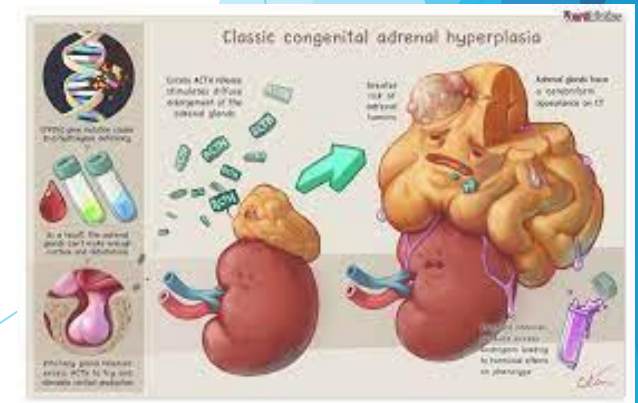


# Congenital adrenal hyperplasia

**Dr. Meghna Khetan**  
**Resident**  
**Department Of Paediatrics**  
**Dr. Dy Patil Medical College**



# Case 1

- ▶ A 7 year old female presented with:
  - Excessive acne
  - Facial hair growth
  - Masculine voice
  - Unusual clitoral growth

Changes noticed since 2 years, gradually progressive



# NEGATIVE HISTORY

- ▶ No ingestion of drugs such as anabolic steroids
- ▶ No history of repeated hospital admissions
- ▶ No history of recurrent diarrhoea
- ▶ No history of recurrent vomiting

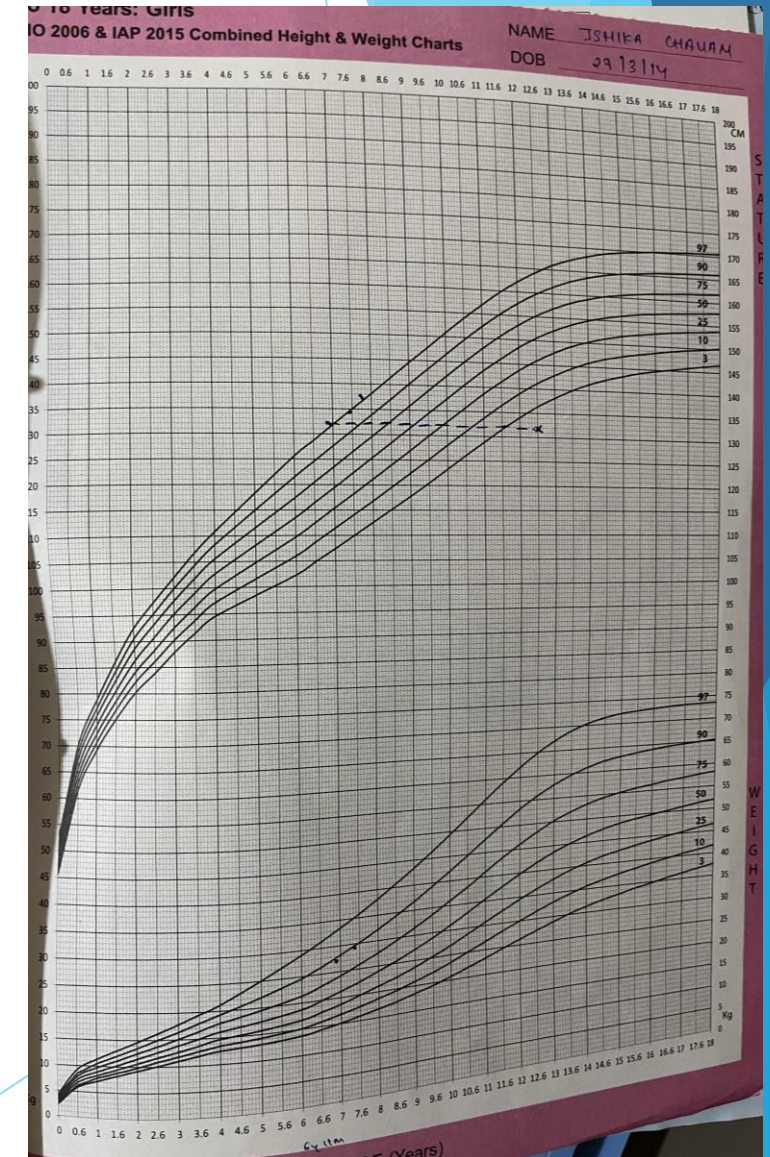
- ▶ **PAST HISTORY:** She was born with atypical appearing genitalia but was not evaluated but at age of 3y of age operated for fused labia.
- ▶ **FAMILY HISTORY** – no history of infertility or similar issues
- ▶ **BIRTH HISTORY** Full term home delivery with an average birth weight, with atypical genitalia noticed at birth.
- ▶ **IMMUNIZATION HISTORY** : completely immunised for age
- ▶ **DEVELOPMENTAL HISTORY** – normal for age

