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MOSAIC ISODICENTRIC Y CHROMOSOME AND 46, X,IDIC(Y) [23]/45,X[17]/46,XY KARYOTYPE: CASE REPORT

Maurício Assis Rodrigues
XY Diagnose Laboratório de Biotecnologia, Campos dos Goytacazes, RJ
http://lattes.cnpq.br/0523569270504213

Thiago Freitas Marotti
Faculdade de Medicina de Campos, Campos dos Goytacazes, RJ
http://lattes.cnpq.br/4119014743056731

Samara Barboza Gomes
Faculdade de Medicina de Campos, Campos dos Goytacazes, RJ
http://lattes.cnpq.br/1132189866585861

Bárbara Gama de Medeiros
Faculdade de Medicina de Campos, Campos dos Goytacazes, RJ
http://lattes.cnpq.br/2316338209282814

Maria Fernanda Vitória Pessanha Campos Azevedo
Faculdade de Medicina de Campos, Campos dos Goytacazes, RJ
http://lattes.cnpq.br/0046321139757460

Abstract: the isodicentric Y chromosome is characterized by a pair of arms identical, mirroring each other through positioning _ _ by the two centromeres, due to their presence are unstable generating a range of mosaicism patterns, with 45.X being the most common and is associated with several manifestations' clinics. Being able vary from a phenotype normal male, with or without fertility, even phenotype feminine with or without stigmas of Turner syndrome. The objective of this work is to report a case of a sex patient male with malformations genitourinary. The karyotype result _ showed a mosaicism atypical of three bloodlines cell phones: 46, X, idic(Y)[23]/45, X[17]/46,XY[10].
Keywords: Change chromosomal; Mosaicism chromosomal; Isodicentric Y chromosome.

INTRODUCTION

The isodicentric Y chromosomes they are often observed as one of the anomalies more common on the Y chromosome (DESGROSEILLIERS *et al.,* 2006), being it is abnormality identified firstly per Jacobs *et al.* in 1966 (JACOBS *et al.,* 1966). It is characteristic per present a pair of arms identical, mirroring each other through _ through positioning _ by the two centromeres (BERGERON *et al.,* 2011).

It is believed that the isodicentric Y chromosomes form _ through processes _ like recombination _ intrachromosomal or chromatid fusion _ _ sisters after breakage occurs _ _ chromosomal on the Y chromosome [PASANTES *et al.,* 2012]. This way, both the genetic material preserved of the Y chromosome as to the manifestation's phenotypic characteristics of carriers of this abnormality structural chromosomal, are dependent on the breaking point, making highly the genetic material of the isodicentric Y chromosome is variable. Thus, if the break point on the isodicentric Y chromosome is in the arm short (p), the arm proximal short, well as the whole arm long (q) will be duplicate. Likewise, if the breaking point occur in the arm long (q), the entire arm proximal long and arm short will suffer from duplication (BERGERON *et al.,* 2011).

Due to the presence of both centromeres, the isodicentric Y chromosomes they are unstable, generating like this a range of mosaic patterns, with 45.X being the most common (DESGROSEILLIERS *et al.,* 2006). Such standards they are associated with vast manifestations clinics like genitalia _ _ ambiguous, gonadal dysgenesis, turner syndrome and low height (TROMBETTA; CRUCIANI, 2017). However, it is known that the phenotype postnatal period varies widely between carriers of such change chromosomal (YANG; HAO, 2019). In addition addition, the patients without changes morphological, carriers of Y chromosome isodicentrism, many times make the diagnosis genetic late, due infertility complaints _ already at phase adult (KALANTARI *et al.*, 2014).

Therefore, the diagnosis precocious per through techniques _ molecular and cytogenetic becomes crucial for patients carriers of the isodicentric Y chromosome. The goal of this work, is to report the case of a recent born at term carrier of isodicentrism on the Y chromosome presenting 3 lineages cell phones, in addition to various changes morphological.

CASE REPORT

Sex patient _ _ male, born at term, with malformations genitourinary as hypospadias peno-scrotum, pouch bifida and bilateral cryptorchidism, in addition to incarcerated inguinal hernia. In this context, he was forwarded to the genetics service. The analysis cytogenetics of 50 cells (figures 1, 2, 3, 4 and 5) from culture of lymphocytes in blood peripheral heparinized revealed presence of

mosaicism chromosomal: 45, X [17]/46, X, idic(Y)[23]/46,XY[10]. In other words, the exam showed the presence of 3 lineages cells: in 23 cells (46%): 46, X, idic(Y); in 17 cells (34%): 45.X; in 10 cells (20%): 46, XY.

