A Child with Mos 45,X/46,XY and Gonadal Dysgenesis

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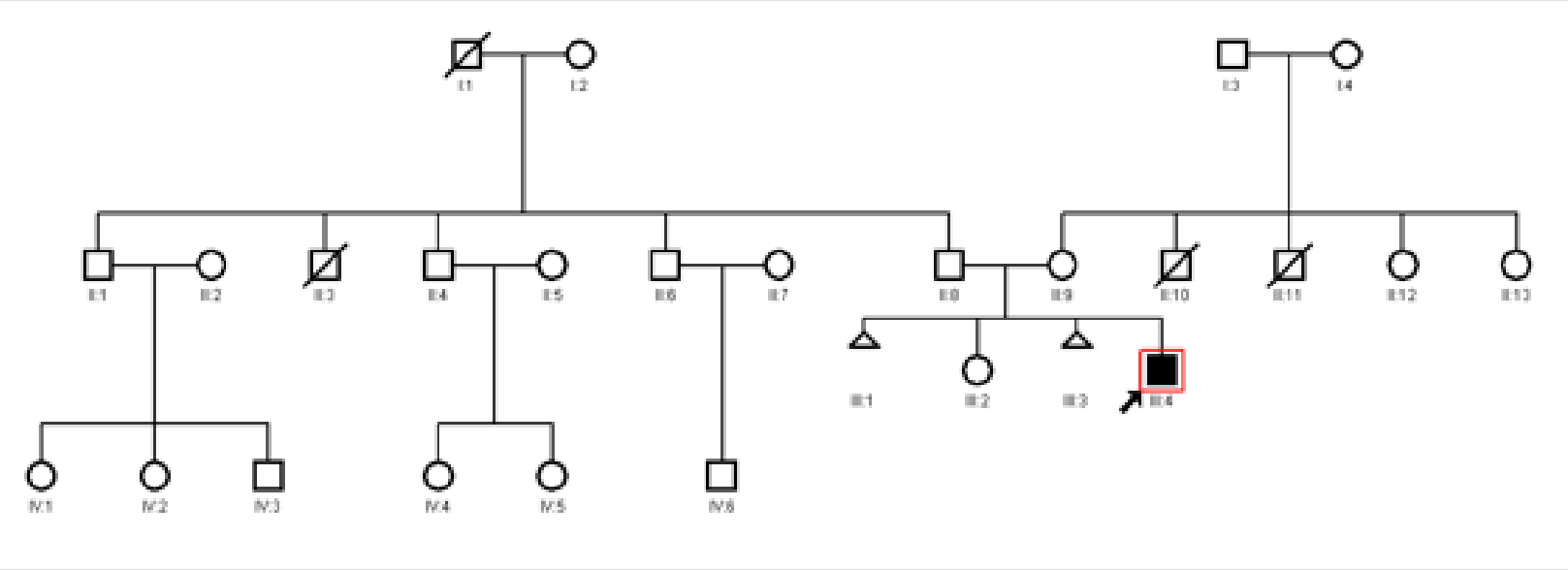
PRESENTED IN CASE SERIES WEBINAR
INDONESIAN SOCIETY OF GENETIC COUNSELOR
SATURDAY, NOVEMBER 27, 2021

History

- A boy was referred by an urologist with severe hypospadias.
- Age of admission 6 months 25 days
- Mother's age: 33 y.o (G4P2A2);
  - 1st miscarriege : Blighted ovum
  - 2nd miscarriage : spontaneus abortion on age 8 week of gestation
- Antenatal history :
  - ANC with Obgyn, No chemical exposure, no medication
- Normal delivery by midwife, aterm
- BW: 3400 gr ; BL: 49 cm

