CARIS

Congenital Anomaly Register and Information Service

Annual Meeting 2024

Virtual Session

GIG CYMRU NHS WALES | Iechyd Cyhoeddus Cymru Public Health Wales

CARIS Annual Meeting 2024 virtual session	
Date	**Tuesday November 26th 2024**
Time	**12.00 - 14.00**
Venue	**MS Teams**

12.00	**Welcome**	*Llion Davies* *Public Health Wales*
12.05	**CARIS update**	*David Tucker* *Public Health Wales*
Focus session: Congenital Cardiac Anomalies Chair: Llion Davies		
12.15	**Covid-19 and congenital anomalies**	*Penny Cresswell-Jones* *Public Health Wales*
12.30	**Detection and care of cardiac anomalies**	*Joyce Lim* *Consultant Paediatric Cardiologist*
12.50	**Paediatric management of cardiac defects**	*Rajesh Viswanathan* *Paediatric Consultant*
13.10	**Screening for congenital heart disease**	*Bill Taylor*
13.30	**Improving the early detection and care of babies with serious heart conditions**	*Tiny Tickers*
13.40	**Epidermolysis bullosa**	*John Helo* *Cardiff University / CARIS*
13.50	**Questions and closing remarks**	
14.00	**Close**	

Welcome

Dr Llion Davies

Public Health Wales

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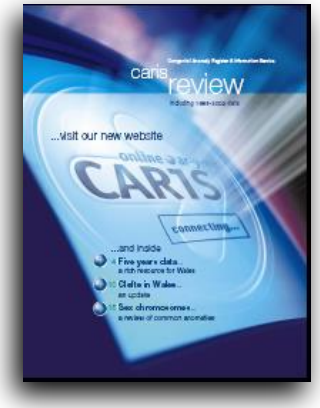

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Public Health Wales

CARIS Team Report

David Tucker

Public Health Wales

Official Statistics Update: 1998-2023

- Congenital Anomalies
  - 40,261 cases registered
  - 4.8% of all births
  - 84.5% liveborn, with 96.9% surviving to 1 year
- Childhood Rare Diseases
  - 381 diseases with published rates
  - Almost 1500 individual rare diseases registered
  - Data on commoner conditions published at LHB level
- Antenatal Detection rates

Team Achievements

- Data expansion (Syphilis and HIV)
- Part of a 4 nations network for rare disease
- Working closely with NHS Exec
- Student placements
- WPSU is now part of the CARIS Team

Adult Rare Diseases

- Alpha-1-antitrypsin, Fabry disease
- Bespoke database now running

Publications

Morris, Joan K., Maria Loane, Charlotte Wahlich, Joachim Tan, Silvia Baldacci, Elisa Ballardini, Clara Cavero-Carbonell et al. "**Hospital care in the first 10 years of life of children with congenital anomalies** in six European countries: data from the EUROlinkCAT cohort linkage study." *Archives of disease in childhood* 109, no. 5 (2024): 402-408.

Urhoj, Stine Kjaer, Joan Morris, Maria Loane, Elisa Ballardini, Laia Barrachina-Bonet, Clara Cavero-Carbonell, Alessio Coi et al. "**Higher risk of cerebral palsy, seizures/epilepsy, visual-and hearing impairments, cancer, injury and child abuse in children with congenital anomalies**: Data from the EUROlinkCAT study." *Acta paediatrica* 113, no. 5 (2024): 1024-1031.

www.eurolinkcat.eu

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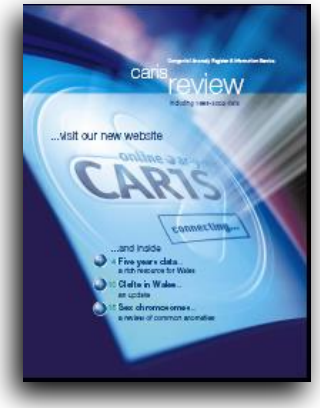

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Public Health Wales

Is maternal COVID-19 infection associated with congenital anomalies in Wales?

Penny Cresswell-Jones

Public Health Wales

Is maternal COVID-19 infection associated with congenital anomalies in Wales?

Penelope Cresswell-Jones
Specialty Registrar in Public Health, Public Health Wales

Llion Davies, Samantha Fisher, David Tucker, Margery Morgan

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Public Health Wales

26.11.24

This presentation will cover:

- What is already known?
- What we hoped to find out
- How we approached the data
- What we found
- What this means

What is already known?

Other infections associated with congenital anomalies

Studies looked at COVID-19 in pregnancy in early pandemic

Small sample sizes and limited first trimester evidence

No clear association between COVID-19 and congenital anomalies

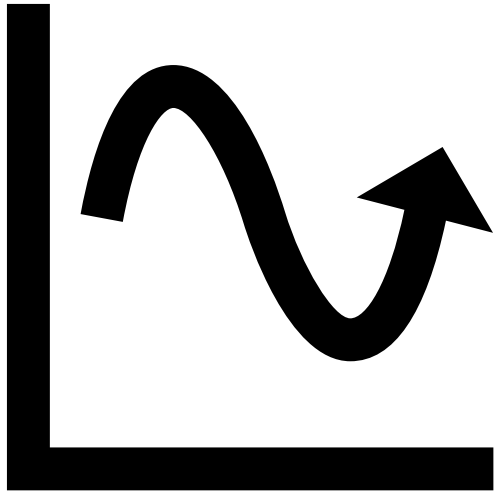
What we hoped to find out

Was maternal COVID-19 infection associated with congenital anomalies in Wales between 2020-2022?

