

CONGENITAL HEART DISEASE & GENETIC CONDITIONS

Lorna Carruthers
ACHD Nurse Specialist
Freeman Hospital

Aims of session

- Brief overview genetic conditions often associated with CHD
- How these genetic conditions can effect patients
- How the conditions are linked to congenital heart disease

Trisomy -21

- Additional chromosome - namely 21
- Varying intellectual levels
- Small skull
- Upward slant of eyes
- Nose is small with the flat nasal bridge
- Mouth has a narrow short palate with small teeth and furrowed protruding tongue
- Ears are small and dysplastic
- Hands are short and stubby
- Single crease on the hand (simian crease) at birth
- Delayed development and behavioral problems
- Cognitive disability

Trisomy 21 - CHD

- 50% Down syndrome children have CHD
- Atrioventricular Septal Defects (AVSDs) – These are the most common in children with Down syndrome.
- Ventricular Septal Defects (VSDs)
- Atrial Septal Defects (ASD's)
- Patent Ductus Arteriosus (PDA's)
- Tetralogy of Fallot (TOF)

Marfans Syndrome

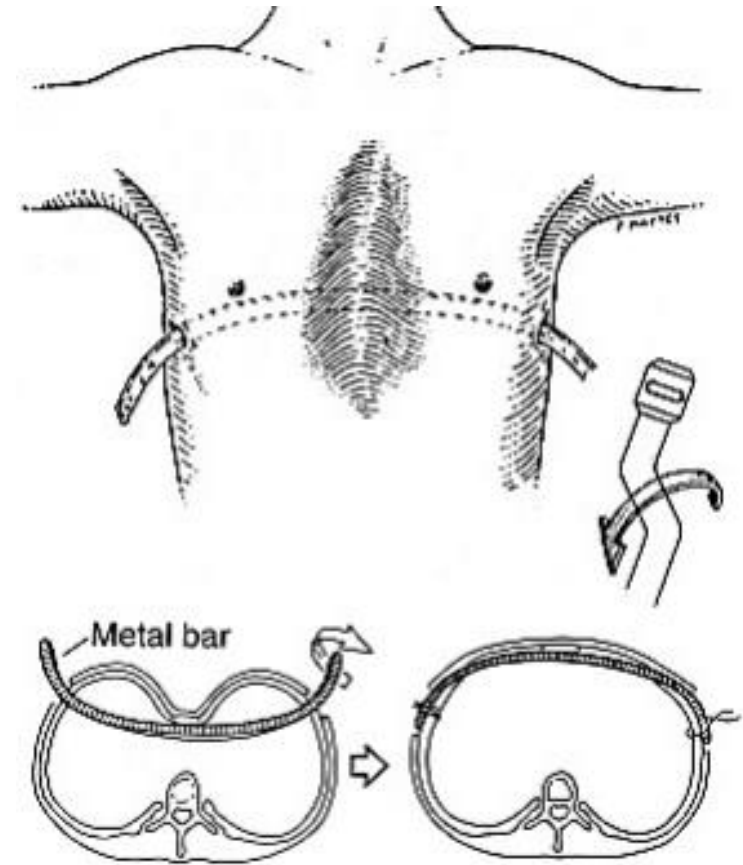
- A genetic disorder that affects connective tissues.
- Common (More than 20,000 cases per year in UK)
- Body produces protein making tissue 'extra stretchy'
- Long arms, legs and fingers
- Tall and thin body type
- Curved spine
- Chest sinks in or sticks out
- Flexible joints
- Flat feet
- Crowded teeth
- Stretch marks on the skin that are not related to weight gain or loss



