*A Case of **SHORT STATURE***

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HISTORY

- A 13yr 8mths adolescent girl from Thiruthani
- 2nd born of 2 degree consanguinous marriage

- With c/o Not gaining adequate height compared to peers.
  c/o Not attained menarche.

History

- She had h/o Recurrent ear infections in the past.
- Good in studies, regular to school
- Appetite and Sleep pattern normal
- Receiving adequate calories and proteins

- Birth h/o: FTND, birth weight 2.6 kg. length and head circumference not known
- Neonatal period uneventful
- Developmental milestones normal
- No family h/o short stature, constitutional delay
- Mother attained menarche at 13yrs.
- Other sibling is male, 15yrs, normal
- Belong to upper middle class
- Immunised uptodate.

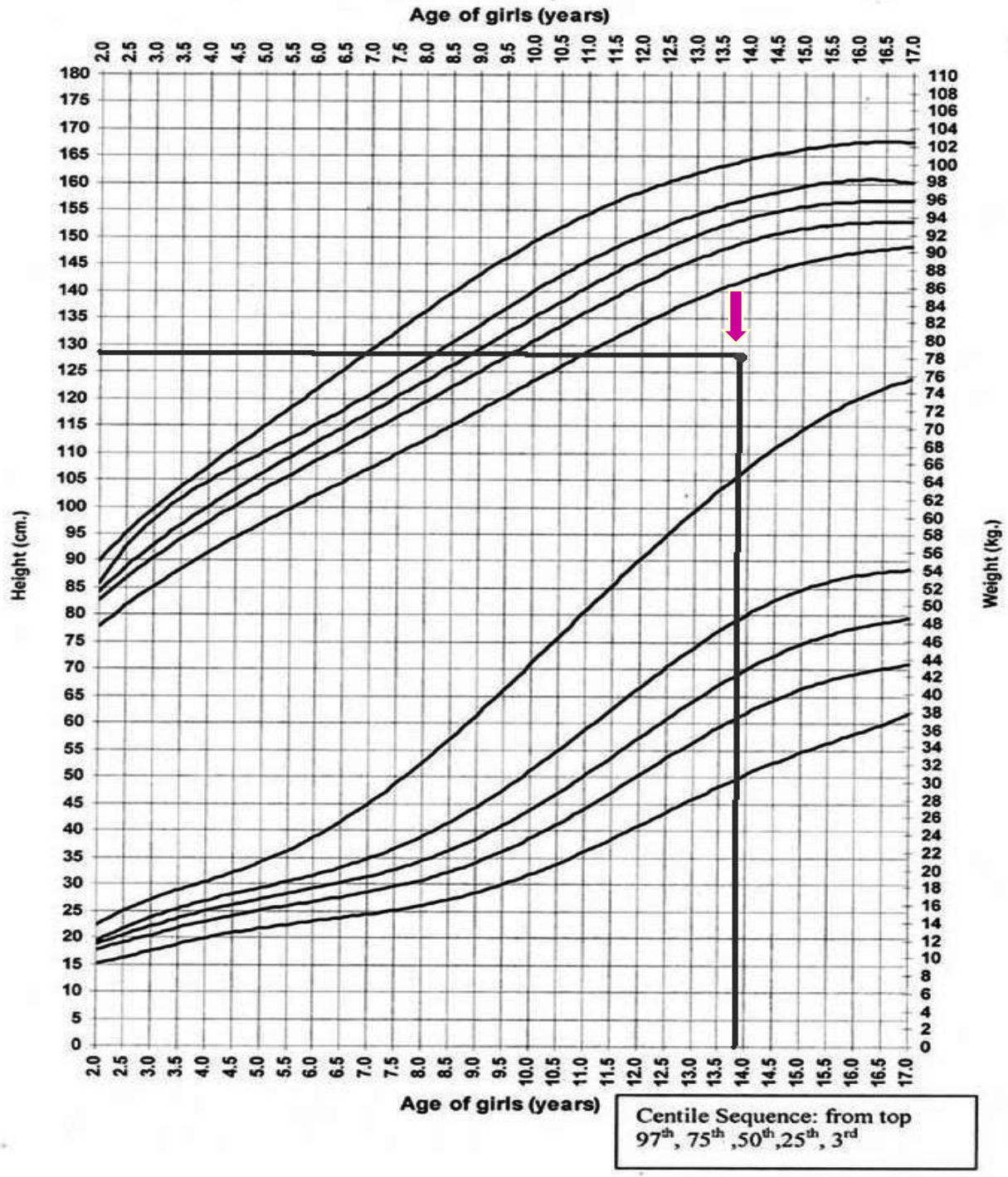
On clinical examination

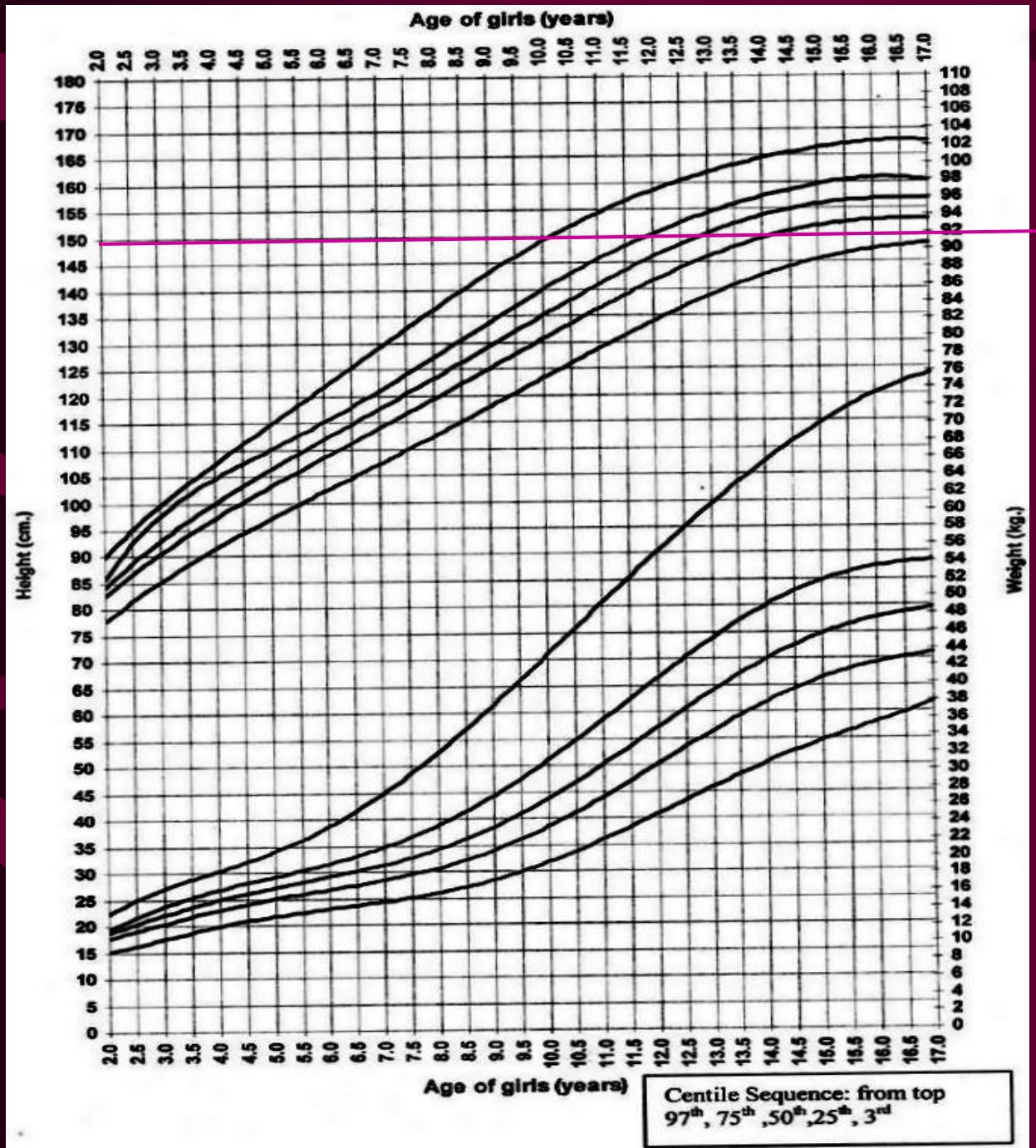
- Conscious, alert, active
- No pallor, icterus, clubbing, edema
- Vitals : Normal
- Anthropometry:

  Weight : 32 kgs (3^{rd} -25^{th} centile)

  Height : 128.4 cms (<3^{rd} centile -IAP)
- Weight for Height - normal

- Head circumference : 51.5 cms (normal)
- Upper seg : lower seg ratio = 1 : 1.09 (normal)
- Arm span : 128 cms
- Father's Height : 164.5 cms
- Mother's Height : 147.5 cms
- Midparental Ht : 149.5 cms (3^{rd}-25^{th} centile)
- Height Age – **9 years**
- BMI – 19.6 (50^{th} -75^{th} centile)


Age of girls (years)
Height (cm.)
Weight (kg.)
Age of girls (years)
Centile Sequence: from top
97th, 75th ,50th ,25th, 3rd



- ✓ Mid Parental Height-149.5

Head to foot examination

- Dolichocephalic skull .
- No midline defects, No dysmorphic facies.
- ENT – R .ear CP with CSOM with discharge
- Oral cavity – High arched palate
- Fundus – normal .
- Neck-No webbing ,No Thyroid swelling.
- Short neck.

- Hyperpigmented Nevi over abdomen +.
- Cubitus Valgus deformity +.
- Flat foot + , Short Great toe.
- Tanner staging – Stage 1 (Preadolescent)
- Systemic examination : Normal
- No kyphoscoliosis

Investigations

- Baseline investigations – Normal
- Renal function tests – Normal
- Liver function tests – Normal
- Serum Calcium – 9.2 mg%
- Serum Phosphorus – 4.9 mg%
- X ray Elbow showed Bone Age 11-13 years.
- ECHO – ASD secundum(3mm)
- Karyotyping sent

- **Thyroid Function Test**: Normal

  T3 – 2 nmol/L

  Free T4 – 15.3 pmol/L

  TSH – 6.2 microunits/ml

- **Estimation of Growth Hormone levels (Clonidine Stimulation Test) – after priming**

  Basal – 0.14 ng/ml

  30 - 0.95 ng/ml

  60 - 5.42ng/ml

  90 - 3.58ng/ml

  120 - 0.77ng/ml

  150 - 0.28ng/ml (GH Deficiency)