Analysis of the Wales congenital anomaly register (CARIS) data

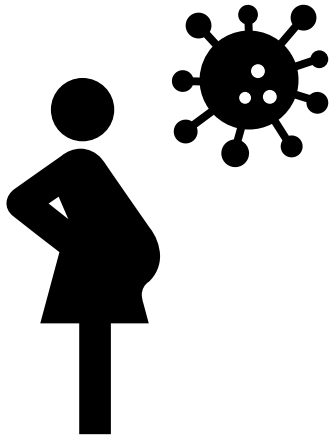
How we approached the data

1. Trends over time before and during the pandemic



CARIS official annual statistics

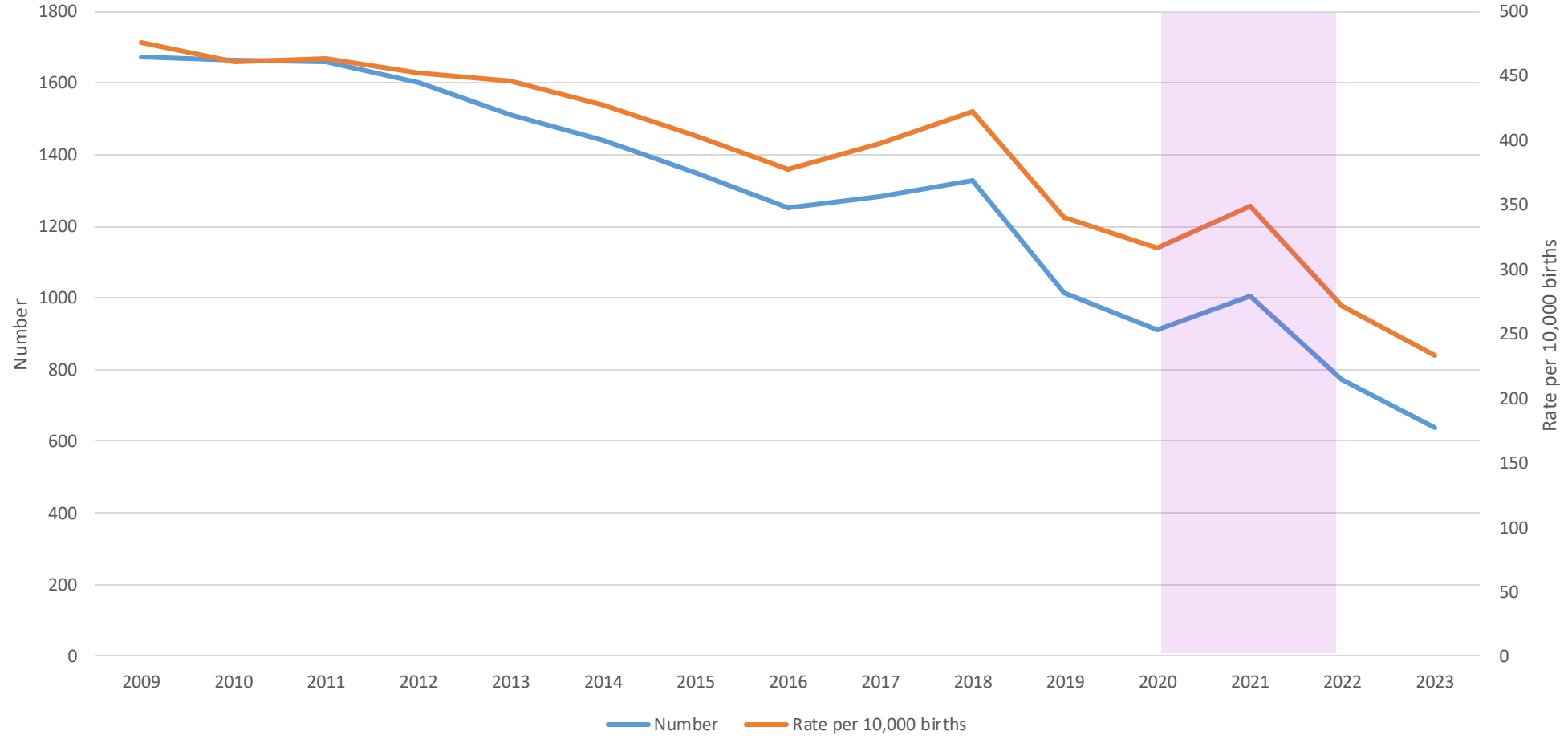
2. Sample of women who had COVID-19



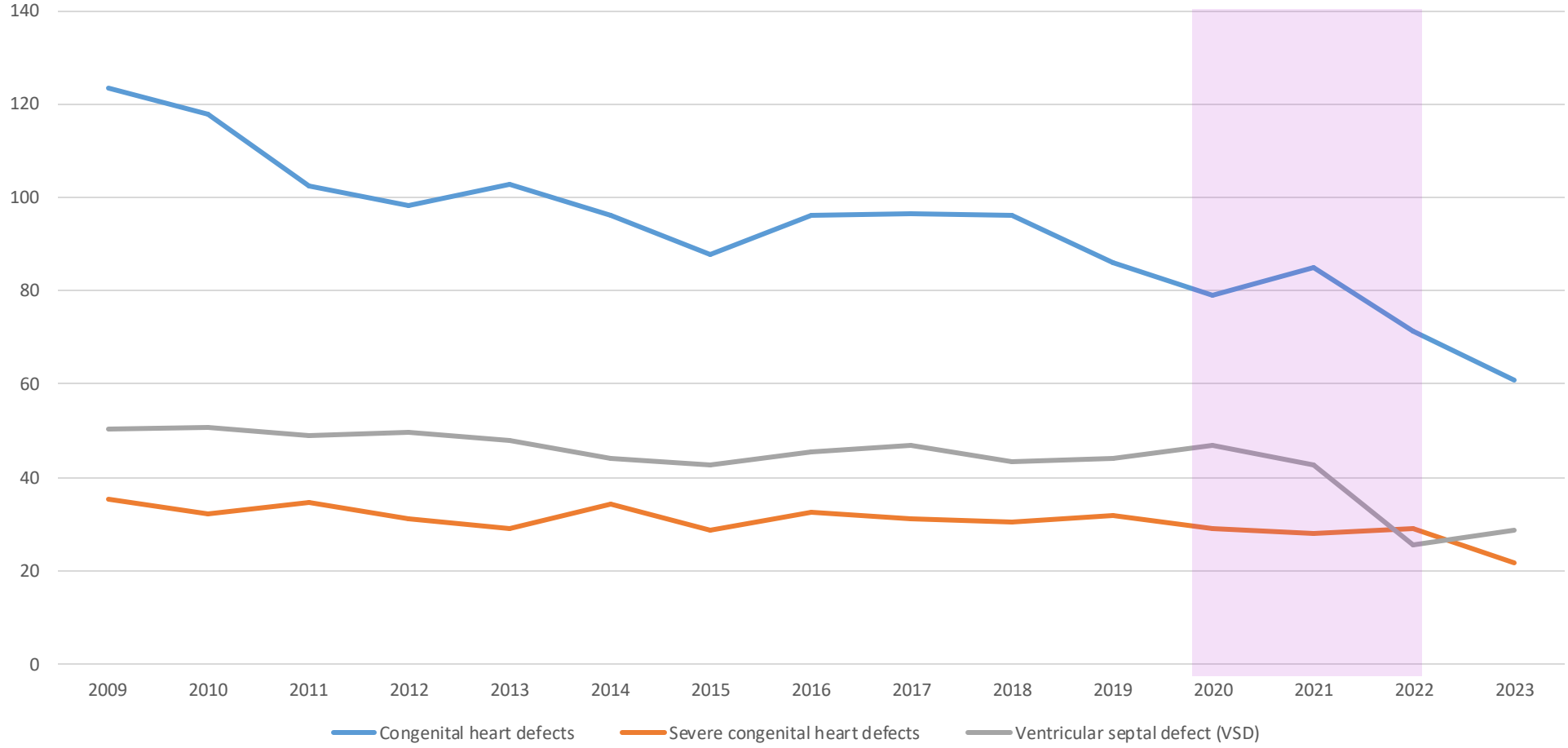
486 women on CARIS with lab-confirmed SARS-CoV-2 (COVID-19)

What we found – Trend data

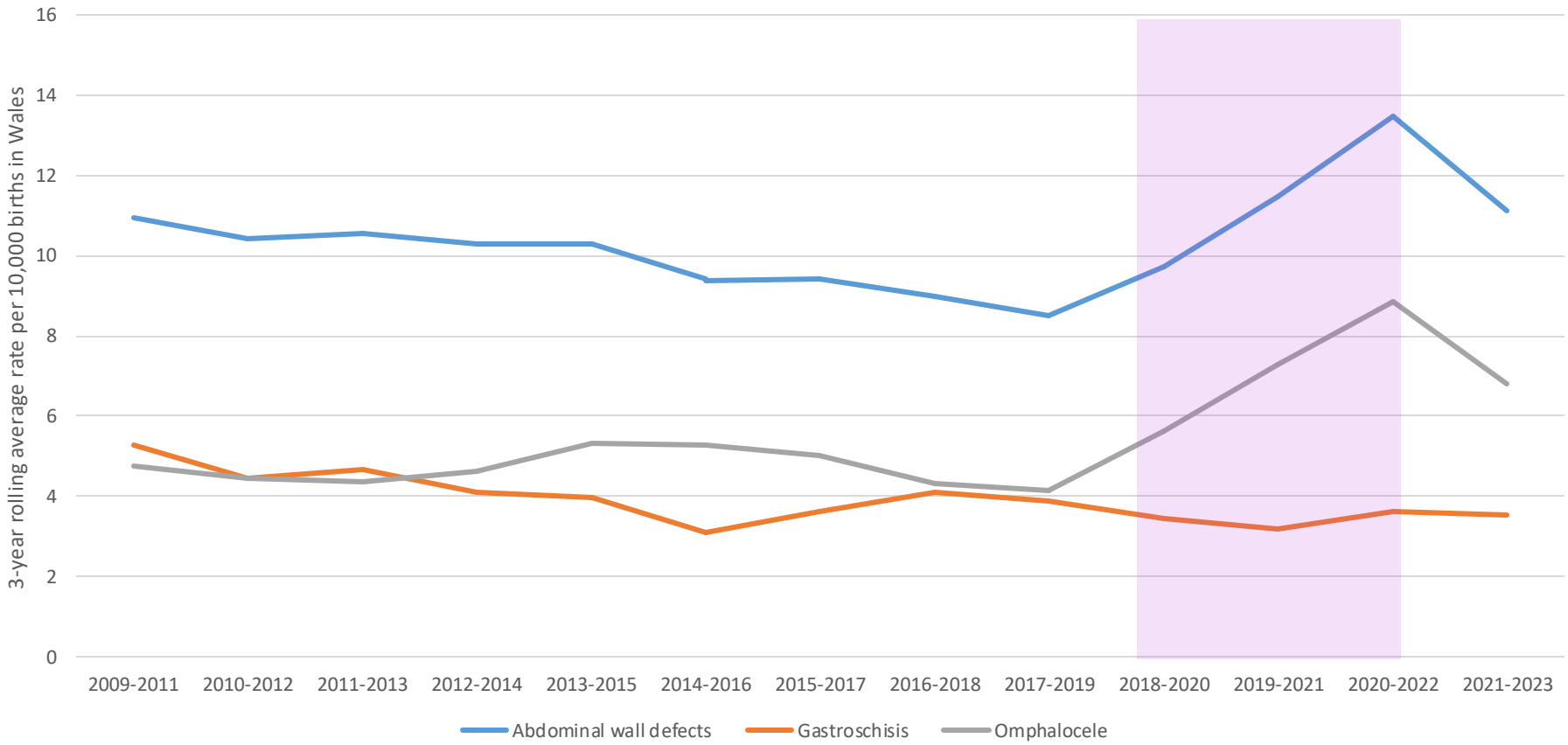
The overall number and rate of congenital anomalies were not greater than pre-pandemic levels



Most types of anomalies in Wales showed similar trends by rate per 10,000 births



Abdominal wall defects (exomphalos/omphalocele) had higher rates in 2020-2022, but the reason for this is unclear

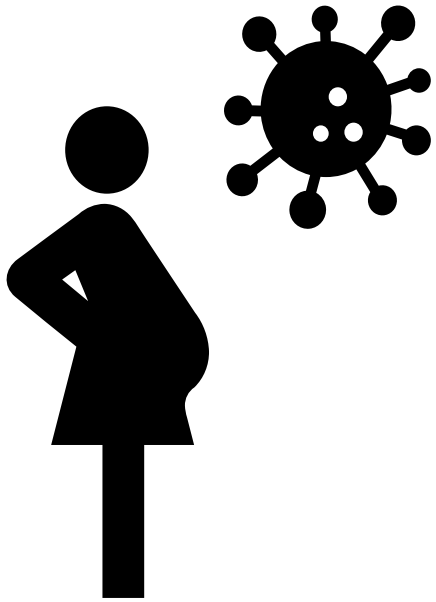


Limitations to using trend data

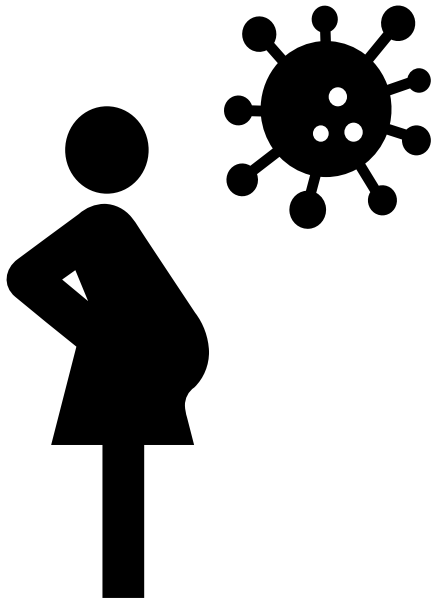
We cannot determine cause

Many potential factors which could influence findings

What we found – Sample



	COVID-19			
	1st trimester	2nd trimester	3rd trimester	Total
Anomaly	41	73	57	171
No anomaly	60	104	151	315
Total	101	177	208	486