# ANTHROPOMETRY

Weight 27kg {between 90<sup>th</sup> to 97<sup>th</sup> centile}

Height 132.5cm {above 97<sup>th</sup> centile }

BMI 15.49 {between 50<sup>th</sup> to 90<sup>th</sup> centile}


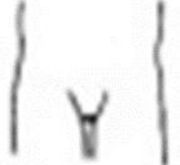










# GENERAL EXAMINATION

- ▶ The child was vitally stable having HR 98,RR 31,PP well felt, BP 104/68 and SPO2 of 99%
- ▶ Tall stature with muscular built
- ▶ Facial hair growth, acne
- ▶ Sexual maturity Rating –
  - Axillary hair – stage III, Pubic hair – stage IV
  - Breast stage –Stage III, no menarche



# TANNER'S STAGING

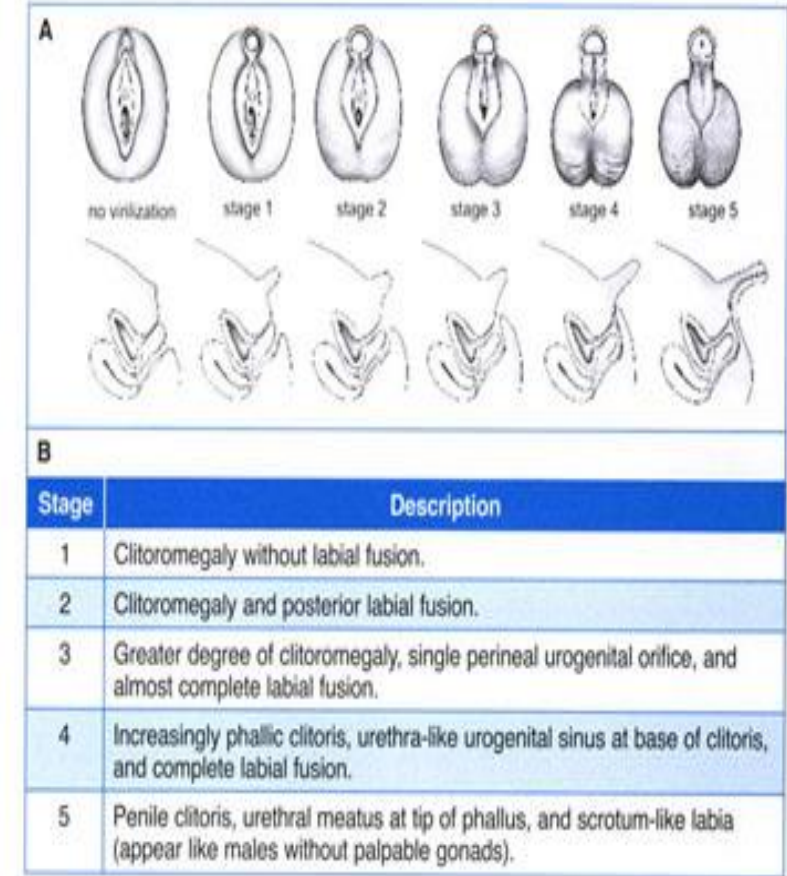
PREPUBERTAL		1		PREPUBERTAL
BREAST BUD		2		PRESEXUAL HAIR
BREAST ELEVATION		3		SEXUAL HAIR
AREOLAR MOUND		4		MID-ESCUTCHEON
ADULT CONTOUR		5		FEMALE ESCUTCHEON

## ► Genital examination -

- Enlarged clitoris, 2cm length x 1 cm width
- Hyperpigmentation present.
- No palpable gonads in labial folds.

**Figure 4.**

The Prader scale (A) is a grading system used to measure the degree of genital virilization (masculinization) on a scale from "0" to "5". Stage "0" refers to an unvirilized female whereas stage "5" reveals complete virilization (a female who appears externally male at birth). The chart below the image (B) describes the images of the Prader virilization stages.



Source: Image reprinted with permission from Emerick, 2012.