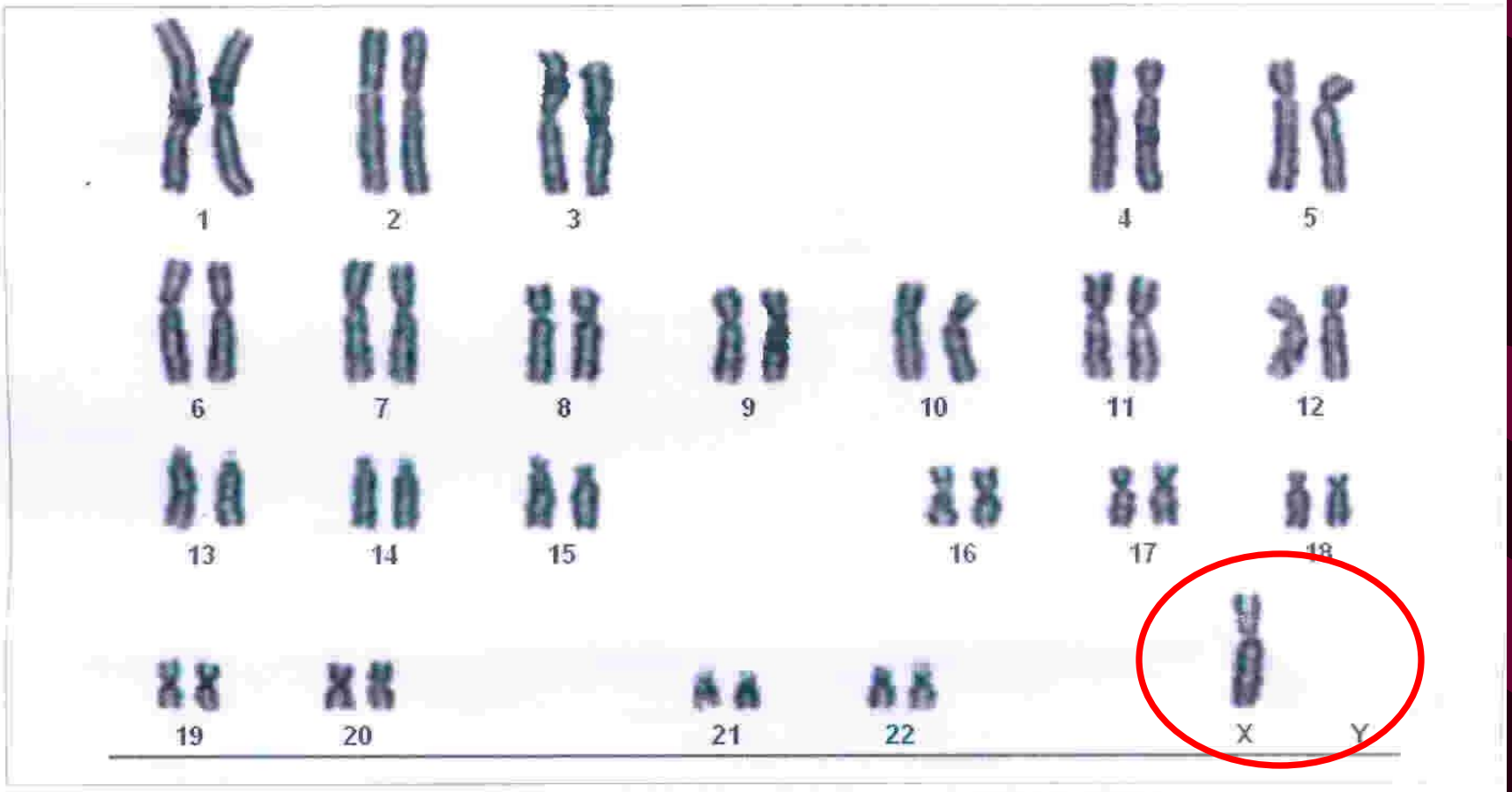
- IGF- I levels – 109 (183-850)
- IGF BP3 levels – 3.73 (3.1-9.5)

- USG Abd – Small uterus , Ovaries not imaged , kidneys N.
- Serum Prolactin – 7.3 ngm/ml (3.34 – 26.72)
- FSH – 137.9 mIU/L (1.79 -5.12)
- LH – 24.4 mIU/L (1.20 – 12.86)

- SHORT STATURE
- DELAYED PUBERTY
- HYPERGONADOTROPIC STATE

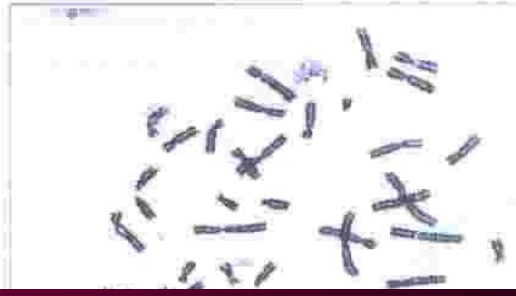
SUSPECTED TURNER SYNDROME

REPORT OF KARYOTYPE ANALYSIS



KARYOTYPE : 45,X

REMARKS : TURNER SYNDROME



DIAGNOSIS

TURNER SYNDROME

Treatment given

- Growth hormone therapy : 0.05mg/kg/day Started on 5 units/day (3units = 1mg)
- R ear CSOM - ENT opinion sought and treated accordingly.
- Flat foot – Orthopedic opinion sought and was given special footwear.
- Gynaecological opinion and psychological counselling given.
- Child is under cardiological evaluation for ASD.