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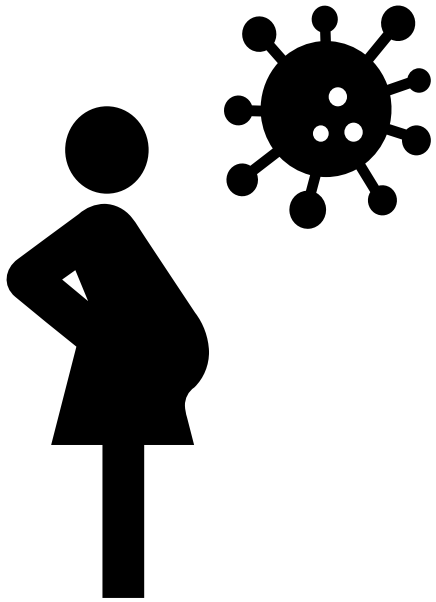

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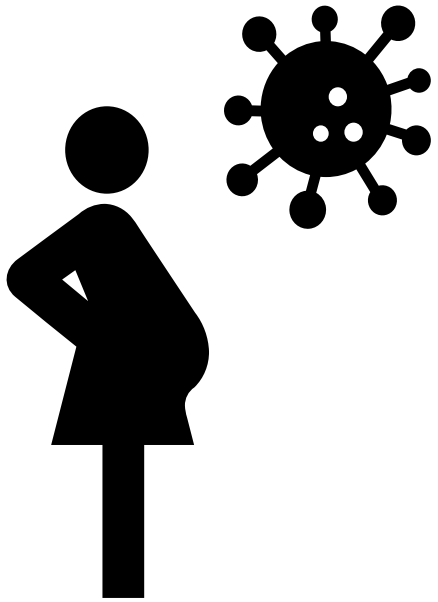

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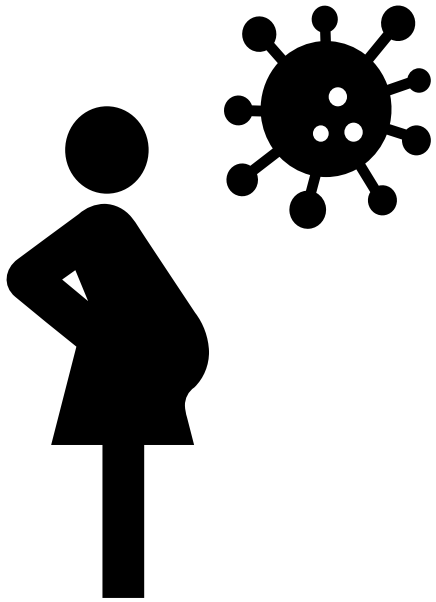

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The most frequently reported anomalies in the sample were similar to pre-pandemic

Congenital hydronephrosis including ureter obstruction	35
Ventricular septal defect	20
Talipes equinovarus	16
Hypospadias	12
Multi-cystic renal dysplasia	6
Tetralogy and pentalogy of Fallot	5
Atrial septal defect	5
Cleft lip with/without palate	5

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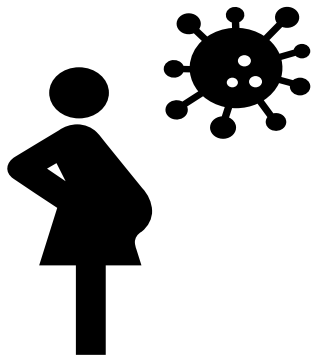
Tetralogy and pentalogy of Fallot	5
Atrial septal defect	5
Cleft lip with/without palate	5
Exomphalos (Omphalocele)	**<5**

Odds of congenital anomaly were not associated with trimester of COVID-19



COVID-19	Adjusted odds ratio	95% CI for adjusted odds ratio	P-value
First trimester	0.654	0.186 – 2.296	0.508
Second trimester	0.838	0.329 – 2.135	0.711
Third trimester	Reference		

Limitations to sample data

- Small sample size
- Reporting and recording lag
- Did not capture whole population of pregnant women with COVID-19

No clear evidence of association between maternal COVID-19 and congenital anomalies in Wales

What this means

Ongoing vigilance is needed

- No clear pattern around COVID-19 and congenital anomalies has emerged
- Associations with rare conditions may need international detection
- Clinicians and CARIS to remain vigilant

Shared learning

- Challenges on staff capacity during the pandemic
- Highlights importance of data linkage
- Opportunities for proactive reporting/recording where a new infection or exposure emerges

Diolch yn fawr.
Thank you

Penelope Cresswell-Jones - Penelope.Cresswell-Jones@wales.nhs.uk

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19.11.24

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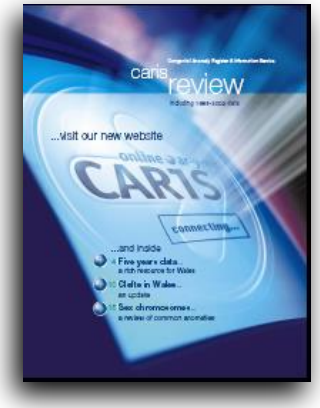

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CARIS *Congenital Anomaly Register and Information Service*

Focus session: Congenital Cardiac Anomalies

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Public Health Wales

DETECTION AND CARE OF CHD PATIENTS

CARIS MEETING
(VIRTUAL MEETING 26/11/2024)

DR JOYCE LIM

Consultant Fetal and Paediatric Cardiologist

Alder Hey Children's Hospital & Liverpool Women's Hospital, Liverpool

Liverpool Women's NHS Foundation Trust

Alder Hey

Congenital Heart Network North West | North Wales | Isle of Man

Overview

- Background of CHD
- FASP targets & NSCARDS data
- Antenatal/Perinatal management

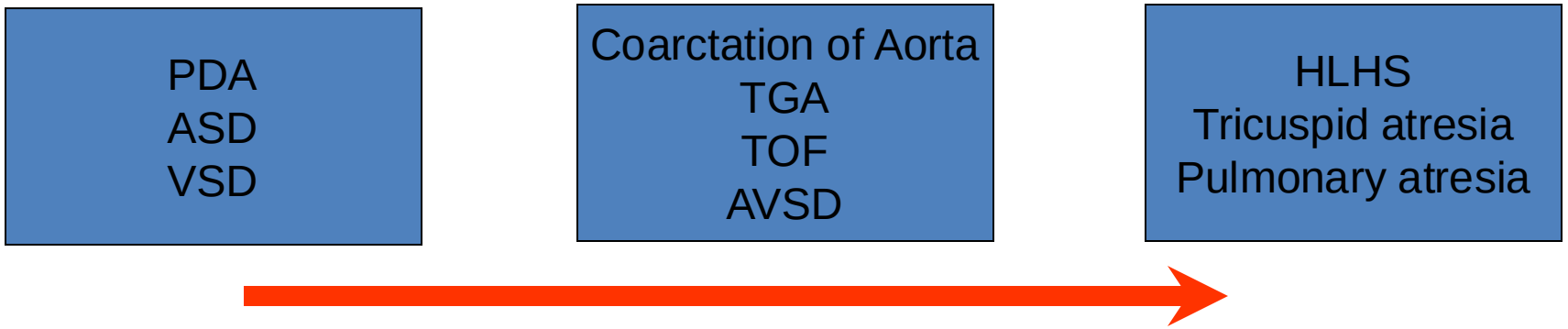
Background

- **Epidemiology of CHD**

  Live-births - 4-11/1000 (0.4-1%)

  Foetuses - higher percentage

- **Spectrum of congenital cardiac anomalies**



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Congenital cardiac anomalies

Difference in prevalence antenatal and postnatal population

	Antenatal	Postnatal
Hypoplastic left heart	17%	7%
Univentricular heart	15%	3%
Atrioventricular septal defect	12%	7%
Double outlet RV small LV	12%	4%
Aortic stenosis	5%	3%
Ebstein's anomaly	5%	2%

Congenital cardiac anomalies

Cause / Pathogenesis

- Chromosomal defects (6% of all CHD but now with WES and WGS, incidence might be higher)
- Multiple genetic abnormalities / part of syndrome (eg: Holt-Oram, VACTERL)
- Environmental factors (eg: infection, maternal disease, toxins)
- "unknown" cause / pure bad luck

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NCARDRS/FASP & NICOR/NCHDA

NCARDRS – National Congenital Anomaly and Rare Diseases Registration Service

NCHDA – National Congenital Heart Disease Audit

NCARDRS/FASP vs NICOR/NCHDA data

NCARDRS/FASP data

- offer a 20-week screening scan to screen for 11 physical conditions. This ultrasound scan is undertaken between 18+0 and 20+6 weeks of pregnancy. The screening pathway must be completed by 23+0 weeks of pregnancy
- Data taken of all fetuses with EDD from 1 April to 31 March of the year
- Specific cardiac screen - Looking at 4 (5) cardiac anomalies (TGA, AVSD, Tetralogy, HLHS, Coarctation of Aorta)
- Updated targets from April 2022

	Pre 2022 targets	Acceptable	Achievable
TGA	≥ 50%	≥ 70%	≥ 99%
AVSD	≥ 50%	≥ 50%	≥ 80%
Tetralogy of Fallot	≥ 50%	≥ 55%	≥ 85%
HLHS	≥ 50%	≥ 80%	≥ 99%
Coarctation of aorta	Not measured	Not set	Not set

https://www.gov.uk/government/publications/fetal-anomaly-screening-programme-standards/fetal-anomaly-screening-standards-valid-for-data-collected-from-1-april-2022

