



# Tall Stature : a diagnosis is sometimes difficult

S. Kherra , H.Bekkakcha, M.BoutabaF.Talbi , Y.Behidj, A. Mekki , Z. Zeroual,  
CHU Parnet Ex. Nafissa Hamoud – Service pédiatrie A



## Introduction

Tall stature is defined as height > 2 standard deviations ( SD ) above the population mean . The most common cause is normal familial tall stature, but some cases are pathological and require special attention .

## Observations

We report four clinical cases corresponding to four diagnostic categories .We describe the diagnostic approach and difficulties encountered through these cases.

### Case N° 01 : An atypical form of adrenal congenital hyperplasia

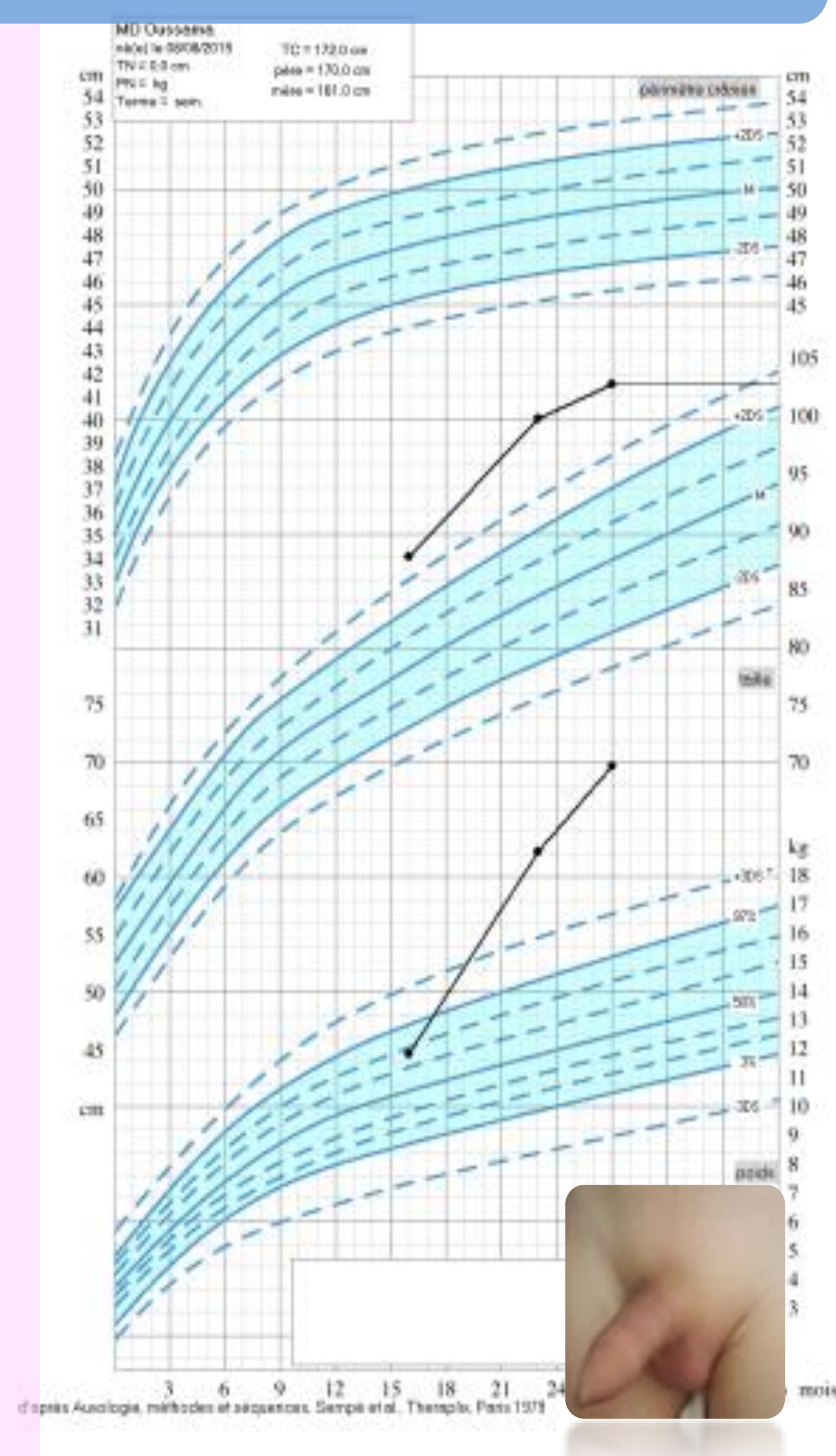
*A diagnosis is sometimes difficult*

A boy aged 25 months was referred for large size, there was a history of repeated hypoglycaemia .

