Abstract citation ID: bvac150.1292

Pediatric Endocrinology

PMON311

45,X/46,XY mixed gonadal dysgenesis: a case report from Saudi Arabia

Abdullah Aljasser, MD and Norah Alsaleh, MD

Introduction: Mixed gonadal dysgenesis is a sex developmental disorder where the gonads are abnormal from there being some cells with XY chromosomes and some with just asingle X, known as chromosome Y mosaicism. This results in a wide range of male/female genitalia that are not typically, or clearly, male or female

The case: 3 years old Saudi boy was born term to 35 year old G5P5 Normal anti natal and history of consanguinity. Birth weight 2 kilogram and found to have genital ambiguity in the form of short phallus structure and hypospiadius testis were felt at the right inguinal canal. his investigation showed abnormal chromosomes 45×46XY musiac . Mother chromosomes were 46XX normal female karyotype.

normal siblings and Wes WES: CNV DESCRIPTION*: seq[GRCh37] Yp11.31q11.23 (mosaic~50% of the cells) SIZE (KB):

~26Mb, GENE COUNT**: 116, INTERPRETATION***: Pathogenic.

RELATED DISORDER: 45,X/46,XY mixed gonadal dysgenesis

The radiological investigations showed small oval shaped mass over the right inguinal canal 0.8/0.6/0.8 cm in size .the left testis couldn't be seen . And Genetogram showed presence of vagina and small uterus.

his laboratory investigations showed at age of 2.5 years LH: less than o.3 and his Testosterone basal was less than 0.09 and Testosterone post HCG 8.29 which should excellent response to HCG.. the case was discussed with the paediatric urologist and the decision was as per the anatomical finding and results of the stimulated Testosterone to give Testosterone injections and asses the response. The child respond very well to the testosterone treatment which leads to improvement in the size and length of the phallus.

Discussion: mixed gonadal dysgenesis is one of the causes of DSD .45, X/ 46, XY mosaic: At birth around 90 per cent of babies with 45,X/46,XY have normal-appearing external male genitals, five per cent have normal-appearing female genitals and in five percent the external reproductive organs are neither fully male nor fully female. They may find one or two gonads that are very inadequate testes, possibly with some ovary-like tubules present. They may find one streak gonad and one immature, dysgenetic or normal testis. They may find an internal vagina, cervix and uterus, although these are usually quite small compared to a normal female. Some people with a 45,X/46,XY karyotype have reduced fertility and some are infertile. No judgment can be made about fertility until well after puberty in the apparently normal males. Children with a 45,X/46,XY karyotype can have any of the medical conditions that are more common among girls with Turner syndrome (who have a 45, X karyotype), which includes heart and disorders, autoimmune disorders and short stature.

Presentation: Monday, June 13, 2022 12:30 p.m. - 2:30 p.m.