CHROMOSOMAL ABNORMALITIES
Downs Syndrome

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What Are Chromosomal Abnormalities?

- Any irregularity in the number or gross structure will cause a disruption in the functioning of an individual.
- Autosomal chromosome abnormalities are present in up to 4 in every 1000 births.
- Sex chromosome abnormalities in 3 births per 1000.
- Within autosomal abnormalities approx 1.5 in 1000 are trisomies (mainly trisomy 21).
What Is trisomy 21?

- DOWNS SYNDROME
- This is the most common autosomal anomaly
- It is called trisomy 21 as there are three chromosomes at this position, so instead of having 46 chromosomes the baby has 47
Diagnosis

- Maternal blood taken at 12-15 weeks to measure alpha-fetoprotein (AFP) is reduced in a Down’s syndrome baby
- Ultra-sound scanning may find flat occiput, small head and typical facial features and a thickening at the back of the head. Heart and intestinal defect may be seen
- amniocentesis
Incidence

- 1 in 600-700 births (overall incidence)
- 1 in 1200 births (at age 20+)
- 1 in 100 births (at age 40+)
- 1 in 40 birth (at age 45+)
Clinical Features- Facial

- Upward slanting eyes
- Prominent epicanthic folds
- Protruding tongue
- Flat nasal bridge
- Brushfields spots
- Short neck
- Flat occiput
- Micrognathia
Clinical Features-limbs

- Short broad hands
- Short incurved little fingers
- Single parmer creases
- Increased risk of congenital hip dislocation
- Gap between first and second toes
Clinical Features-cardiovascular

- Atrial and ventricular septal defects and atrioventricular canal defects are the most commonest
- Cardiac defects are present in 40% of cases
Clinical Features-Gastrointestinal

- Gut stenoses and atresias (especially duodenal) are more common, including Hirschsprung’s disease
- Tracheoesophageal fistula
Clinical Features- Neuromuscular

- Hypotonia
- Feeding difficulties
- Motor delay
Management

- Inform parents as early as possible
- Confirm the diagnosis with chromosomal analysis
- Give parents information on prognosis and general management
- Genetic counselling as this can affect further attempts for children