- Examination revealed: weight: 19kg (2+ SD), Height :101 cm (+ 4.5 SD), Tanner: G2P3A1 with testicles : 04 ml testicles .
- Blood Pressure : 90/55 mmHg .
- Bone age 07 years ,
- Hormonal status : FSH : 5,5 mui/ml ( < 4,6 ) LH: 3,9 mui/ml ( < 3,6), testosterone:03.88 ng/ml ( 2,8-8 ), 17hydroxyprogesterone : 4.91 nmol/L, D4 : 17.73 nmol /L , 11Desoxycortisol : 141.10 nmol/L
- Abdominal ultrasound showed bilateral adrenal hyperplasia .

**A diagnosis of precocious puberty with 11beta deficiency was made , a genetic analysis showed a deficiency of CYP11B with an homozygosis mutation p.G379V**

- Therapeutic approach : Hydrocortisone : 15 mg/m<sup>2</sup>/day with Decapeptyl : 1 injection 3.75 mg/ 28 days

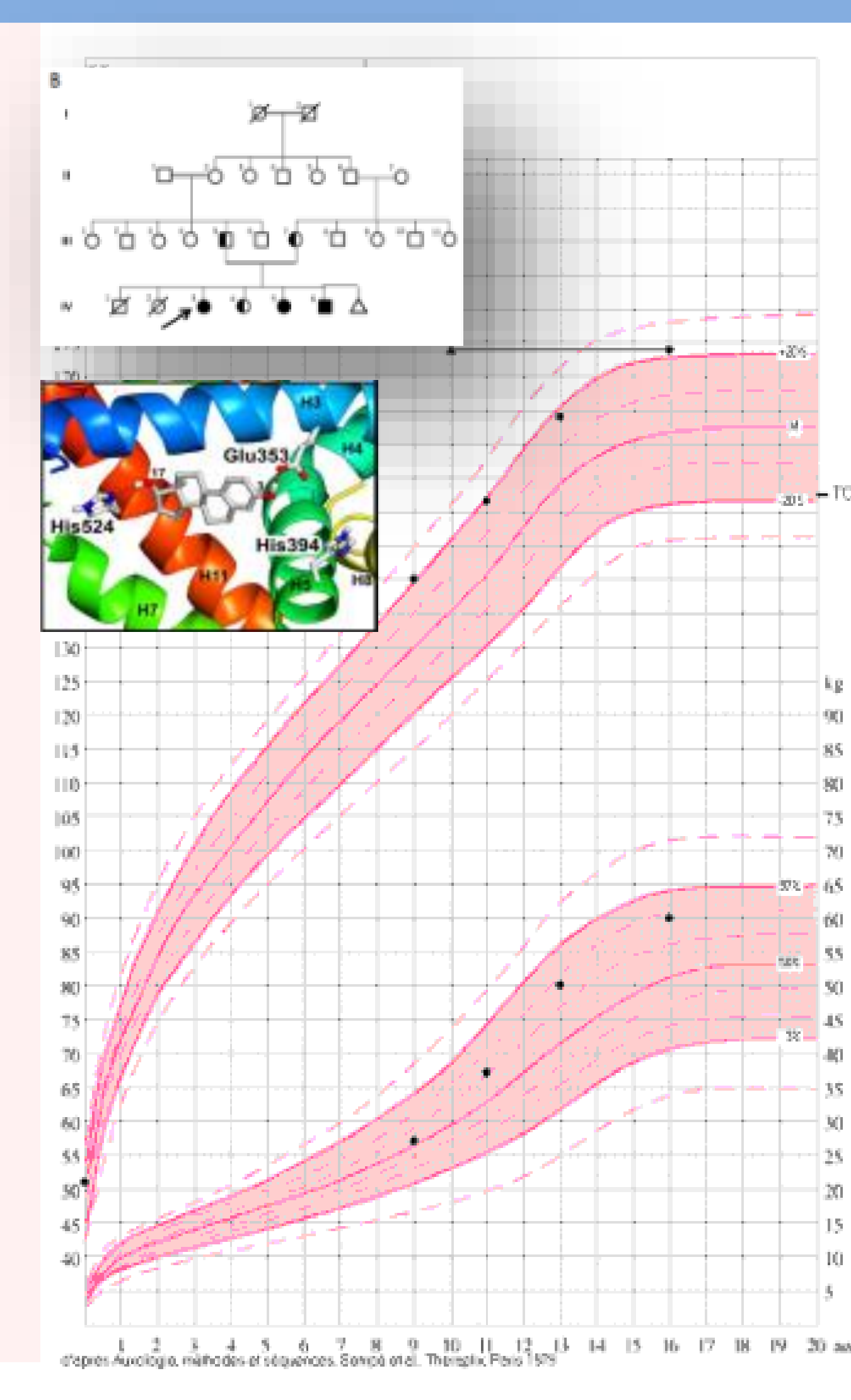


### CASE N° 02 : Estrogen insensitivity caused by mutation in the estrogen receptor α

*A rare genetic disease can be revealed by a tall stature*

•A girl aged 16 years , from a consanguineous family was referred because of primary amenorrhea.

- On examination she was tall, Height : 176cm (+2 SD ), weight :64 kg (+1.5 SD ), BMI: 20.66 kg/m<sup>2</sup> .Tanner :B1P5 , hirsutisme ( Ferriman - Gallwey Score : 22 ) .
- Bone age :10 years .
- Hormonal profile : 17β-estradiol level of 9035 pmol/L (normal 120 to 300 pmol/L during early follicular phase ) ,(LH: 24 IU/L [N: 2 to 8 IU/L], FSH: 13 IU/L [N: 2 to 10 IU/L])
- Karyotype : 46 XX,
- Pelvic ultrasonography showed enlarged bilateral multicystic ovaries with a small uterus.
- This clinical and biological presentation strongly suggested an estrogen resistance syndrome confirmed by genetic studies which showed a c.1181G>A mutation in the fifth coding exon of ESR1 at 394 (Arg 394His) of the ESR1 ligand-binding domain .