DISCUSSION

Turner Syndrome - Introduction

The condition is defined as the combination of characteristic phenotypic features accompanied by complete or partial abscense of the second X chromosome with or without mosaicism

Incidence: 1 in 1500-2500 live born females

CHROMOSOME ABNORMALITIES IN TURNER SYNDROME

1	45 X	60%
2	MOSAIC XX/X	20%
3	ISOCHROMOSOME Xq or Xp	5%
4	46 X del (X)	5%
5	46 X RING (X)	5%
6	WITH Y CHROMOSOME	5%

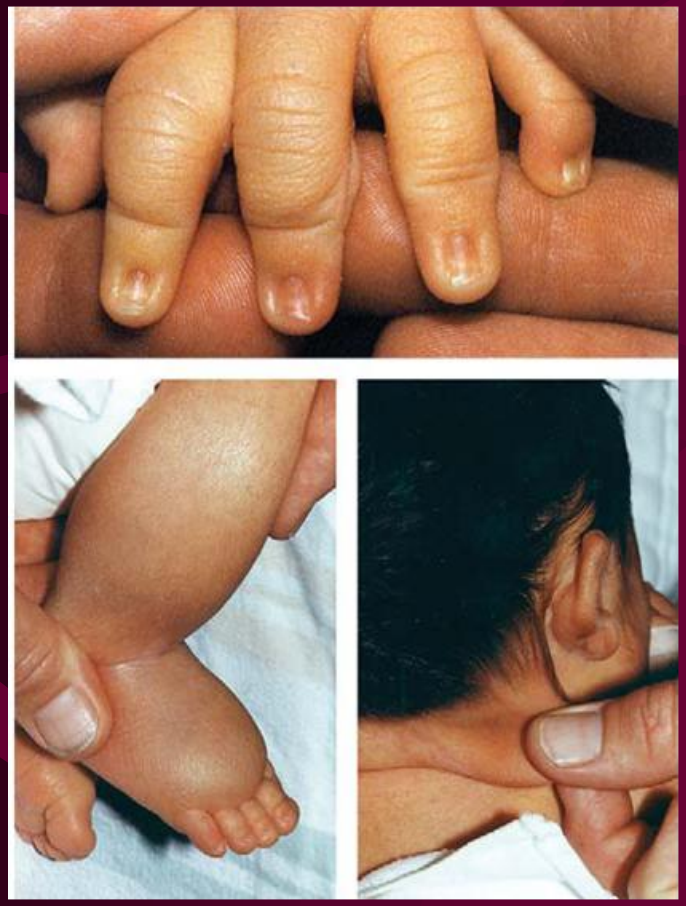
95-99% of 45 XO conceptions are miscarried

- Parenteral age has no effect on incidence
- X chromosome is maternal in origin in 76% and paternal in 24%
- XY cell line in mosaic Turner may be assoc with virilization and 20% risk of Gonadoblastoma.
- *SHOX gene (short stature homeobox containing* gene) located on the long arm of the X chromosome is a key contributor to growth restriction.

CLINICAL FEATURES

- Classical turner (45XO) show many characteristic abnormalities
- Short stature and streak ovaries are constant findings irrespective of chromosomal constitutions.

- At birth: Edema on dorsum of hands/ feet, loose skin-folds on nape of neck, low birth weight, decreased length.



- Delay or failure in the development of the communication between the lymphatic system and the venous system.