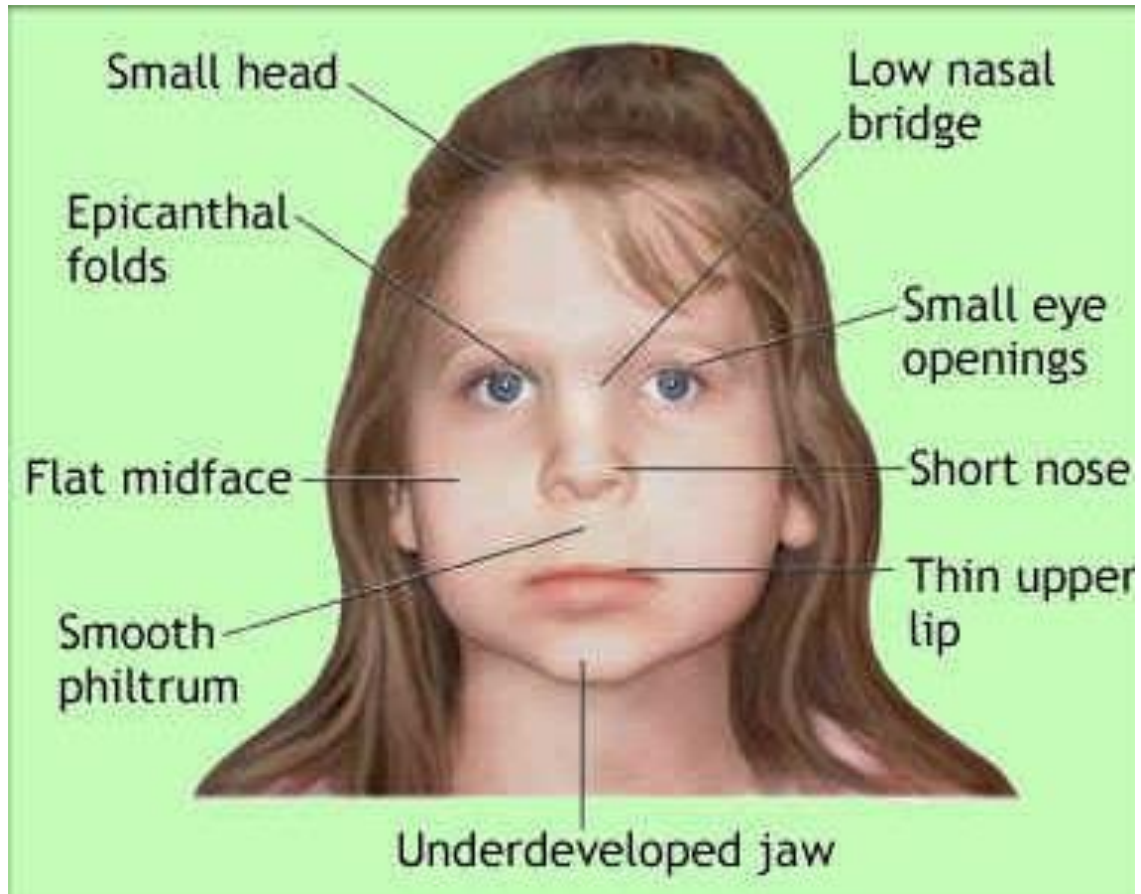
Marfans Syndrome - CHD

- **Aortic aneurysm** - due to stretchy connective tissue it is common for 'bulging' to happen, commonly found at the aortic root.
- **Aortic dissection** - Due to a tear in the aortic tissue wall, this can present as severe back or chest pain. When a dissection happens, it causes a weakness in the wall structure, it can rupture or depending on the origin of the dissection it can be medically managed.
- **Valve malformations** - People who have Marfan syndrome can have weaker tissue than normal in their heart valves. This can produce stretching of the valve tissue and abnormal valve function. When heart valves don't work properly, your heart often has to work harder to compensate. This can eventually lead to heart failure
- **Chest Malformations** - Pectus excavatum, a 'depression' of the sternum. Can have a 'Nuss procedure'

NUSS PROCEDURE



Di George Syndrome -22q11.2 deletion



- Caused by the deletion of a small segment of chromosome 22.
- Low set ears
- congenital heart problems
- specific facial features
- Frequent infections
- Developmental delay- varying degree
- Learning problems
- Cleft palate

Di George Syndrome

- Heart Murmurs
- Aortic regurgitation
- Ventricular septal defects
- Tetralogy Fallot

What is Ehlers Danlos Syndrome?