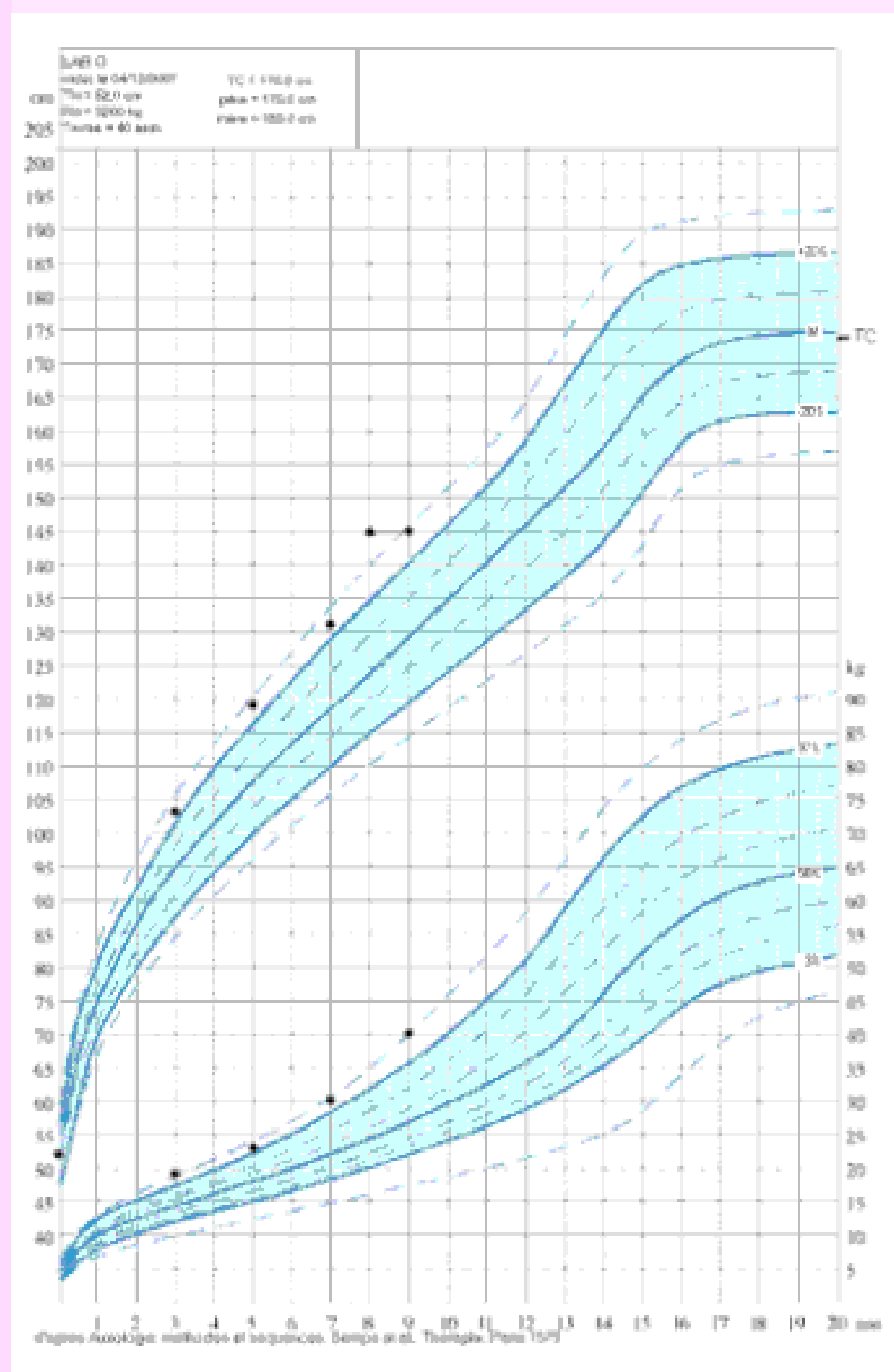


### CASE N° 03 : an abnormality of the X chromosome , 47 XXY

*Klinefelter 's syndrome a common cause of tall stature*

A13-year-old patient from a consanguineous family was referred with tall stature

- BW : 4000 g , HW : 51 cm , H.C : 35 cm
- There was a facial dysmorphism associated with mental retardation, gynecomastia and micropenis, stretched length 3cm) testis : 1.8x1 cm
- Hormonal status :
  - Testosterone : 2.3 ng/ml .
  - FSH : 30 Mui /ML LH : 10 .2 Mui /ml,
- Bone age : 08 years
- Karyotype : 47 XXY
- Diagnosis : **Klinefelter 's syndrome**