- Childhood:

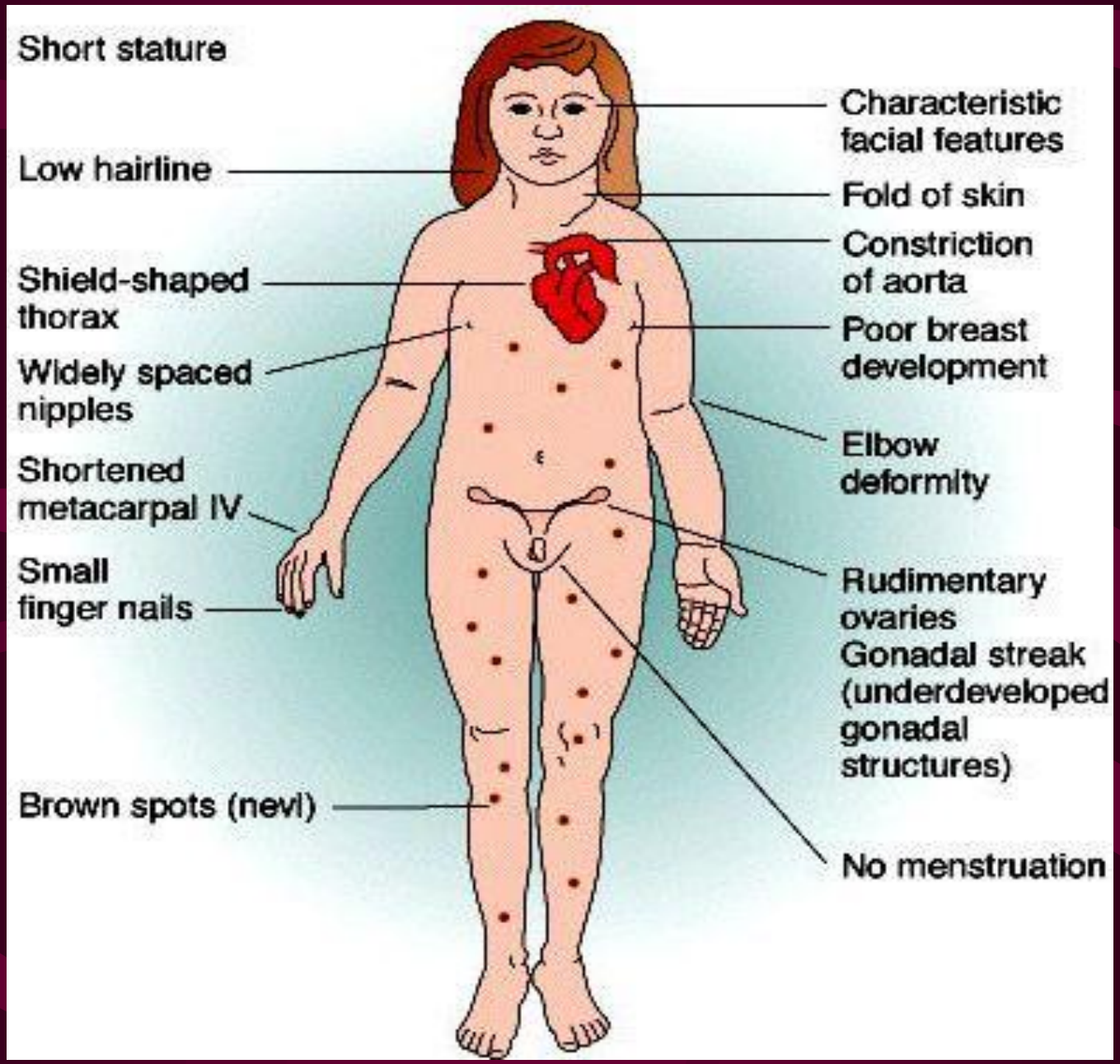
  Webbing of neck, low posterior hairline, small mandible, prominent ears, epicanthic folds, high arched palate, broad chest with widely spaced nipples, cubitus valgus, hyperconvex fingernails, excessive number of nevi, Short 4^{th} metacarpal .