NICOR/NCHDA data

- Data collected from the intervention/surgical perspective – all types of cardiac anomaly which required intervention before age 1 year
- Uses PPD (Procedures with Prenatal Diagnosis) data
- Data collected between 1 April to 31 March following year

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NICOR (NCHDA) DATA

Over half of all procedures in infancy have a successful prenatal diagnosis

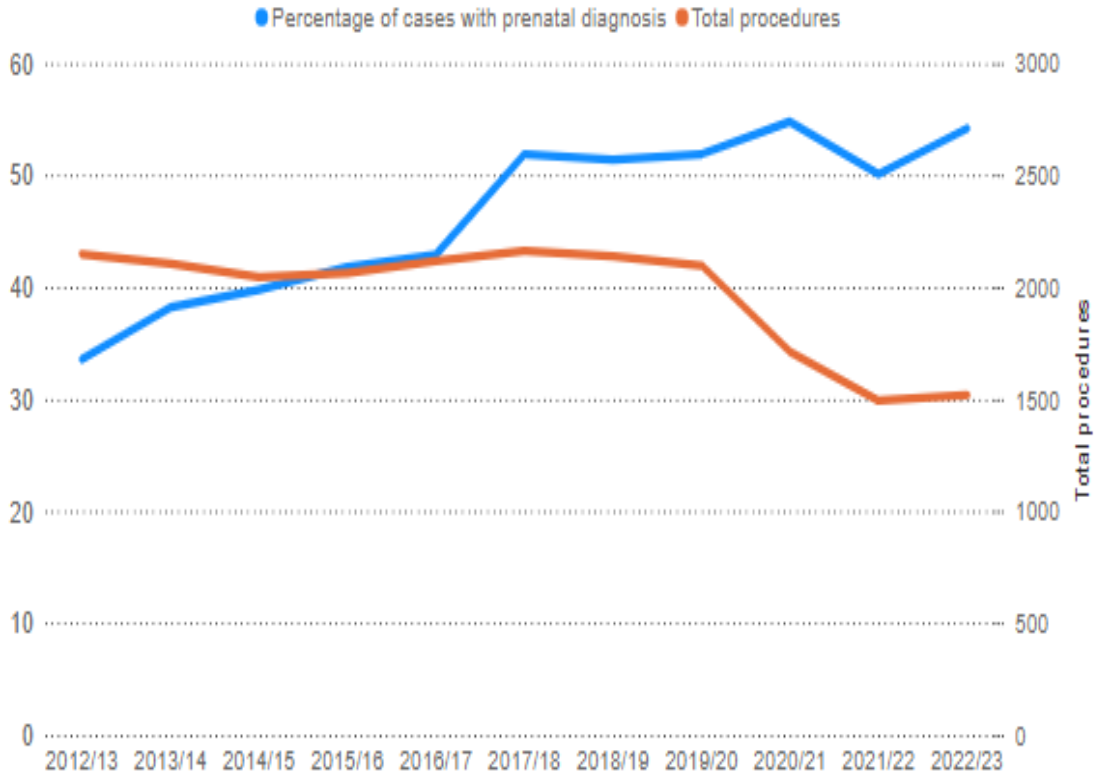
The audit collects data for babies diagnosed antenatally with a cardiac defect who then undergo an intervention in the first year of life.

In 2022./23, the detection rate was 54.2% for all infants requiring a procedure in the first year of life (up from 50.1% in 2021/22).

Over the last six years, the rate of antenatal diagnosis has plateaued at just over 50%.

Note: the data excludes spontaneous intrauterine deaths, termination of pregnancy, non-intervention after birth and unrecognised death in community or non-tertiary centre. For additional information, see here.

Percentage of CHD procedures in the first year of life for infants that had a prenatal diagnosis



Prenatal diagnosis of lesions has improved for some types but worsened for others

The figure shows the prenatal detection rate of four individual cardiac lesions where a procedure is performed in the first year of life:

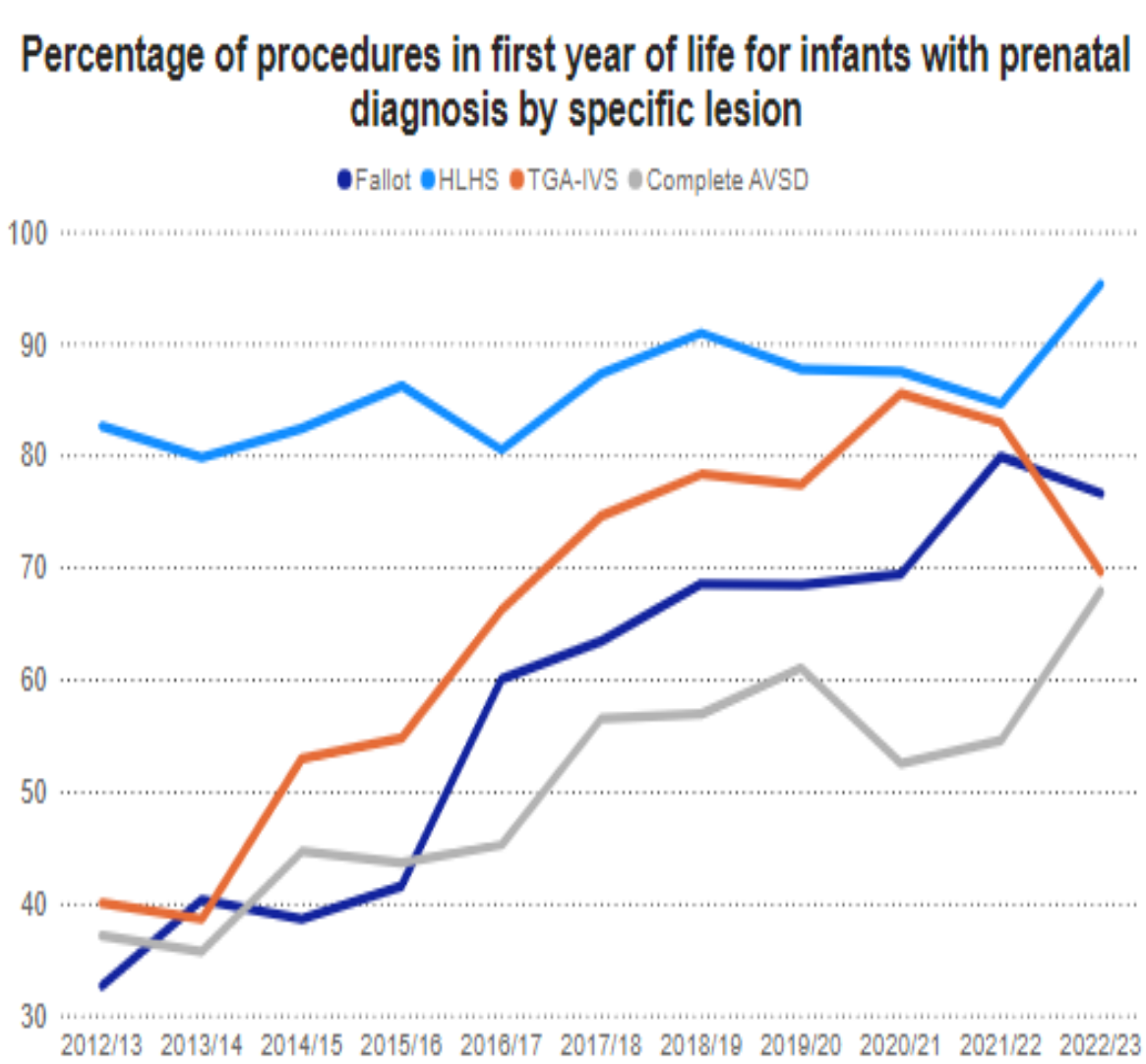
- Fallot's tetralogy
- Hypoplastic left heart syndrome (HLHS)
- Transposition of the great arteries with an intact intraventricular septum (TGA-IVS)
- Atrio-ventricular septal defect (AVSD)

For those requiring a procedure in the first year, the prenatal detection of HLHS was highest in 2022/23 at 95%. There was also an improvement in diagnosis of complete AVSD.