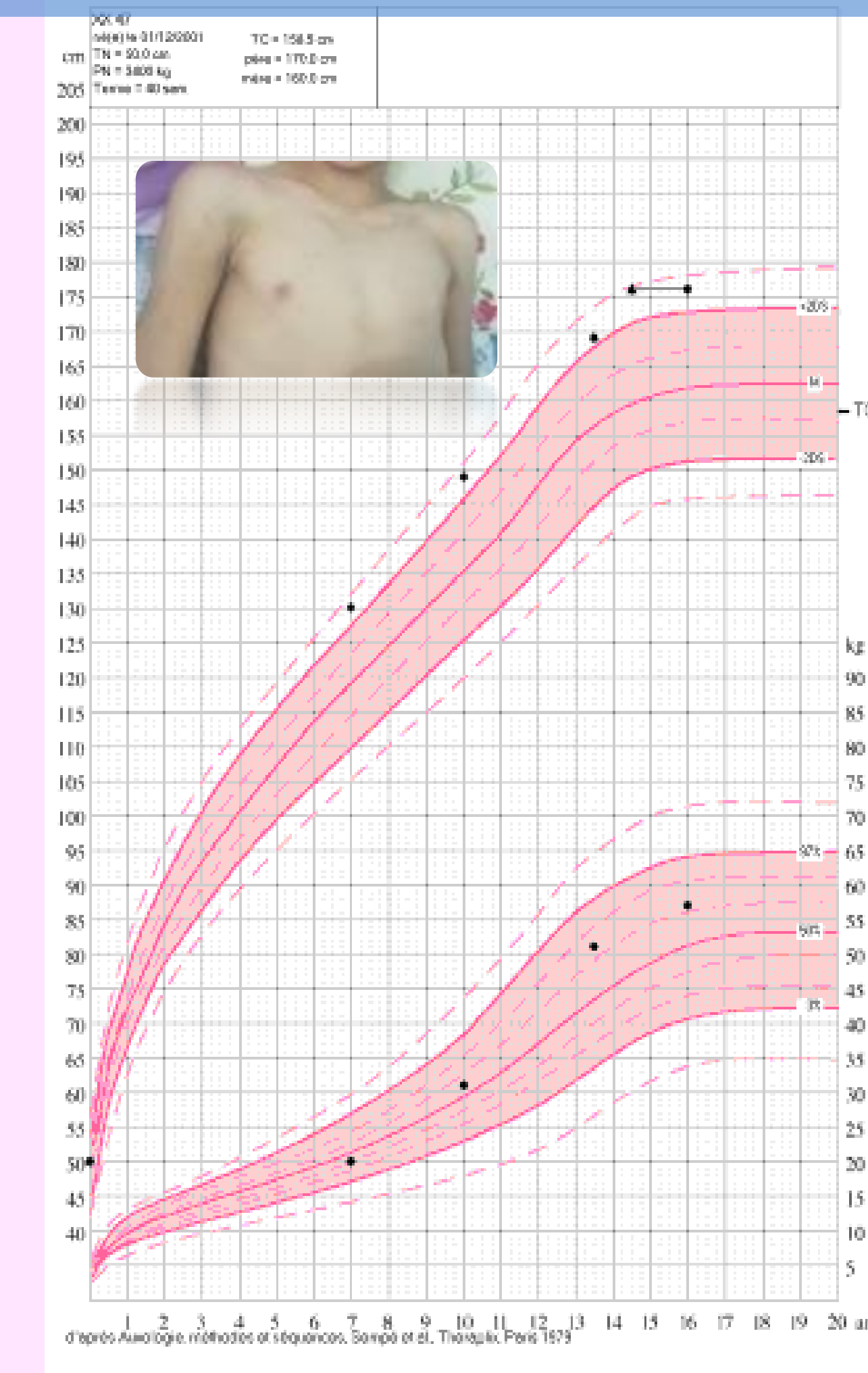


### CASE N° 04 : Another X chromosome anomaly

*Triple X Syndrome , a diagnosis to consider with tall stature*

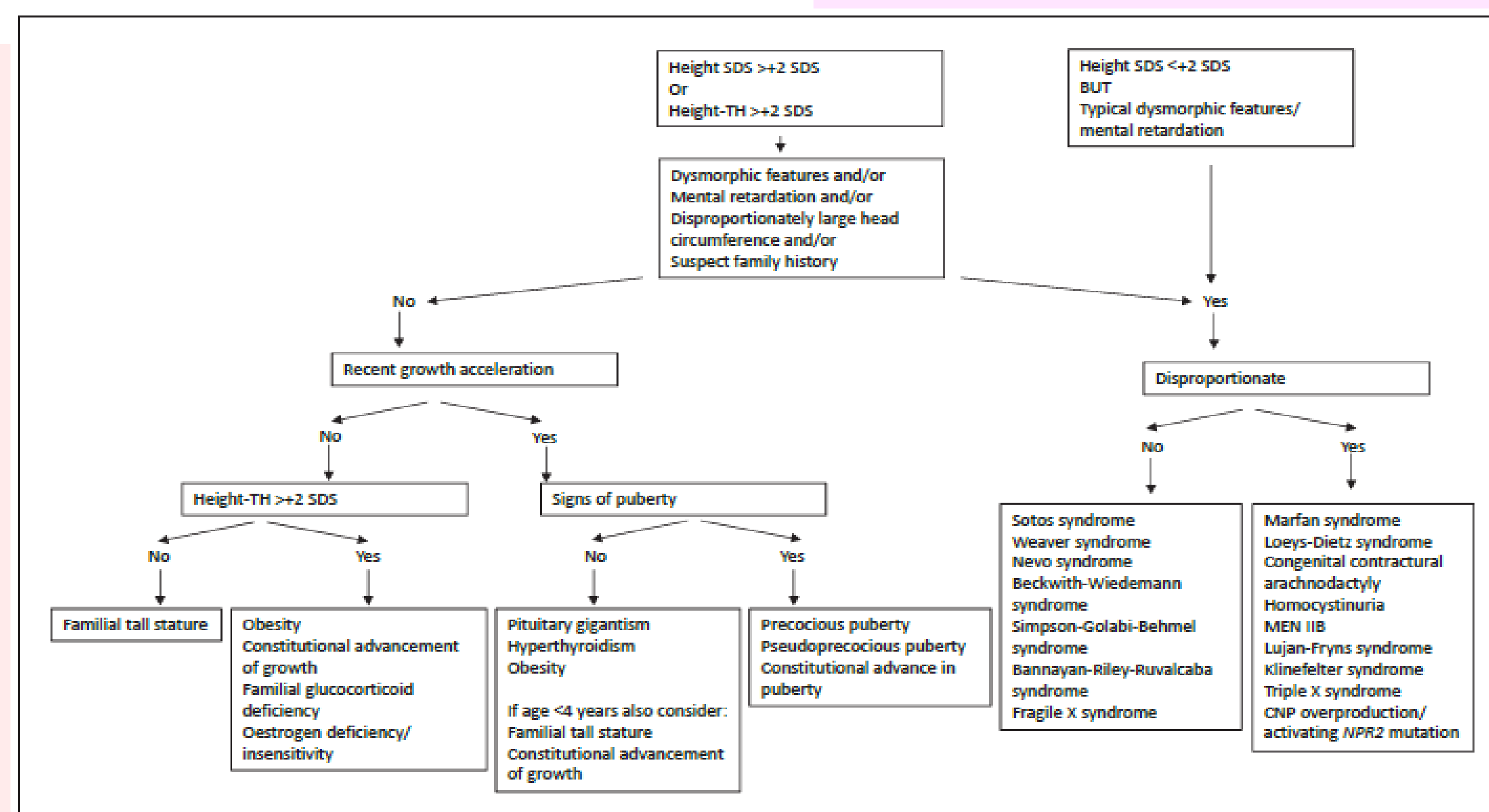
A 16-year-old girl from a 2<sup>nd</sup> degree consanguineous family and a history of epilepsy, was referred for delayed puberty with mild mental retardation IQ = 50 .

- Clinical examination showed : height: 175 cm > +2SD ), mental retardation,
- Tanner : A1S2P2 , hypoplastic labia minora
- Hormonal profile : revealed primary gonadal failure with LH : 24 mui/ml ( N : 2,4 – 12,5 FP) FSH : 30 mui/ml ( N: 3.5 – 12,5 PF ) with estradiol : 3.5 ng / ml ( N :12.5 – 166 PF ).
- Pelvic ultrasound : small uterus
- Bone age : 12 years .
- Karyotype : **47 XXX** indicating triple X syndrome
- Prescribed Treatment: incremental dose of estrogen therapy .



## DISCUSSION AND CONCLUSION

- The diagnostic approach in a child with tall stature is based on a combination of: family and personal history; growth assessment including height velocity and pubertal status; general features (dysmorphism, developmental delay), and the bone age .
- In the presence of developmental delay a genetic causes should be considered; including Klinefelter's syndrome. When the speed of growth is accelerated, an endocrine cause such as true precocious puberty, precocious pseudpuberty and true gigantism must be excluded. .. Rare causes of tall stature include non-classical congenital adrenal hyperplasia while estrogen insensitivity syndrome is particularly rare.
- The third and fourth cases in our series illustrate the importance of karyotype analysis in the etiological research of tall stature , especially when there is dysmorphism and developmental delay. Genetic tests are important in atypical cases of tall stature, including Fragile X syndrome.
- While constitutional advance and familial tall stature remain the commonest causes of tall stature the clinician should always search for a pathological cause of tall stature when this is inappropriate for the family heights and/or if there are accompanying features.



Visser et al ; Hormon Rec Pediatric 2016

Reference : 1 . Edoarda V A and al ; diagnostic and therapeutic approach of tall stature ; European journal of endocrinology 2017 / 2 . T. Edouard, M. Tauber et al ; Grande taille EMC . 2014