- Short stature :

  Mean adult height 143 cm (132-155cm)

- CVS: Bicuspid aortic valve (33%), aortic stenosis, aortic coarctation, anomalous pulmonary venous drainage.
- Renal malformations (33%): pelvic kidney, horse shoe kidney, double collecting system, complete absence of one kidney, uretero-pelvic junction obstruction.
- Others : Recurrent bilateral otitis media (75%), sensorineural hearing deficits. Anterior segment dysgenesis and keratoconus
- Lymphocytic thyroiditis.
- Inflammatory bowel disease ,Osteoporosis

- Ovaries (USG): Streak ovaries in 90% by 4-10 years age. Failure of sexual maturation, amenorrhoea, 10-20% may have some breast development, pregnancy rarely reported.
- CNS: Normal intelligence, Problems with gross & fine motor- sensory integration, language dysfunction, Mental retardation in patients with ring chromosome, Defects in perceptual spatial skills.


Short stature
Low hairline
Shield-shaped thorax
Widely spaced nipples
Shortened metacarpal IV
Small finger nails
Brown spots (nevi)
Characteristic facial features
Fold of skin
Constriction of aorta
Poor breast development
Elbow deformity
Rudimentary ovaries
Gonadal streak (underdeveloped gonadal structures)
No menstruation

Treatment / Prognosis

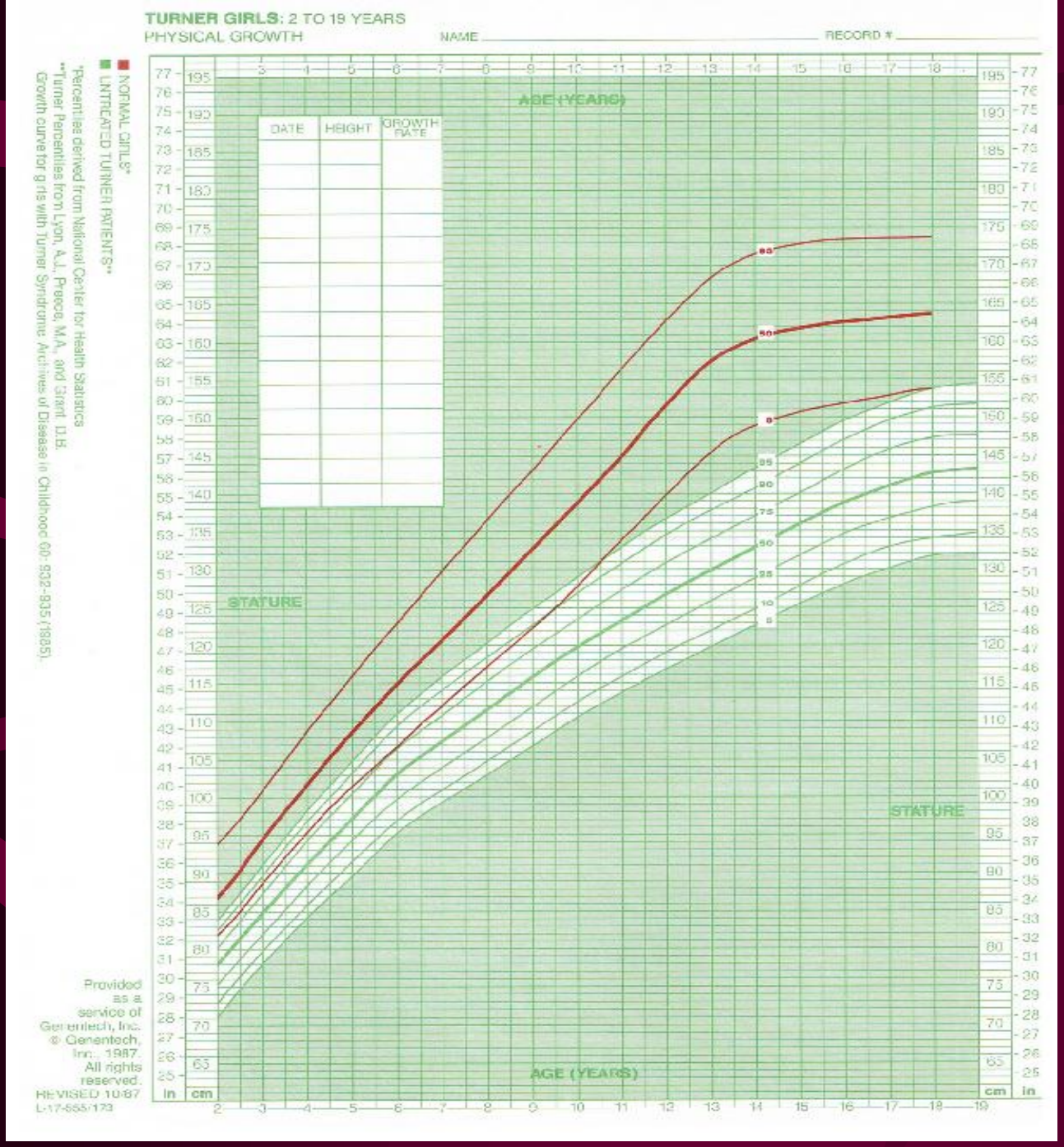
- Growth hormone for height.
- Estrogen therapy for puberty.
- Psychosocial support.
- Plastic surgery for physical abnormalities.