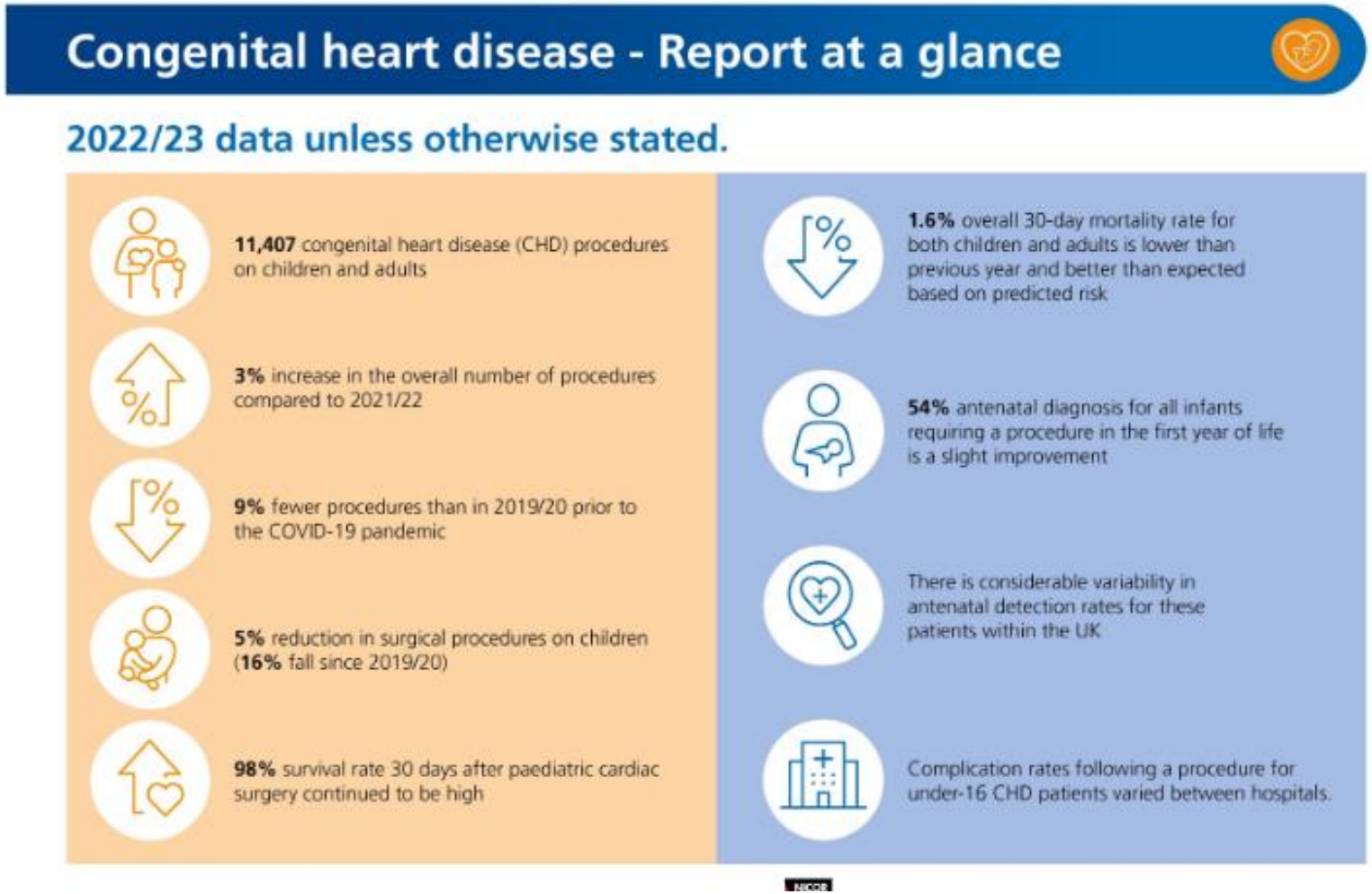
Conversely, prenatal diagnostic rates for Fallot's tetralogy and TGA-IVS both deteriorated. This could represent a genuine fall in detection or might, for example, relate to an increase in the termination of pregnancy for these diagnoses. Linkage with the National Congenital Anomaly and Rare Disease Registration Service would help clarify such issues.

Differences in screening timing and methods used by sonographers may also drive variations in prenatal diagnosis.

A full table is available here (link to QI document).

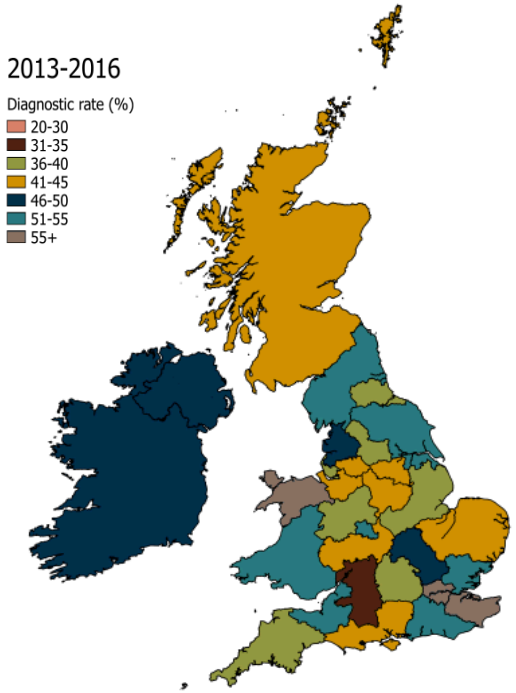


NICOR/NCHDA data (2022/2023)

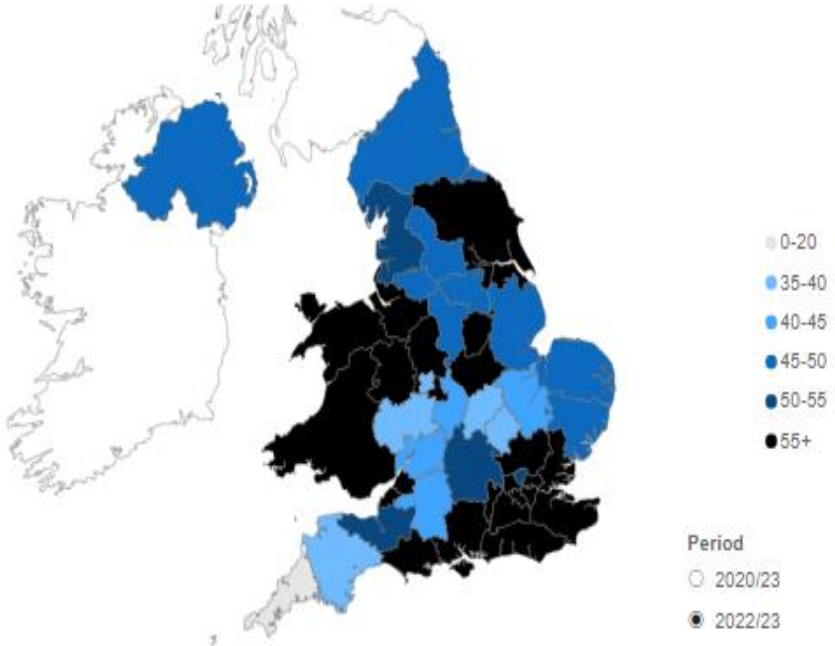


Source: NICOR Annual Report 2024 (published 10/4/2024)

NICOR 2022/23 - Percentage of Prenatal Dx for CHD procedures in 1st year of life



2013-2016



2022-2023

Covers

- 42 Integrated Care Boards (ICB) in England
- 7 University Health Board (Wales)
- Northern Ireland

For 2022/23 – highest rate of 72% prenatal dx by Leicester, Leicestershire and Rutland ICB

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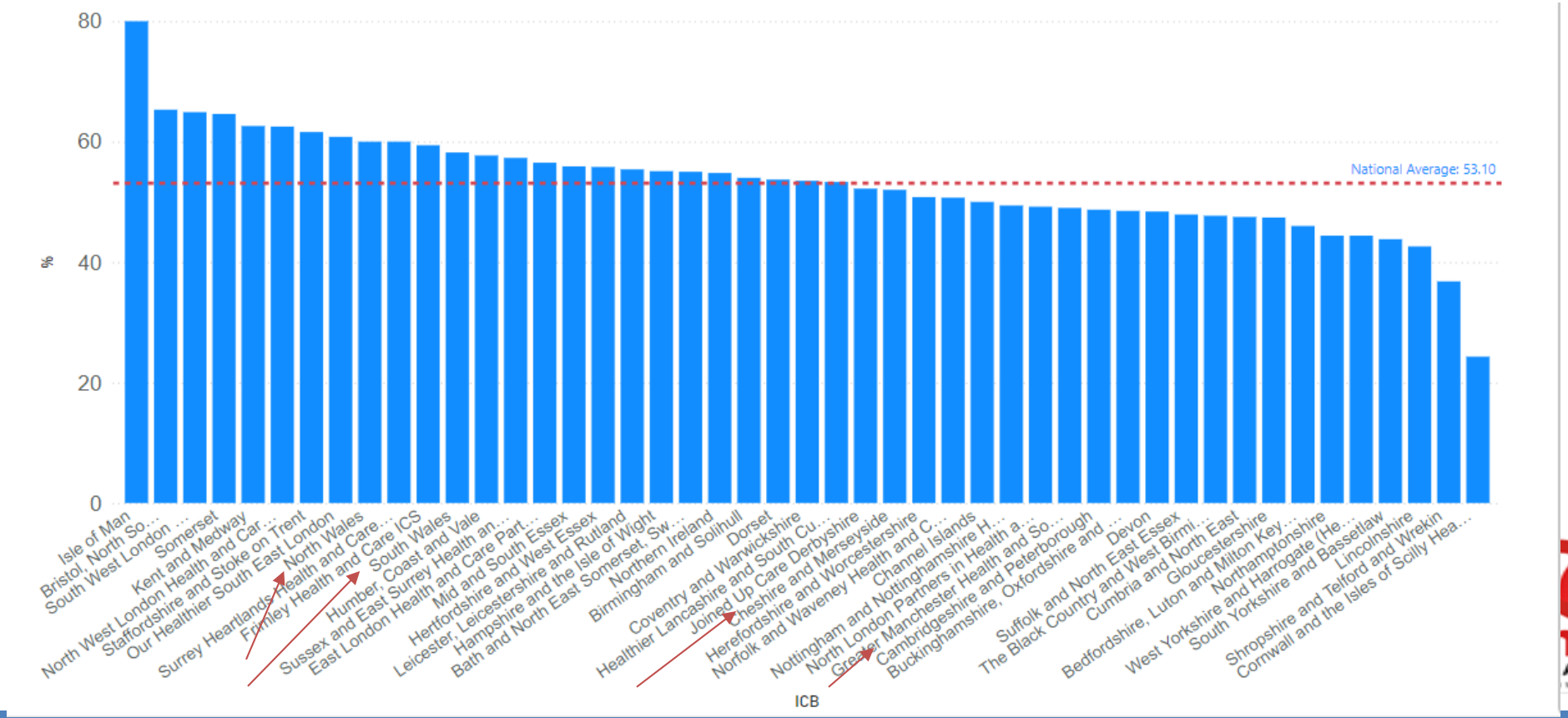
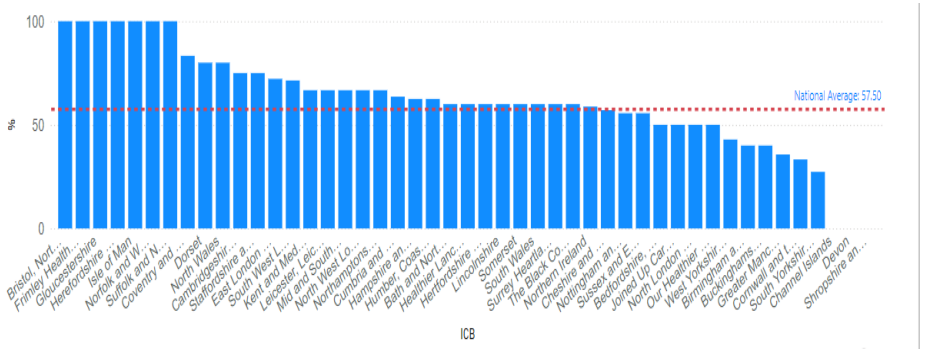
ALL (AVSD, HLHS, TGA, Tetralogy) – 2022/2023