Individuals with EDS have a defect in their connective tissue, the tissue that provides support to many body parts such as the skin, muscles and ligaments. The fragile skin and unstable joints found in EDS are the result of faulty collagen. Collagen is a protein, which acts as a "glue" in the body, adding strength and elasticity to connective tissue

Signs & Symptoms

Symptoms vary widely based on which type of EDS the patient has. In each case, however, the symptoms are ultimately due to faulty or reduced amounts of collagen. EDS typically affects the joints, skin, and blood vessels.

Pain	Fatigue	Prolapse
Dislocations	Chiari	Preterm labor
Subluxations	Sprains	IBS
Hypermobility	Gastrointestinal issues	Dysautonomia
Osteoarthritis	Atrophic scarring	Flat feet
Osteoporosis	Muscle spasms	Swan neck deformity
Skin Tearing	Poor healing	Degenerative Joint Disease
Stretchy skin	TMJ	Gastritis
Soft skin	POTS	Arthralgia
Mitral Valve Prolapse	Organ rupture	Myalgia
Easy bruising	Aneurysms	Surgical complications

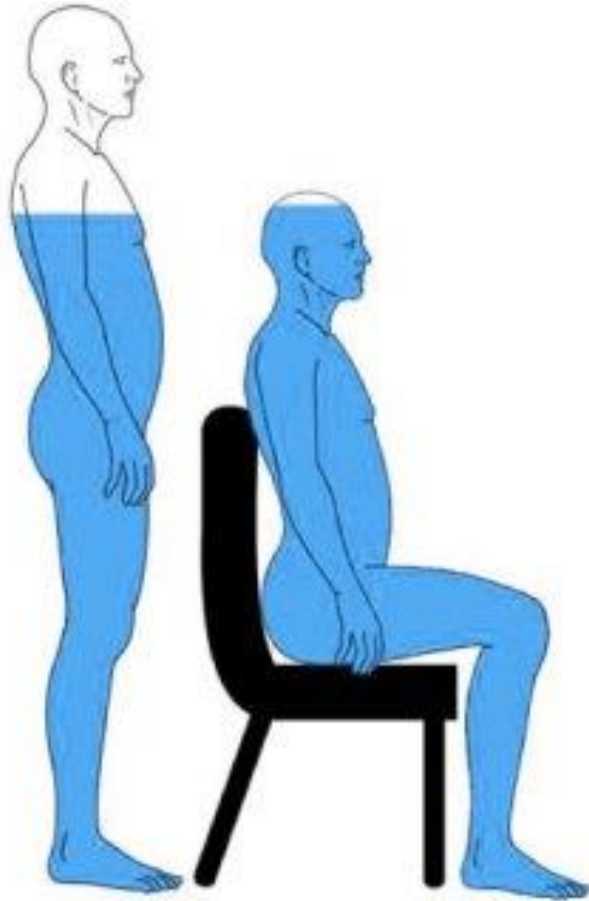
FB you know you have EDS when



Ehlers Danlos -CHD

- Vascular EDS
- Scar tissue
- Weakened aortic tissue
- Rupture/dissection risk
- POTS Syndrome

Postural Orthostatic Tachycardia Syndrome



Symptoms



Dizziness



Sweating



Fatigue



Shortness
of Breath



Chest Pains
& Heart
Palpitations

How To Treat It



Eat Smaller
Meals & Fewer
Carbohydrates



Increase
Fluid Intake



Avoid Caffeine



Increase
Salt Intake



Avoid
Prolonged
Standing

Loeys Dietz syndrome

- Genetic connective tissue disorder
- Similar to Marfans
- Similar to Ehlers Danlos
- Weakened connective tissue
- Aortic aneurysms
- Aortic dissections
- Early detection essential in paediatric patients



Turners Syndrome

- Also Known as 45X, 45 X0
- Females is missing X Chromosome
- Classical / Mosaic
- Webbed neck, Short stature, swollen hands, swollen feet
- Underdeveloped ovaries - infertile

Turners -CHD

- Skeletal abnormalities
- Muscular abnormalities
- Aortic problems- dilatation
- Regular surveillance - imaging

Didn't mention.....

- Noonan's
- Williams
- Shones complex

QUESTIONS??????