- Normal life expectancy, lead normal, independent, productive lives.

TREATMENT

- Growth Hormone – Dose 54 mcg/kg/day
- Start at around 5 to 6 yrs of age, continue till around 15 – 16 yrs of age.
- The gain appears to be 6 – 8 cms after 5 to 7 yrs of Rx.
- Patients receiving growth hormone must be monitored closely for increased intracranial pressure, hypothyroidism, glucose intolerance, edema, and any changes in the nevi.

- Replacement therapy with estrogens :
- Estrogen treatment may need to be delayed until 14–15 yr of age.
- A conjugated estrogen (Premarin) 0.3–0.625 mg, or micronized estradiol 0.5 mg . Given daily for 3–6 mo is usually effective in inducing puberty.
- The estrogen then is cycled (taken on days 1-23), and a progestin(5–10 mg daily) is added(days 10-23).
- In the remainder of the calendar month, during which no treatment is given, withdrawal bleeding usually occurs.



- Specialized growth curves for girls with TS

Review

- **Atrial septal defect as an uncommon cardiovascular malformation with Turner's syndrome.**

Hirose H, Takagi M, Tada S, Kugimiya T.

Department of Cardiovascular Surgery, Shin-Tokyo Hospital, Chiba, Japan.

Abstract

Cardiovascular malformations are frequently observed in Turner's syndrome. Bicuspid aortic valve and coarctation of aorta are commonly associated with Turner's syndrome while an atrial septal defect is unusual. A rare case of atrial septal defect with Turner's syndrome reported by

Department of Cardiovascular Surgery, Shin-Tokyo Hospital, Chiba, Japan.

References

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- V.V. Khadilkar, A.V. Khadilkar, M. Nandy and G.B. Maskati **Growth hormone in Turner syndrome** Indian Pediatrics : Mar 2006;43:236 – 240
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- Rosenfeld RG, Brasel JA, Burstein S, Chernausek SD, Johanson AJ. Growth hormone therapy of Turner's Syndrome: beneficial effect on adult height. J Pediatr 1998; 133: 803-804.
- Davenport ML, Punyasavatsut N, Stewart PW, Gunther DF, Savendahl L, Sybert VP. Growth failure in early life: an important manifestation of Turner syndrome. Horn Res 2002: 57: 157-164.

Take home messages

- High index of suspicion for Turner syndrome is required in adolescent girls presenting with short stature and delayed puberty.
- Karyotyping is advised for all girls presenting with short stature
- Timely intervention helps in achieving increase in height and psychosocial wellbeing.

THANK YOU