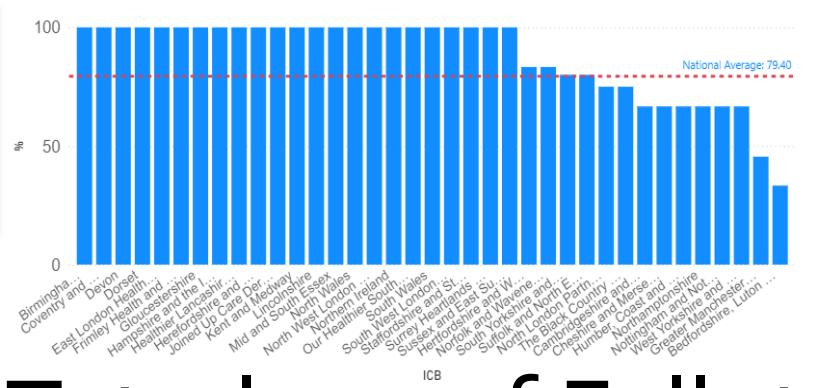


NICOR 2022/2023 data

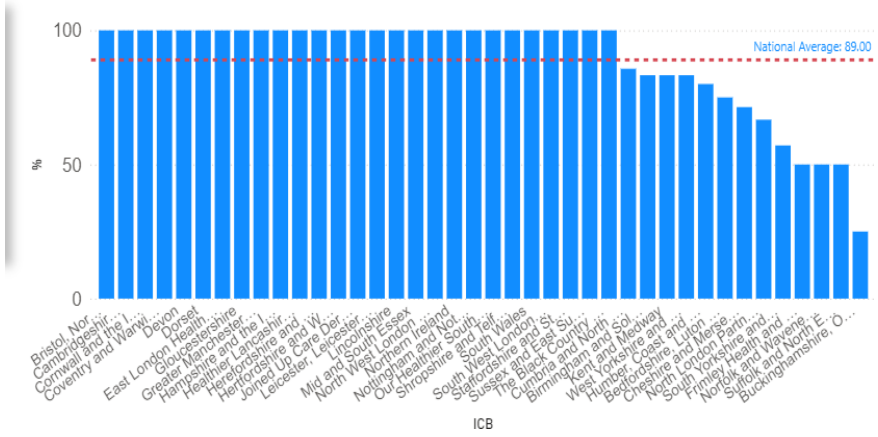
AVSD



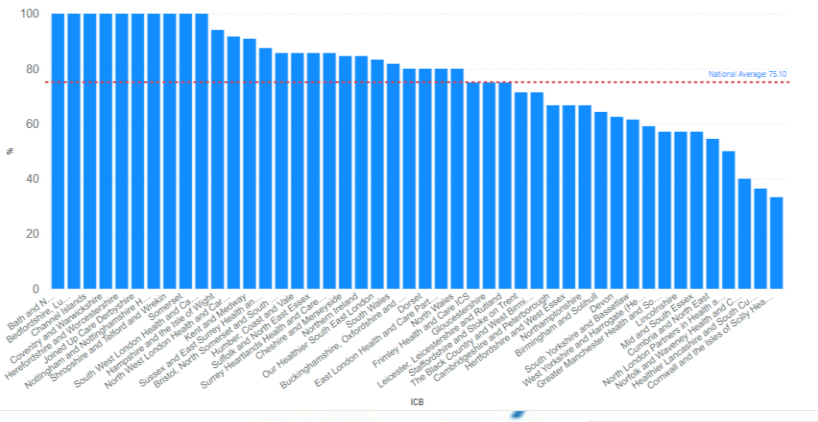
TGA



HLHS



Tetralogy of Fallot



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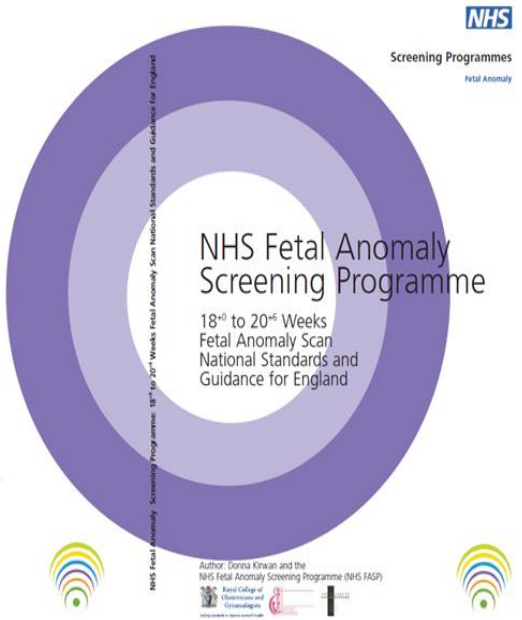
We are dependant on sonographer pick up at anomaly scan

- 5 standard fetal cardiac views
- Increased referrals for abnormality since 3VT
- PHE will also monitor outcomes for 4 (5) major heart lesions via NCARDRS



Challenges to improving detection in 'low risk' pregnancies

- Training
  - Vast numbers of sonographers performing 20 week scan with high turnover
  - Huge variability across region and within centres
  - Hands on training (Tiny Tickers, NW Congenital Heart Network, other courses)
- Time
  - 58% allow 15-20 minutes, 33% allowing 20-30 minutes
  - 9% of units only allow 10-15 minutes
- Equipment
  - 65% of units reported they have sufficient needs for their workload and 61% provided adequate image quality
- Innovation
  - AI-assisted technology
  - Simulation

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Benefits of Prenatal diagnosis

- Parents are better prepared for postnatal course
  - Planned delivery
  - Allows planning for family logistics
  - Time to consider options

- Baby

- Allows for option for fetal intervention (limited)

- perinatal plan improves clinical status of baby immediately postnatally (less likely to present in extremis); smoother clinical course

- Stable baby = better surgical/interventional outcome – short, medium, longer term. Less co-morbidity

- Local teams

– better prepared/ know what to expect

- Allows for planning

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Management

Alder Hey Children's NHS
NHS Foundation Trust

Diagnosis

↓

Counselling

(Further investigations, treatment, options explained, cardiac liaison sister)

↓

Follow-up review

(evolution of lesion)

↓

Planned delivery

(issues with prematurity, size)

↓

Postnatal review and management

IUD / Termination

Active Management

Compassionate Care

Fetal Cardiac Counselling

- Making the diagnosis
- Attempting to predict potential clinical course antenatally
- Predicting possible postnatal clinical course antenatally
- Considering associated problems
- Options to consider
- Multidisciplinary team involvement
  - Paediatric/Fetal cardiologist
  - Fetal Medicine Consultant
  - Fetal Cardiac Nurse Specialists
  - Neonatal team
  - Local neonatal/paediatric/obstetric teams

  - Depending on extracardiac involvement (geneticist, general surgeon, neurosurgeon, urologist, cleft palate teams)
  - Hospice/Palliative care team
  - Specialist Midwife
  - Others – mental health team, etc

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Predicting antenatal course

- Cardiac
  - Natural history of cardiac lesion during pregnancy
  - Potential for improvement/no change/deterioration
  - Potential for new findings with subsequent reviews
  - Likely scenario for each possibility

- Extra-cardiac
  - Variable depending on abnormality seen
  - Chromosomal issues / part of syndrome
  - Impact of these on the CHD – extracardiac issues alone may have reasonable outcome; combination may make certain conditions non-survivable

Important to see at least 6-8 weeks apart to be able to see any significant change(s)

Antenatal course – complicating issues

- Prematurity
- Size at birth
- Associated extracardiac abnormality
- Associated chromosomal issues / possible syndrome
- Potential functional issues
- Multiple pregnancy and implications

Antenatal Dx - Parental predicament…..

- Uncertainty of outcome in some cases (especially borderline ventricles or complex DORV)
- Unable to ascertain degree of extracardiac involvement / potential functional issues
- Parents want definite answer
- Need postnatal confirmation before decide on management
- Religious / social constraints
- Multiple pregnancy

Perinatal planning

- Place of delivery
  - Where to deliver (local team preference, parental preference, local guidelines)

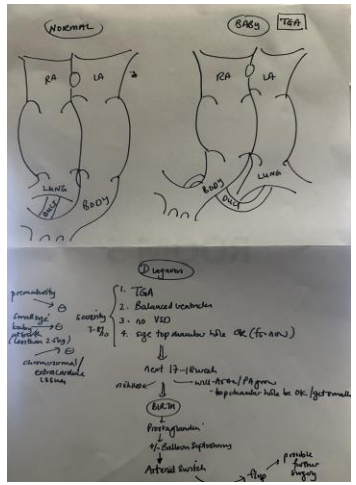
- Timing of delivery
  - 39-40 weeks induction Vs spontaneous labour
  - Maternal disease, multiple pregnancy, IUGR, T21

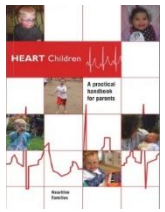
- Method of delivery
  - SVD vs assisted delivery

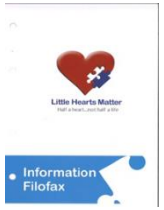
- Immediate postnatal management
  - Prostaglandin dependent/not
  - Acceptable saturations
  - Timing of transfer to Surgical unit/ Home
  - Monitoring / follow-up plan

Information provided
- what do parents already know?
- what do they understand?