From the findings _ clinical and laboratory procedures, the conduct adopted was to subject the patient to a bilateral inguinal herniorrhaphy surgery and bilateral orchidopexy. During assessment surgery, it was noted that there was suffering in the testicle left (testicular torsion); testicle looking _ dysmorphic, with epididymis dissociated; cord spermatic thickened, with a tubular shaped cyst and content mucoid. After the interventions, the newborn was stable on the fourth day post-operative. The present study obeyed to the aspects ethical and legal related to research involving _ beings humans with approval by an Ethics Committee number CAAE 56679822.3.0000.5244. _

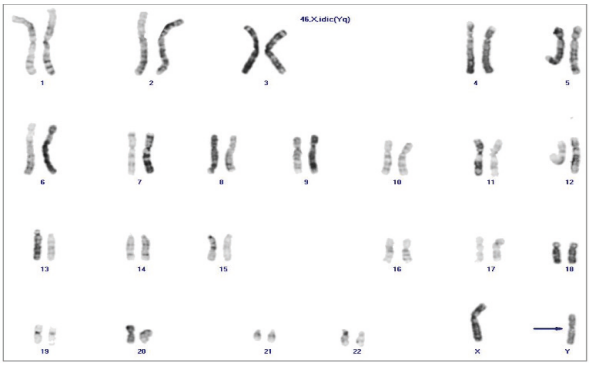

Figure 1: Image karyotype G of a cell 46, X, idic(Y).

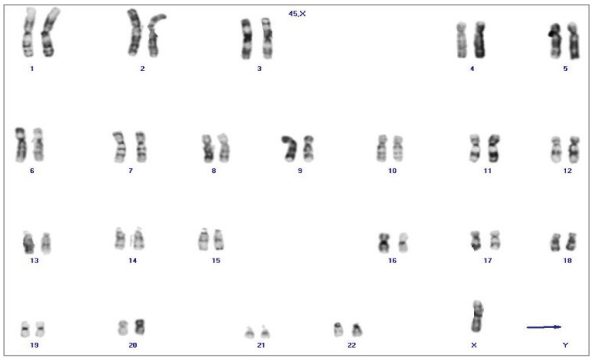

Figure 2: Image karyotype G of a cell 45, X.

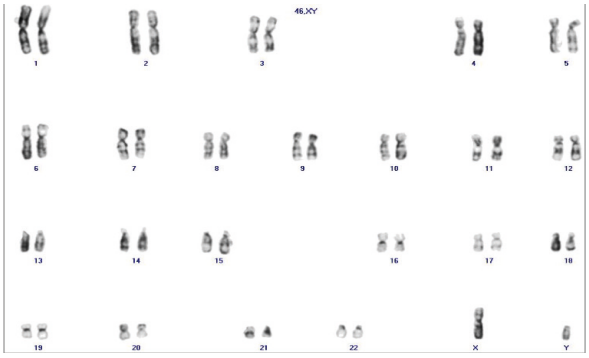

Figure 3: Image karyotype G of a cell 46, XY.

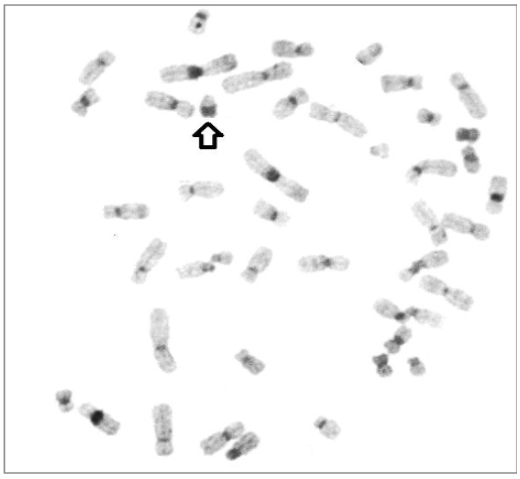

Figure 4: Image karyotype C bands of a cell 46, XY.

Y chromosome highlighted.

