cubitus varus*, *pes-planus*, thoracic vertebrae fusion, bilateral femur-patellar arthrosis, bilateral inguinal hernia, hypergonadotrophic hypogonadism, single malformed kidney, mild intellectual disability, emotional immaturity, anxiety, impulsivity and obsessive-compulsive behaviors.

He evolved with osteoporosis, seizures (14y), hypertension, insulin resistance, obesity, dyslipidemia (18y), pre-diabetes (23y), testicular volume was of 5 ml as an adult, infertility due to azoospermy.

He was treated by a multidisciplinary team: pediatric endocrinologist, orthopedist, neurologist, cardiologist, nephrologist, psychologist, fono-audiologist and occupational therapist.

The family reported signals of cardiac insufficiency a week before a sudden death at age of 24 years. Necropsy showed bilateral pulmonary thromboembolism.

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.

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Syndromes : Mechanisms and Management

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