# SYSTEMIC EXAMINATION

- ▶ CARDIOVASCULAR SYSTEM : S1 S2 normal,  
No murmur
- ▶ CENTRAL NERVOUS SYSTEM : conscious,  
alert, oriented to time place and person
- ▶ RESPIRATORY SYSTEM : bilateral air entry  
equal on both sides
- ▶ PER ABDOMEN: soft, non tender with no  
organomegaly

# BLOOD INVESTIGATIONS

- ▶ Hemogram , liver function tests, renal function tests and electrolytes were done with were all within normal limits

	Sodium (135-155)	Potassium ( 3.5-5.5)
On admission	141	3.8
After 7 days	138	3.5

# BLOOD INVESTIGATIONS

## ▶ HORMONAL PROFILE –

- ▶ 17 OHP : **7.13 mmol/l** ( <3.03mmol/l) ↑
- ▶ TESTOSTERONE : **282.95ng/dl** (2.69-10.29ng/dl) ↑
- ▶ CORTISOL : **282.95ng/dl** (2.69-10.29ng/dl) ↑
- ▶ ACTH: **282.95ng/dl** (2.69-10.29ng/dl) ↑

## ▶ PUBERTAL HORMONAL PROFILE –

- ▶ LH :0.67IU/L ( >0.3 pubertal range) ↑
- ▶ FSH:4.32IU/L (pubertal range)
- ▶ Estradiol : **32pg/ml** (prepubertal <20pg/ml) ↑

# RADIOLOGICAL INVESTIGATIONS

- ▶ USG ABDOMEN PELVIS
- ▶ Uterus in pubertal size
- ▶ Adrenal Glands: 14\*3 suggestive of hyperplasia of the gland
- ▶ BONE AGE – 12.7 YEARS , Chronological Age – 6.11 YEARS ( advanced)

# GENETIC PROFILE

- ▶ Karyotype 46xx
- ▶ GENETIC TESTING confirmed homozygous mutation

# FINAL DIAGNOSIS

- ▶ Simple virilizing congenital adrenal hyperplasia with
- ▶ central precocious puberty

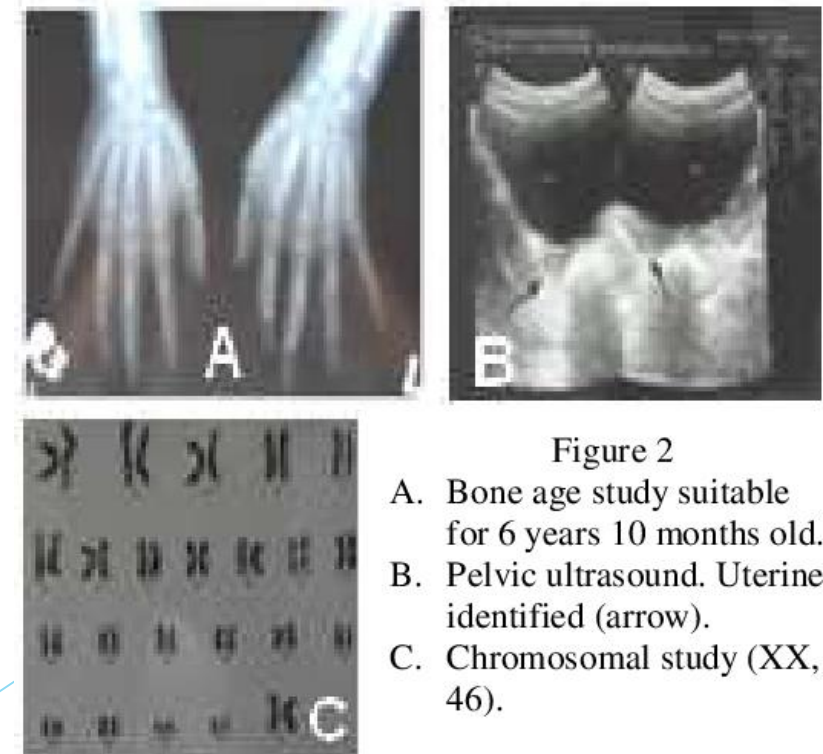


Figure 2

- A. Bone age study suitable for 6 years 10 months old.
- B. Pelvic ultrasound. Uterine identified (arrow).
- C. Chromosomal study (XX, 46).



