BIRTH DEFECTS

A case of Down Syndrome

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Case History

- Mrs. B.K, 41 yrs homemaker from Taplejung
- Referred from Amda hospital
  - BP: 200/110 Rt and 190/120 mm Hg on left armLt
  - Headache still persistent
  - Cap Depin 10 mg po stat given with tab Amlod 10 mg po stat
- DOA: 077/11/11 at 6:30pm
- Prov diagnosis:

  G4P3L1 at 36+5 weeks POG with severe pre-eclampsia

Clinical status

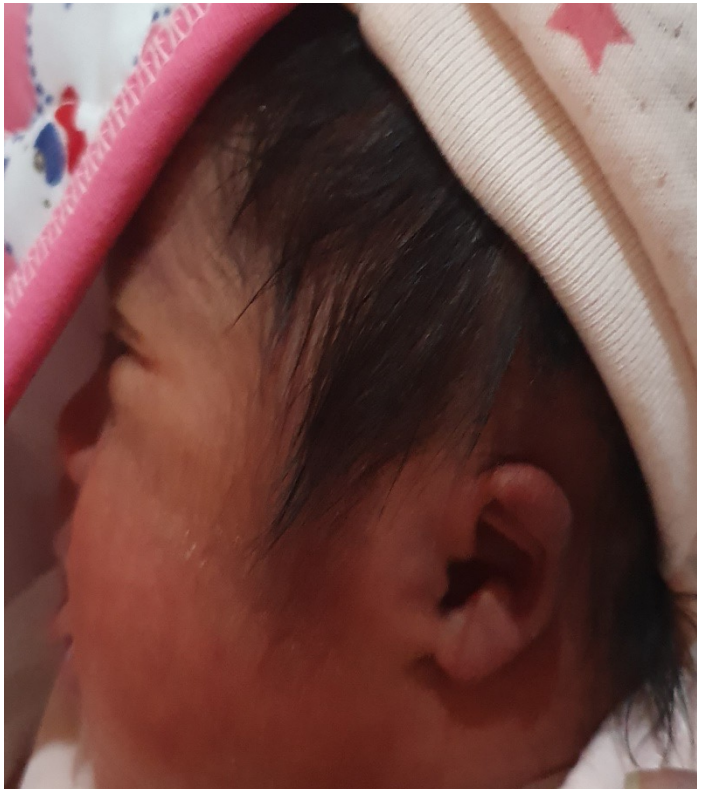
077/11/11

- Frontal headache, pedal edema
- P=106bpm, BP=150/100mmHg
- P/A- ut 34/52, cephalic, no contraction, FHS+
- MgSO4/Dexamethasone/Depin

077/11/12

- Planned for Em LSCS
- Hb-11.2 gm%
- Platelets-5,94,000/cumm
- PT-15sec
- LFT/RFT/LDH:WNL
- Urine albumin +
- Hematuria +

DOR on 077/11/15



Postnatal

- Delivered S/L/M at 1:20 pm with A/S 8/10 and 9/10, weight: 2.5 kgs.
- Examined by pediatrician, advised for NICU admission due to features suggestive of Down syndrome but denied by pt

Features of DS

- a flat nasal bridge
- brachcephaly or shortened frontal lobe
- clinodactyly /short fifth finger
- hypotonia
- epicanthic folds
- upward slanting palpebral fissures

- white spots on the iris known as Brushfield spots
- excessive joint laxity including atlantoaxial instability
- excessive space between large toe and second toe
- small ears and mouth

- Absent fetal nasal bone- in 2/3rd cases of DS
- Reversed a wave in ductus venosus
- Tricuspid regurgitation

Soft tissue marker

1. Single umbilical artery
2. Short femur
3. Short humerus
4. Single transverse palmer crease

Diagnosis

Any of 2 soft tissue marker should be positive to diagnose down syndrome

Screening tests

- First trimester Screening test
  - Nuchal translucency
  - Biochemical test free B HCG and PAPP-A
- 2nd trimester Screening test
  - Triple test
  - Quadruple test
- Integrated test
- Cell free DNA test(non invasive prenatal testing)

Interpretation

Double test

-B-hcg inreased and PAPP-A decreased

Triple Test

-B-hcg increased,MSAFP decreased and serum estriol decreased

Quadruple Test

-B-hcg increased,MSAFP decreased,serum estriol decreased and Inhibin A increased

Down Syndrome Detection Rate

Screening test at 30 years of age	Detection rate, %
DOUBLE TEST	60-65
TRIPLE TEST	65-70
QUADRUPLE TEST	70-75
1ST TRIMESTER COMBINED TEST	90
CELL FREE DNA TEST	99

Risk of Down Syndrome by AGE

Maternal Age	Incidence of Down syndrome	Maternal Age	Incidence of Down syndrome	Maternal Age	Incidence of Down syndrome
20	1 in 2,000	30	1 in 900	40	1 in 100
21	1 in 1,700	31	1 in 800	41	1 in 80
22	1 in 1,500	32	1 in 720	42	1 in 70
23	1 in 1,400	33	1 in 600	43	1 in 50
24	1 in 1,300	34	1 in 450	44	1 in 40
25	1 in 1,200	35	1 in 350	45	1 in 30
26	1 in 1,100	36	1 in 300	46	1 in 25
27	1 in 1,050	37	1 in 250	47	1 in 20
28	1 in 1,000	38	1 in 200	48	1 in 15
29	1 in 950	39	1 in 150	49	1 in 10

Whom to screen?

- Maternal age ≥35 years
- Past H/O Down syndrome

- Down syndrome (DS) or Down's syndrome, also known as trisomy 21, is a genetic disorder caused by the presence of all or part of a third copy of chromosome 21
- Down syndrome is the most common chromosome abnormality in human beings
- Prevalence is approximately 1:500

Take Home Message

I care....

Because birth defects affect all of us

AND

Because raising awareness can help babies around the world.

Acknowledgement

To

Prof Dr Gehanath Baral

Head of the Department in Obstetrics and Gynaecology

For inspiring us and organizing today's World Birth Defect Day Celebration

Thank You