Congenital Problems

April 20th 2022

Dr. Mikey Bryant
Introduction:

• Children are often born with congenital problems.
• Some comprise a range of conditions which can have an impact on the child’s development.
• Often multiple systems involved.
• It's important that families are informed about the condition of their child and offered some kind of management plan.
Down Syndrome:

- 1/600 births – surprisingly common!
- Incidence continues to increase as mother’s age increases.
- Caused by trisomy 21, usually due to maternal oogenesis.
- 2% mosaic with a normal cell line also produced – essentially leading to milder conditions.
Clinical Features: Facial:

- Usually noticeable from birth.
- Generalised hypotonia, often see an obvious head lag.
- Facial features remain very distinctive: small low-set ears, up-slanting eyes, prominent epicanthic folds, a flat facial profile and protruding tongue.
- Later will notice brushfield spots on the iris.
- Flat occiput (brachycephaly) with a short neck.
Down Syndrome:
Clinical Features: Remaining body:

- Limb features: short broad hands (brachydactyly), short curved little fingers (clinodactyly), single transverse palmar crease and a wide “sandal” gap between toes.
- Mildly short stature.
- Intellectual impairment with IQ between 25 and 70.
- Social skills often advanced, very friendly and personable.
- High levels of interaction ongoing
Associated Conditions:

• 40 – 50% have congenital cardiac defects, including AVSD, VSD, ASD, Fallot Tetralogy.
• GI problems such as duodenal atresia, anal atresia.
• Developmental hip dysplasia.
• Leukaemia risk increased (1% patients suffer).
• Deafness may be both conductive and sensorineural.
• Eczema
• Hypothyroidism.
Growth failure
Mental retardation
Flat back of head
Abnormal ears
Many "loops" on finger tips
Palm crease
Special skin ridge patterns
Unilateral or bilateral absence of one rib
Intestinal blockage
Umbilical hernia
Abnormal pelvis
Diminished muscle tone

Broad flat face
Slanting eyes
Epicanthic eyelid
Short nose
Short and broad hands
Small and arched palate
Big, wrinkled tongue
Dental anomalies
Congenital heart disease
Enlarged colon
Big toes widely spaced

Some clinical features
Diagnosis:

• Ideally should be carried out by chromosomal analysis, e.g. interphase FISH.
• This is often difficult to attain in our setting.
• As a result a clinical diagnosis is appropriate.
• However be wary of other conditions.
Management:

• Ensure as detailed cardiac assessment as possible, carry out echocardiography.
• Genetic counselling to ensure the whole family are aware of the risks moving forward.
• Long term follow-up to be carried out by multidisciplinary team including physiotherapy, nursing team etc.
• Monitor thyroid function tests.
• Annual ophthalmic reviews
Klinefelter’s Syndrome:

- Chromosomal abnormality is 47 XXY.
- Usually non-disjunction during maternal oogenesis.
- By mid-puberty the testes begin to involute and do not develop appropriately.
- Hypergonadotrophic hypogonadism with low testosterone is the hallmark.
- May be very tall but have a feminine body build.
- IQ marginally reduced.
- Usual presentation is later in life with infertility.
Tall stature
Slightly feminized physique
Mildly impaired IQ (15 points less than average)
Tendency to lose chest hairs

Frontal baldness absent
Poor beard growth
Breast development (in 30% of cases)
Osteoporosis
Small testes

Female-type pubic hair pattern
Management:

• Essentially not a curable condition.
• Needs to be explained that patients can lead a normal life with a good quality.
• Advise to avoid traditional fertility cures.
Patau Syndrome: Trisomy 13:

- 1/6000 births.
- May well be diagnosed on prenatal USS.
- Include:
  - Holoprosencephaly, microcephaly.
  - Cleft lip/palate.
  - Congenital cardiac diseases such as ASD, VSD etc.
  - Renal anomalies.
  - Severe mental retardation.
Management of Patau Syndrome:

- Children suffering from cleft lip and palate should be managed as such.
- Be aware of the high risk of aspiration pneumonias in these children.
- Ensure this is treated promptly.
- Education on safe feeding also essential.
Turner Syndrome:

- Girls with a single X chromosome.
- Short stature, wide carrying angle at elbows (cubitus valgus), widely spaced nipples, low posterior hairline, excessive pigmented naevi.
- Neonates often have puffy hands and feet.
- Commonly get congenital cardiac problems including co-arctation of the aorta, often with VSD.
- Commonly see renal agenesis or horseshoe kidney.
- May be subtle!
Turner Syndrome:

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

• Cyanosis.
• Heart failure signs or symptoms
• Murmur auscultated.
• Persistent respiratory distress with persistently low oxygen saturations despite oxygen therapy.
• Children often going place to place treated for “chest infections”.
• Faltering growth.
Heart Failure:

• Large left to right shunt e.g. large VSD.
• Left-sided obstructive lesions such as co-arctation of aorta.
• Later in life: Cardiomyopathy e.g. Hypertrophic obstructive cardiomyopathy.
• Myocarditis.
• Endocarditis.
• Rare: Tachy-arrhythmias.
Clinical Features:

• Sweating (increased compensatory sympathetic drive).
• Breathlessness, tachypnoea, creps heard on lungs despite constant antibiotic/bronchodilator treatment.
• Poor feeding in infants.
• Poor weight gain.
• Cardiomegaly, also tachycardia with a gallop rhythm.
• Rarely the same as adults!
Categorisation: Acyanotic lesions:

- VSD
- ASD
- Patent Ductus Arteriosus.
- Pulmonary Valve Stenosis.
- Coarctation of the aorta.
- HOCM
Categorisation: Cyanotic lesions:

- Tetralogy of Fallot.
- Transposition of the Great Arteries.
- Tricuspid Atresia.
- Total anomalous pulmonary drainage.
Types of murmurs and approach:

• Murmurs not always easy to pinpoint or diagnosis easy to reach exactly using auscultation alone.
• Continuous murmur:
• Long systolic murmur:
• Ejection systolic murmur:
Types of murmur and approach:

• Murmurs not always easy to pinpoint or diagnosis easy to reach exactly using auscultation alone.
• Continuous murmur: e.g. machinery: patent ductus arteriosus.
• Long systolic murmur: VSD
  Tetralogy of Fallot.
• Ejection systolic murmur: ASD/AVSD

  Pulmonary stenosis
  Coarctation of aorta
Management:

- Ultimately most of these conditions require surgery.
- However initial management of heart failure includes:

  - Oxygen therapy (or CPAP if needed).
  - Sit up at 45 degrees.
  - Consider low dose diuretics.
  - Antibiotics appropriate as needed if concomitant infection.
Trisomy 18 (Edward’s Syndrome)

- unusually small head
- back of the head is prominent
- ears are malformed and low-set
- mouth and jaw are small (may also have a cleft lip or cleft palate)
- hands are clenched into fists, and the index finger overlaps the other fingers
- Clubfeet (or rocker bottom feet) and toes may be webbed or fused
Trisomy 18:

• Similar cardiac defects to above
• Managable with accurate diagnosis and good quality care.
• Can consider physical therapy options.
• Further types of cardiac disease to be covered in the next session!