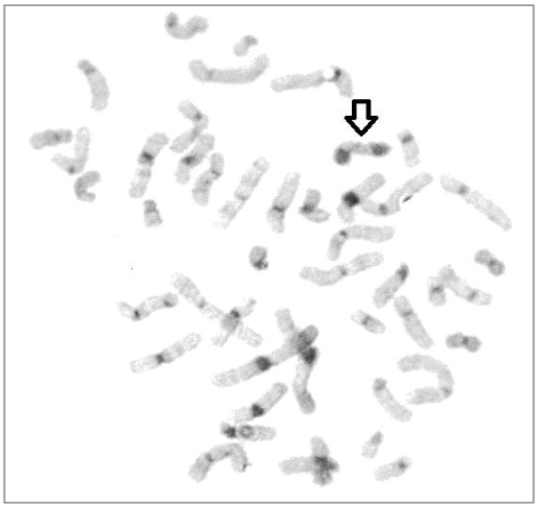

Figure 5: Image karyotype C bands of a cell 46,X,idic(Y).

Chromosome idic (Y) highlighted.

DISCUSSION AND CONCLUSION

Due to chromosome instability isodicentric during division, we have many different types of cells that are found at big most patients _ chromosome carriers _ isodicentric (DESGROSEILLIERS *et al.*, 2006). The Y chromosome has several genes involved at sex determination, spermatogenesis, growth and development. _ The phenotype of patients is variable, some have stigmata of Turner syndrome, azoospermia, hypospadias and testicles abnormal or small (YANG; HAO, 2019).

You several degrees of mosaicism in patients reflect at chromosome instability _ isodicentric and in its moment of stabilization during embryogenesis. _ In addition Furthermore, if we compare per example with the series of 9 cases of isodicentric Y chromosome by DESGROSEILLIERS *et al.*, 2006, we will see that only 1 of the 9 cases had the presence of 3 lineages genetics in blood samples _ peripheral, and even so, this case presented one percentage very small cell with the third lineage pattern _ cell (only 2 cells), getting difficult to know if such mosaic really influenced the phenotype of such patient. Thus, according to our study of the literature, our patient was the only one found a balance at percentage of the 3 lineages, demonstrating thus the rarity of such karyotype.

How does the chromosome isodicentric not have the SRY gene, individuals have phenotypes female and the SRY action is done through the production of androgens and _ ducts mullerians, inducing the differentiation of Wolffian ducts into vesicles seminal, epididymis, duct ejaculatory (SHINAWI *et al.*, 2010). In this case report, the patient it presents hypospadias peno-scrotum, pouch bifida and bilateral cryptorchidism, phenotypes connected directly the presence of the chromosome isodicentric and consequently the absence of the SRY gene, impairing development embryonic reported.

REFERENCES

BERGERON, Mélanie Beaulieu et al. Correlation of intercentromeric distance, mosaicism, and sexual phenotype: molecular localization of breakpoints in isodicentric Y chromosomes. **American Journal of Medical Genetics Part A**, v. 155, n. 11, p. 2705-2712, 2011.

DESGROSEILLIERS, M. et al. Phenotypic variability in isodicentric Y patients: study of nine cases. **Clinical genetics**, v. 70, n. 2, p. 145-150, 2006.

JACOBS, Patricia A.; ROSS, Alan. Structural abnormalities of the Y chromosome in man. **Nature**, v. 210, p. 352-354, 1966.

KALANTARI, Hamid et al. Delineating the association between isodicentric chromosome Y and infertility: a retrospective study. **Fertility and Sterility**, v. 101, n. 4, p. 1091-1096, 2014.

PASANTES, J. J. et al. 47, X, idic (Y), inv dup (Y): a non-mosaic case of a phenotypically normal boy with two different Y isochromosomes and neocentromere formation. **Cytogenetic and genome research**, v. 136, n. 2, p. 157-162, 2012.

TROMBETTA, Beniamino; CRUCIANI, Fulvio. Y chromosome palindromes and gene conversion. **Human genetics**, v. 136, n. 5, p. 605-619, 2017.

SHINAWI, Marwan et al. Mixed gonadal dysgenesis in a child with isodicentric Y chromosome: Does the relative proportion of the 45, X line really matter?. **American Journal of Medical Genetics Part A**, v. 152, n. 7, p. 1832-1837, 2010.

YANG, Yang; HAO, Wang. Clinical, cytogenetic, and molecular findings of isodicentric Y chromosomes. **Molecular Cytogenetics**, v. 12, p. 1-9, 2019.