DISCUSSION

48,XXYY is a very rare sex chromosome abnormality (aneuploidy) characterized by the presence of one extra X and Y chromosomes in males. It is estimated to occur in 1:30000 – 1:50000 male births. Phenotypically it was considered a variant of Klinefelter syndrome (47,XXY), but currently, due to mental deficiency and behavioural characteristics associated, it's considered to be a separate genetic condition.

It is associated with neurodevelopmental and psychological involvement. 48,XXYY syndrome results from non-disjunction during gametogenesis. The extra set of chromosomes almost always comes from the sperm due to non-disjunction during meiosis.

Neurodevelopmental and psychological disorders are common with the presence 48,XXYY. Other features include dental defects, peripheral vascular disease, deep vein thrombosis, dysmorphic features, elbow abnormalities, as well as allergy and asthma. The risk of developing type 2 diabetes is also high.

It is accepted that XYY spermatogonia are able to produce altered 24,XY and 24,YY spermatozoa, although the ejaculate of XYY men have generally shown a very small increased fraction of them. In a small percentage of patients, mitotic non-disjunction occurring during the early stages of a 46,XY zygote can result in 48,XXYY syndrome.

This rare chromosome aneuploidy is exceptionally discovered during childhood because there are no specific modifications. Mostly cases are diagnosed after puberty because its phenotype similar to the Klinefelter's syndrome. It is important to differentiate it from Klinefelter's syndrome (46,XXY) because there are a variety of behaviours, learning disabilities and emotional problems that are unique to patients with XXYY syndrome.