# SUMMARY

- ▶ A 7 year old female presented with acne, hirsutism, cliteromegaly having a tall stature . On further evaluation blood hormones were raised for her age with adrenal gland hyperplasia on usg abdomen pelvis , chromosomal study indicative of 46XX
- ▶ All suggestive of simple virilising type of congenital adrenal hyperplasia

# MANAGEMENT

- ▶ Tab. Hydrocortisone 5mg twice a day
- ▶ (Dose –  $10\text{mg}/\text{m}^2/\text{day}$ )
- ▶ Inj. Depot Medroxy progesterone acetate 150mg monthly for treatment of Central precocious puberty ( to postpone menarche)

# FOLLOW-UP

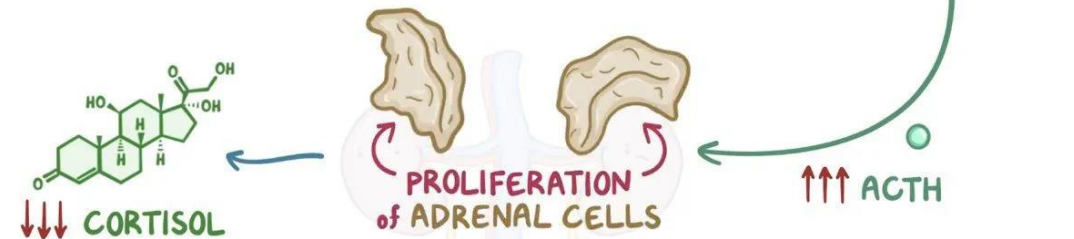
Age	Height	Weight	Clinical features	17OHP (ng/ml)	Testosterone (ng/dl)	Management
7 years	132.5cm	27kg	Clitoromegaly with acne and hirsutism	7.13	282.95	T hydrocortisone 5mg BD and inj DMPA 150mg monthly
7.5 years	135cm	29.5kg	Reduced clitoromegaly with decreased acne		12.98	Tab hydrocortisone 5mg BD with Inj DMPA 300mg monthly
7.7 years	138cm	30.6kg	Slight Clitoromegaly			Tab hydrocortisone 5mg BD with Inj DMPA 300mg monthly



## CASE 2

### CONGENITAL ADRENAL HYPERPLASIA

- \* ENLARGED ADRENAL GLANDS at BIRTH
- \* CAUSED by ENZYME DEFICIENCIES in the ADRENOCORTICAL STEROID PATHWAY
  - ↳ ↓↓↓ ADRENOCORTICAL STEROIDS



▶ A 14 day old male child presented to casualty with complains of

1. Lethargy
2. Non acceptance of feeds
3. Increased work of breathing
4. Fever

On examination the child had a GCS 9/15 HR of 70, RR of 20, having feeble pulses with cold extremities with a bsl of 50mg/dl

The child was admitted in nicu where he was mechanically ventilated

Given 2 boluses followed by maintenance fluids thereby stabilising him

- ▶ **Birth history:** born via normal vaginal delivery having a birth weight of 2.2kg cried immediately at birth and discharged at day 2 of life, born out of a non consanguineous marriage 2<sup>nd</sup> by birth
- ▶ No significant antenatal history
- ▶ **Family history :** no similar issues in elder sibling
- ▶ **Immunization history :** completely immunised till age