Pedigree



- No consanguinity
- No other family member with the same syndrome

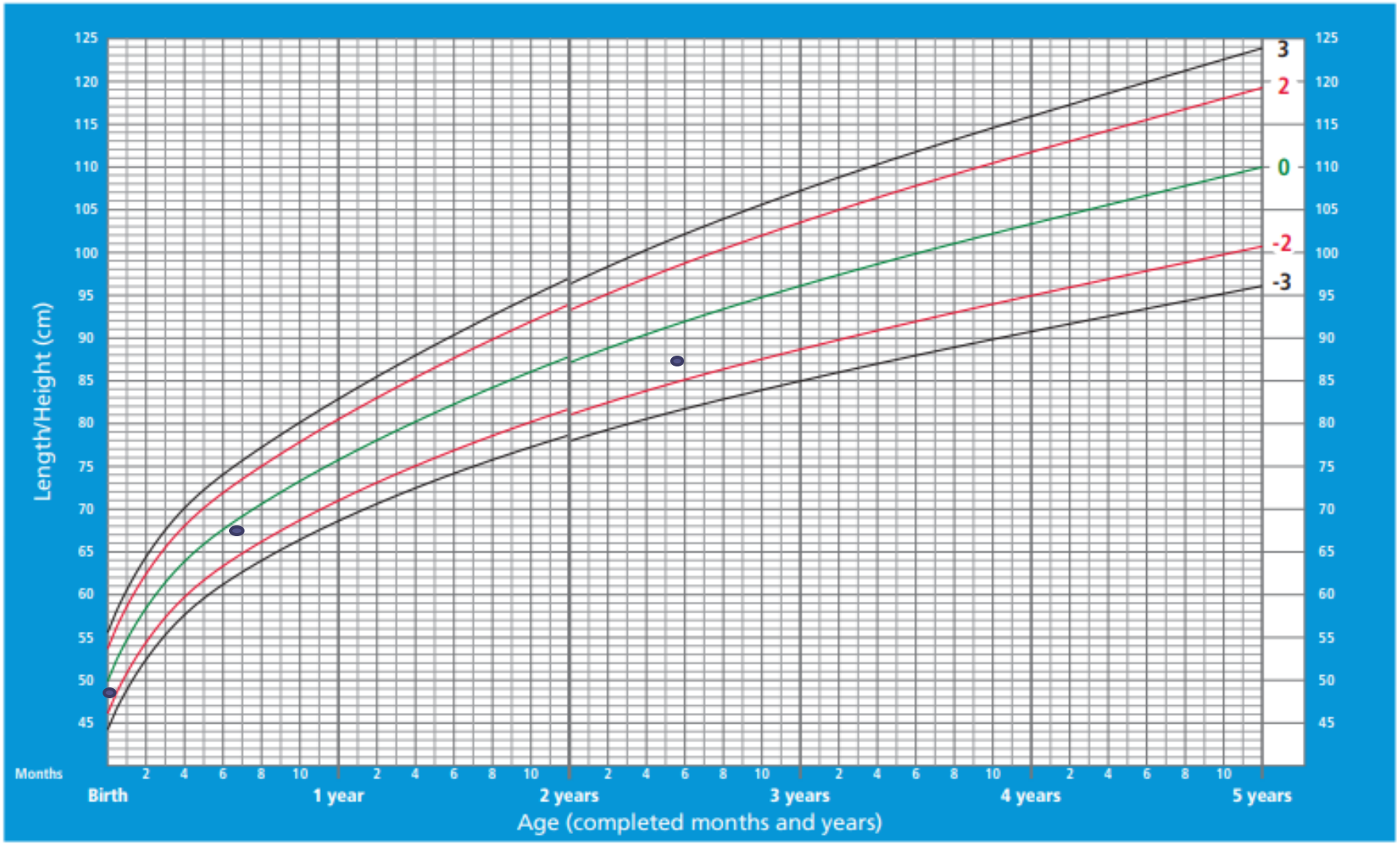
Physical Examination

- Physical examination on age 6 months 25 days
  - Weight: 7,5 kg; Lenght : 67 cm –HAZ= >-2 ; Short stature : (-)
  - Head : mesocephalus
  - Neck :Extraskin of neck(+)minimal; Low posterior hair line (+)
  - Thorax : shield chest (-), cardiac murmur: (-) and pulmonal abnormality (-)
  - Abdomen : abdominal mass (-)
  - Extremities :
    - Tappering finger (+/+), clinodactily of 5th finger (+/+)
    - Cubitus valgus (-)