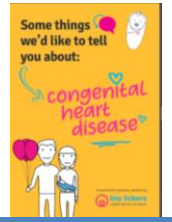
- Cardiac Diagnosis
  - Drawings
  - Printed information/handbooks
- Extracardiac diagnosis
- Option for Amniocentesis – microarray/whole exome
- Independent advice
  - Antenatal Results and Choices (ARC)
  - Little Heart Matters
  - Max Appeal
- Contact details for Fetal cardiologist/Fetal cardiac nurse specialist (fCNS)







Children's Heart FEDERATION



MAX APPEAL
IMPROVING LIVES OF 22q11 FAMILIES

Parental support

- Contact details important – usually more questions after clinic
- Fetal cardiac nurse specialists (fCNS)
  - vital role especially for continuing pregnancies
    - Contact with other families
    - Further information about cardiac condition
    - Practical advice – support at/after diagnosis (emotional and practical)
    - "link" between antenatal and postnatal status
    - Professional Liaison with Obstetrician and Cardiologist
    - Signposting
    - Decision Making contact
  - Link for future pregnancies
- Fetal midwives – especially for non-continuing pregnancies
- Hospice team – for complex CHD/single ventricle physiology/multiple anomalies

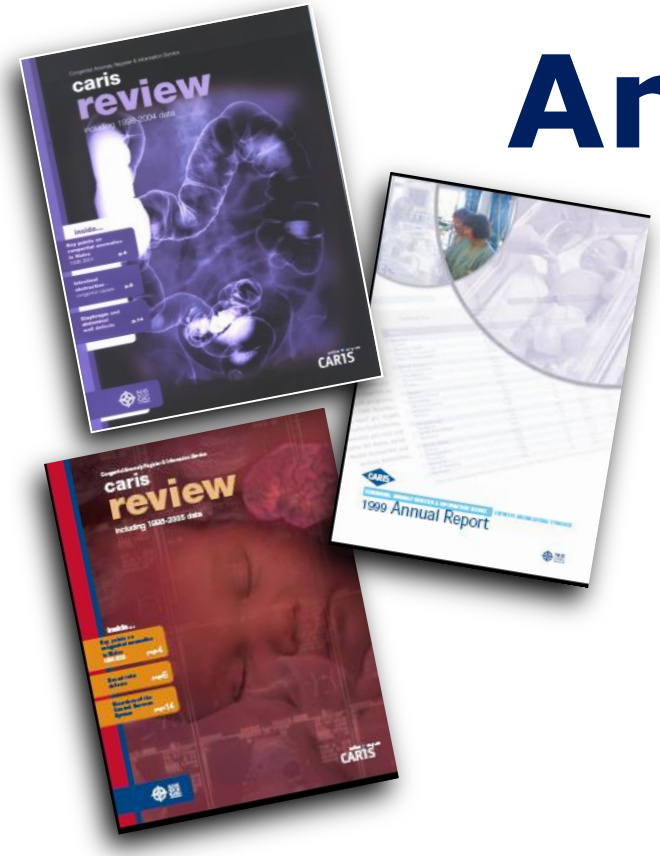
QUESTIONS?

Alder Hey

Congenital HEART NETWORK
NORTH WEST | NORTH WALES | ISLE OF MAN

CARIS

Congenital Anomaly Register and Information Service



Annual Meeting 2024

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Iechyd Cyhoeddus Cymru

Public Health Wales

Paediatric management of Congenital heart defects

Dr Rajesh Viswanathan

Consultant Paediatrician

Wrexham Maelor Hospital

- Overview of Congenital heart defects:

Classification

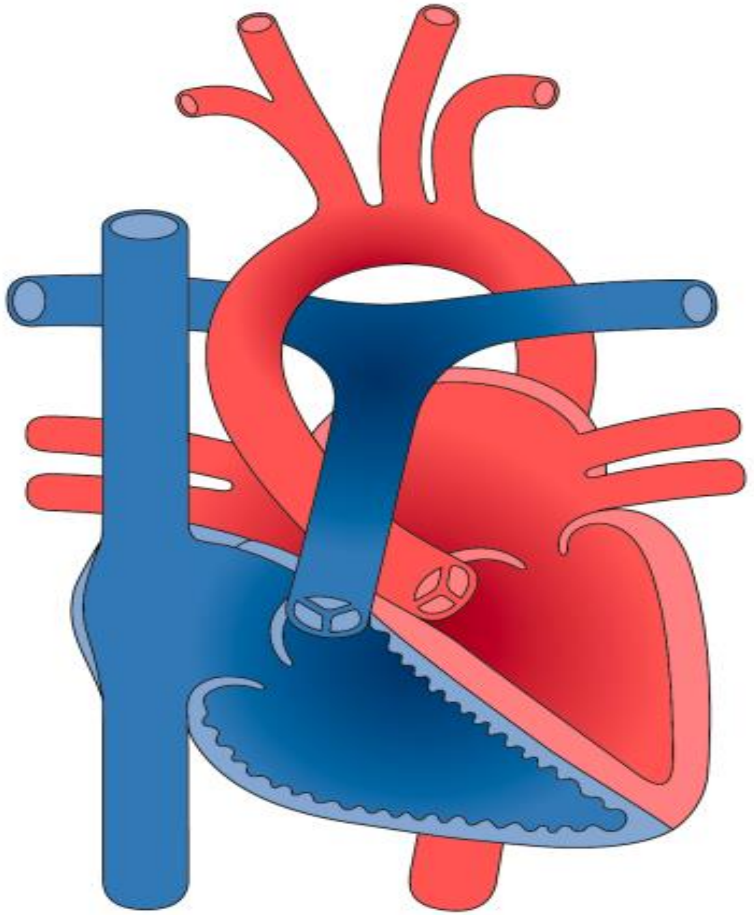
Examples from each class with definite management

- Paediatric role

Classification of heart defects

- Structure
- Function (Cardiomyopathy)
- Conduction system

Normal heart



Classification of Structural heart defects

- Acyanotic:

Obstructive lesions:

Regurgitation lesions:

Left to Right shunts:

- Cyanotic:

Decreased pulmonary blood flow:

Increased pulmonary blood flow:

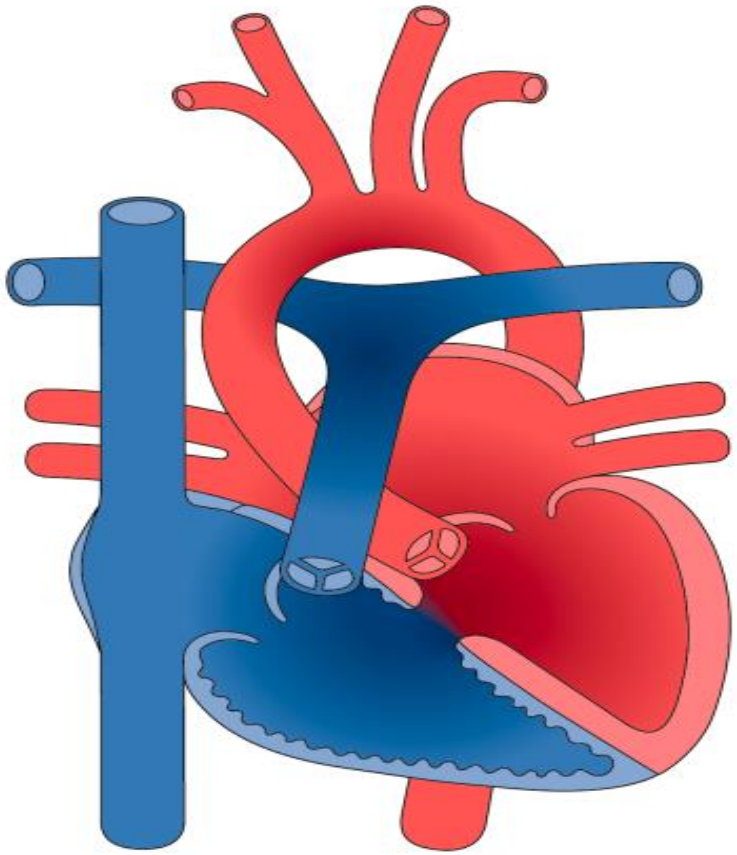
Left to right shunts

- Atrial septal defect
- Ventricular septal defect
- Patent ductus arteriosus
- Partial anomalous pulmonary venous return
- Atrioventricular septal defects
- Aortopulmonary window