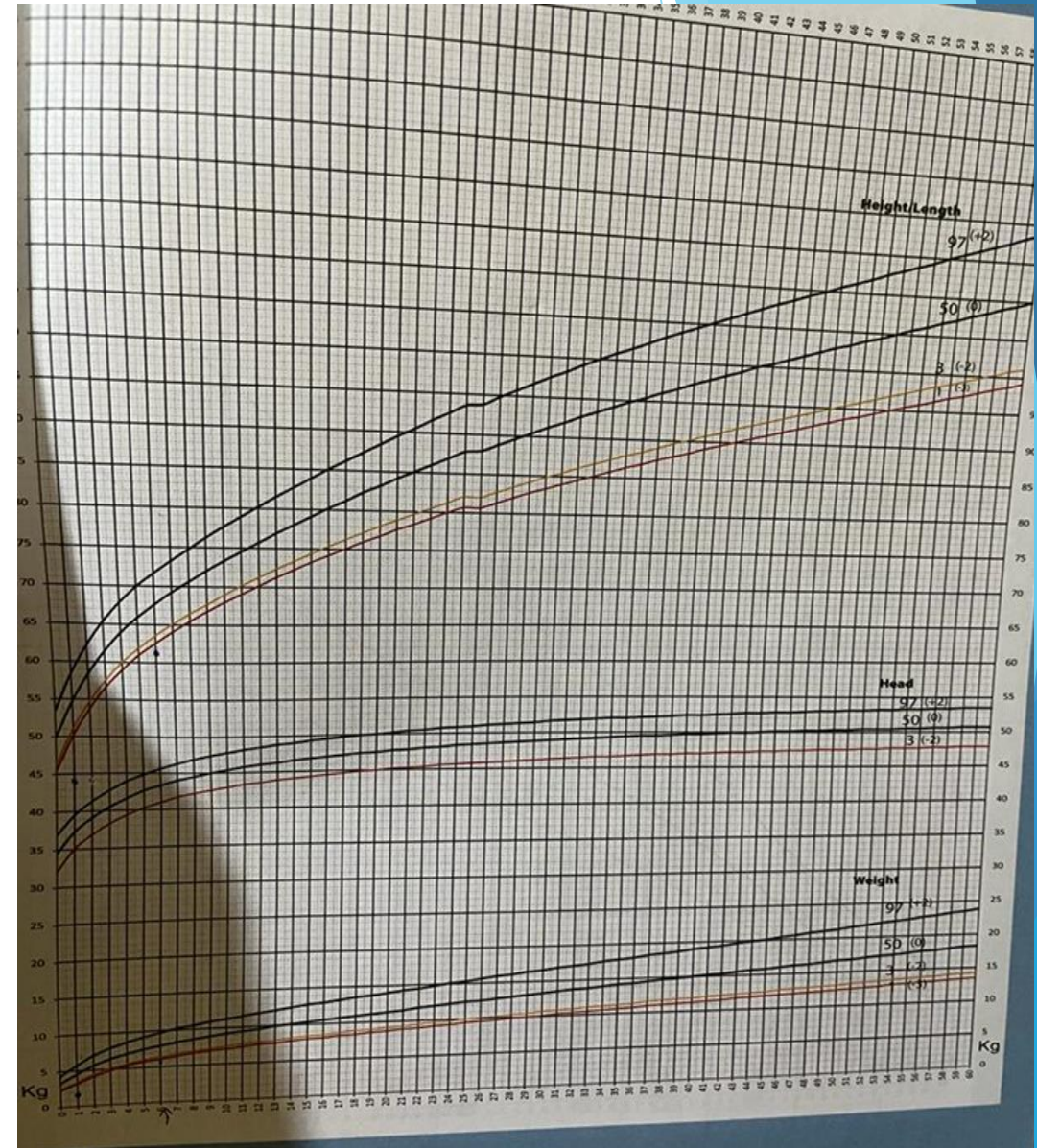


# ANTHROPOMETRY


Birth weight : 2.2kg

Length : 44cm

Head circumference:  
35cm



# INVESTIGATIONS

- ▶ Hemogram , liver function tests, renal function tests and electrolytes and blood gas analysis were sent
- ▶ There was severe hyponatraemia with hyperkalamia
- ▶ There was severe metabolic acidosis
- ▶ Renal function tests were suggestive of acute renal failure
- ▶ Serum cortisol and adrenocorticotrophic hormone were also sent as we were suspecting adrenal crisis which were in turn markedly raised.
- ▶ ACTH 88 

# INVESTIGATION

	DAY 1	DAY 3	DAY 7	DAY 14
Sodium	109	119	125	130
Potassium	7	6.1	5.9	4.5
pH	6.8	7.2	7.3	7.4
Bicarbonate	<3	9	10	12
Urea	161	89	68	40
Creatinine	9.1	5.2	4.5	1.8

# FINAL DIAGNOSIS

SALT WASTING TYPE OF  
CONGENITAL ADRENAL  
HYPERPLASIA



# MANAGEMENT

- ▶ The child was admitted in nicu and was mechanically ventilated. Was started on maintenance fluids after giving 2 boluses.
- ▶ All the electrolyte imbalances were corrected
- ▶ The child was started on iv hydrocortisone at a dose of 100mg/kg/day for treatment of adrenal crisis
- ▶ Gradually over a period of the labs of the child improved and was In a stage of resolving AKI.
- ▶ The child was removed from mechanical ventilator on day 3 of admission and was gradually started on feeds
- ▶ At the end of 14 days the child was able to accept full feeds

# SUMMARY

- ▶ A 14 day old male child presented to casualty in shock, mechanically ventilated and admitted in nicu for 2 weeks.
- ▶ Labs suggestive of diselectrolytemia, with deranged renal function tests and raised acth levels and treated for the above
- ▶ Started on iv hydrocortisone and treated as salt wasting type of congenital adrenal hyperplasia



# FOLLOW UP

AGE	17 OHP	Testosterone	Management
26 days	>20ng/dl	66.37ng/dl	Tab hydrocortisone at 20mg/kg/day Tab fludrocortisone at 50mcg BD
1.5 month	14ng/dl	45ng/dl	Tab hydrocortisone at 20mg/kg/day Tab fludrocortisone 50mcq BD
7 month	10ng/dl	34ng/dl	Tab hydrocortisone at 20mg/kg/day Tab fludrocortisone 50mcq BD