  - External Genital examination:
    - Phallus length: 2,5 cm
    - Scrotum : bifida (+), hyperpigmentation (-), rugae (+), chordae (+)
    - OUE : phenoscrotal
    - Perineum: one ending
    - Gonad: R: 1 mL /scrotal ; L: <1 mL/retractile (streak gonad?)
- Age 2 years 5 months :
  - Weight: 11 kg; Height : 87 cm

Length/height-for-age BOYS

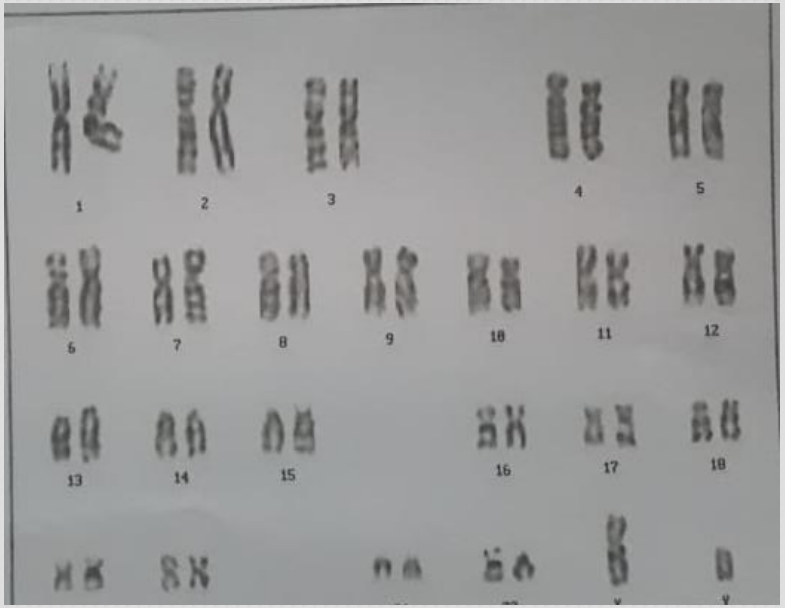
Birth to 5 years (z-scores)

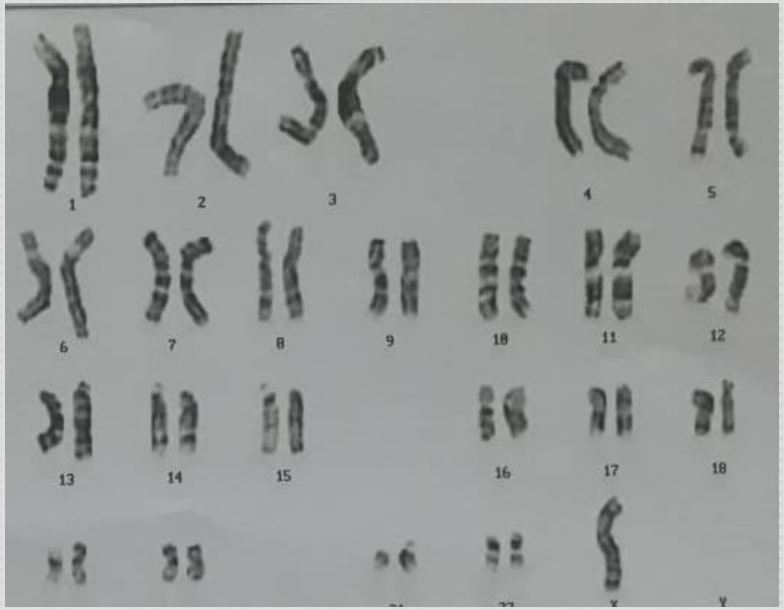
World Health Organization



WHO Child Growth Standards

Karyotipe 45,X[12]/46,XY [88]





ISCN 2016

Laboratory findings

- Hormonal data : 14th January 2020
  - LH : 0,33 mIU/mL (L)
  - FSH : 0,6 mIU.mL (L)
  - Testosterone : 8, 12 ng/dL (N)
  - Testosterone post HCG : 2, 46 ng/dL
  - HCG test (-)
- Ultrasound :
  - Uterus (-)
  - Adnexa (not mentioned)
  - Testicular structure (not mentioned)
  - Horse shoe kidney (-)

Diagnosis

- Mos 45,X [12]/46,XY[88] with mixed gonadal dysgenesis

Management

- Ultrasound of left testicular structure to identify the presence of streak gonad
- HCG test by andrologist to know gonadal function
- Genital correction by Urologist planning (after gender adjustment team meeting):
  - Orchidopexy or orchidectomy for the left gonad
  - Chordectomy
  - Hypospadia correction

Genetic Counseling

- Inform to the parents of the patient regarding :
  - The sex chromosomal abnormality caused disorder of sexual development in this patient
  - The number of karyotipe 46,XY>45,X defined as male, but possible to have gender disphoria in the future
  - To confirm the gender and help sex assignment need: SRY gene and psychological analysis
  - Mostly de novo with recurrent risk <1%
  - Possibility to have short stature and gynecomastia in the future
  - Possibility to have reproductive disorder and gonadoblastoma due to the unfunctional gonads

Disscussion

- It is still in debate whether this patient can be included as turner variant due to dysmorphological appearance was not typicaly for Turner syndrome

Thank you

Hindawi
Case Reports in Obstetrics and Gynecology
Volume 2019, Article ID 3719178, 3 pages
https://doi.org/10.1155/2019/3719178

Case Report

Mosaic Turner Syndrome Presenting with a 46,XY Karyotype

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Received 16 December 2018; Revised 17 March 2019; Accepted 18 March 2019; Published 11 April 2019

Academic Editor: Seung-Yup Ku

Although Turner syndrome is most commonly associated with a 45,X genotype, other mosaic genotypes are present in approximately half of all cases. We describe a case of Turner syndrome with a 46,XY genotype by conventional 5-cell karyotype who was subsequently found to have a mosaic genotype of 18% 45,X and 82% 46,XY by 50-cell FISH analysis. Individuals with a mosaic 45,X/46,XY genotype have a variety of phenotypic presentations ranging from male to female which are not correlated with the percentage of mosaicism. Our case represents an extreme example where the genotype is predominately 46,XY and the phenotype typical of Turner syndrome.

Mosaic Turner Syndrome With 45,X/46,XY Mosaicism and Apparent Absent Uterus

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Abstract

Turner syndrome (TS) is a relatively common chromosomal abnormality in females. Short stature, gonadal dysgenesis, and somatic dysmorphic features are the characteristic features of the syndrome. The chromosomal abnormalities of TS are highly variable; 45,X/46,XY mosaicism accounts for 10-12% of cases of Turner syndrome. Despite the presence of hypogonadism, affected females typically have a uterus. Here, we report the case of a 22-year-old female who presented at 15 years of age with primary amenorrhea. She was diagnosed with Turner syndrome mosaicism with a karyotype of 45,X/46,XY. Her pelvic imaging showed an absent uterus and ovaries. Due to the presence of a Y chromosome, she underwent prophylactic gonadectomy. Histopathology of her removed gonads confirmed the diagnosis of mixed gonadal disorder. She was started on estrogen replacement. Four years after treatment, she developed her menses. Her repeated pelvic magnetic resonance imaging showed the presence of a small uterus.