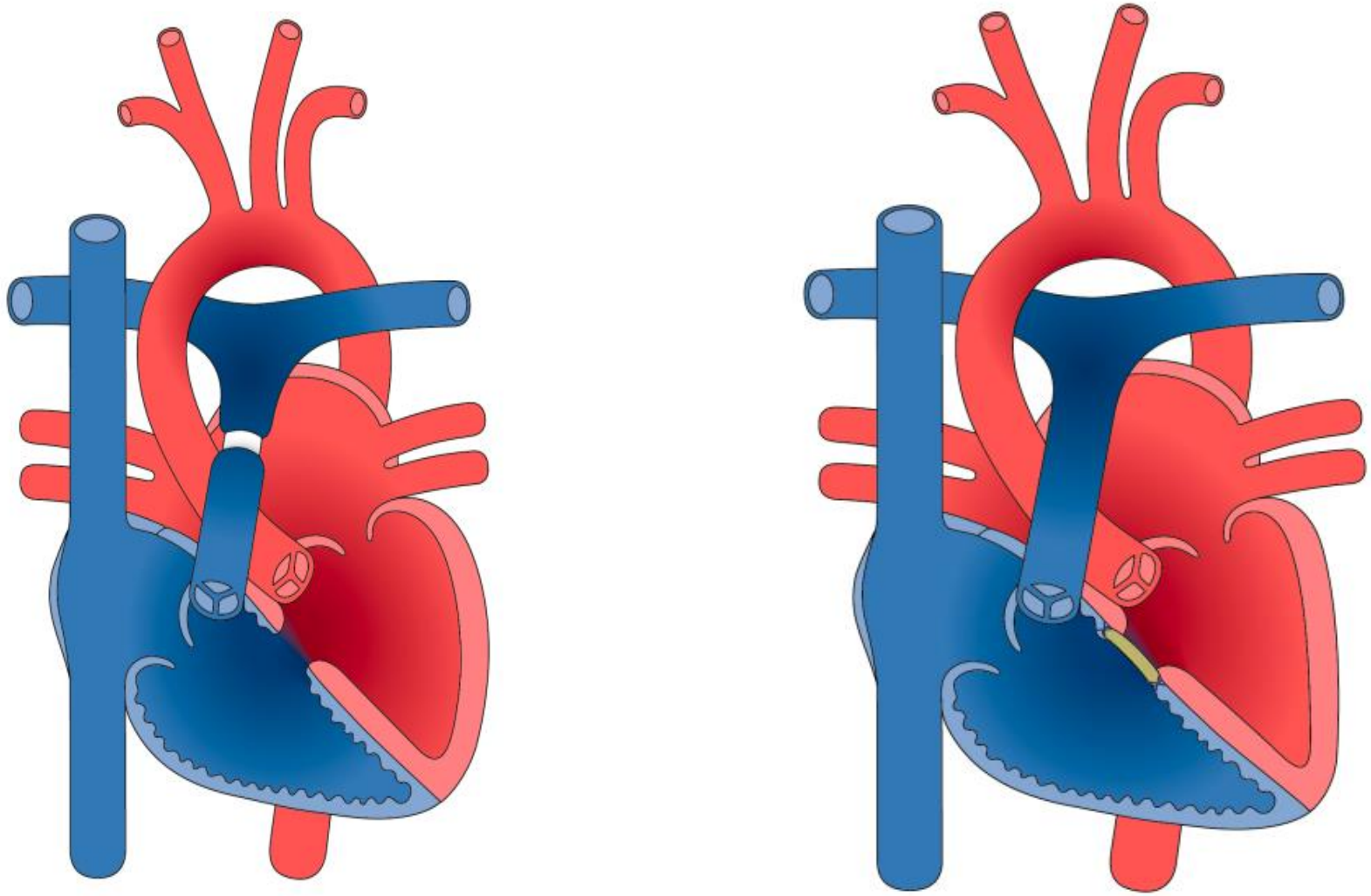
Communication between systemic and pulmonary circulation

Heart failure

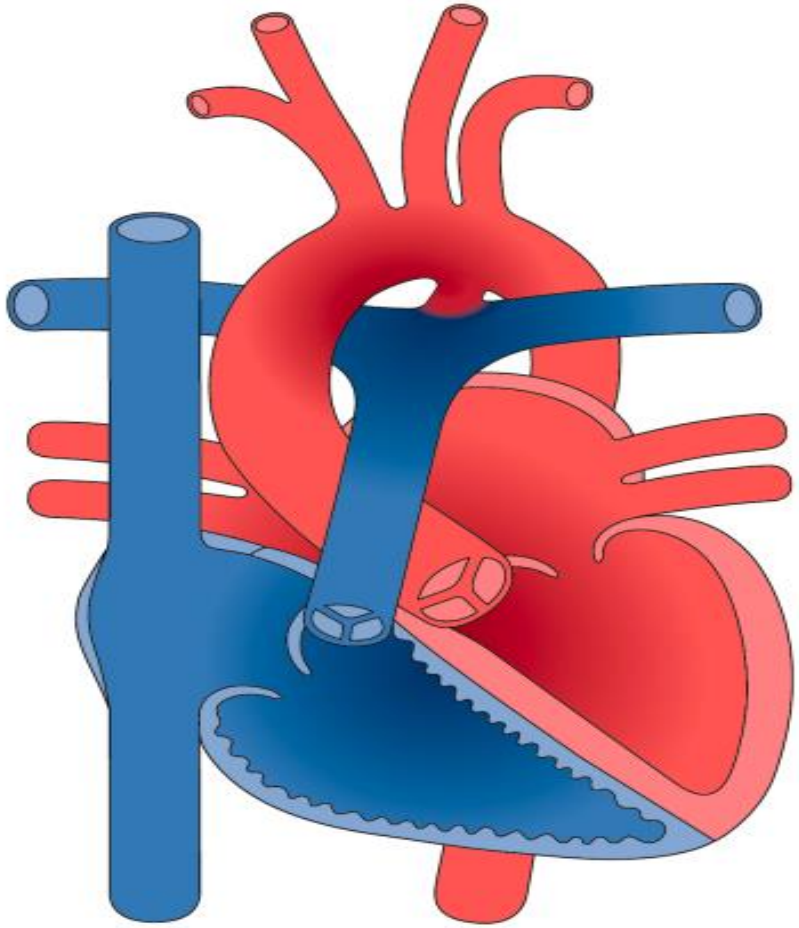
Ventricular septal defect



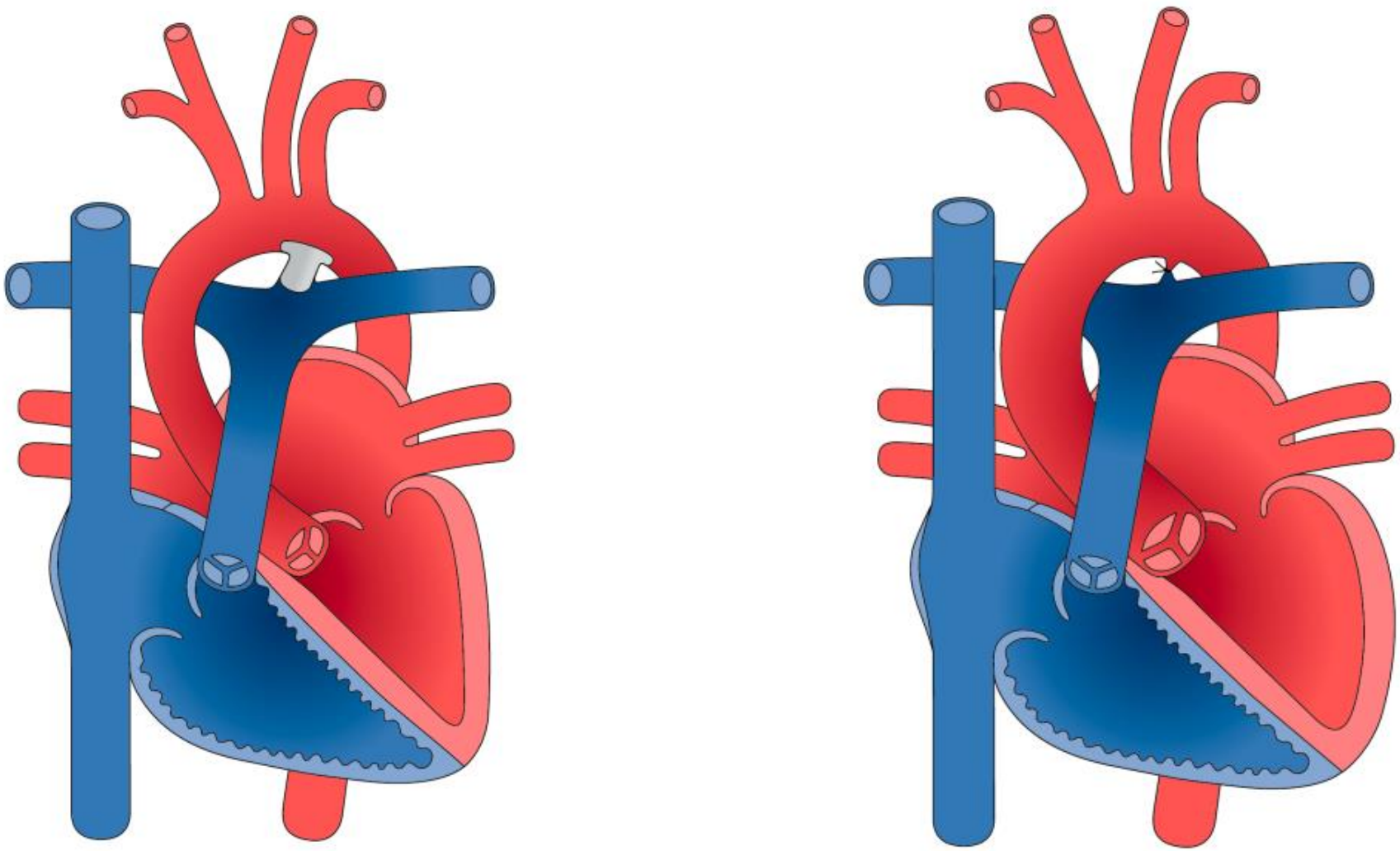
Treatment



Patent Ductus arteriosus



Treatment



Obstructive lesions

- Obstruction to ventricular outflow:

Valvular: Pulmonary stenosis, aortic stenosis

Below the valve: subaortic membrane, double chambered RV

Above the valve: supravalvular aortic stenosis, coarctation of aorta

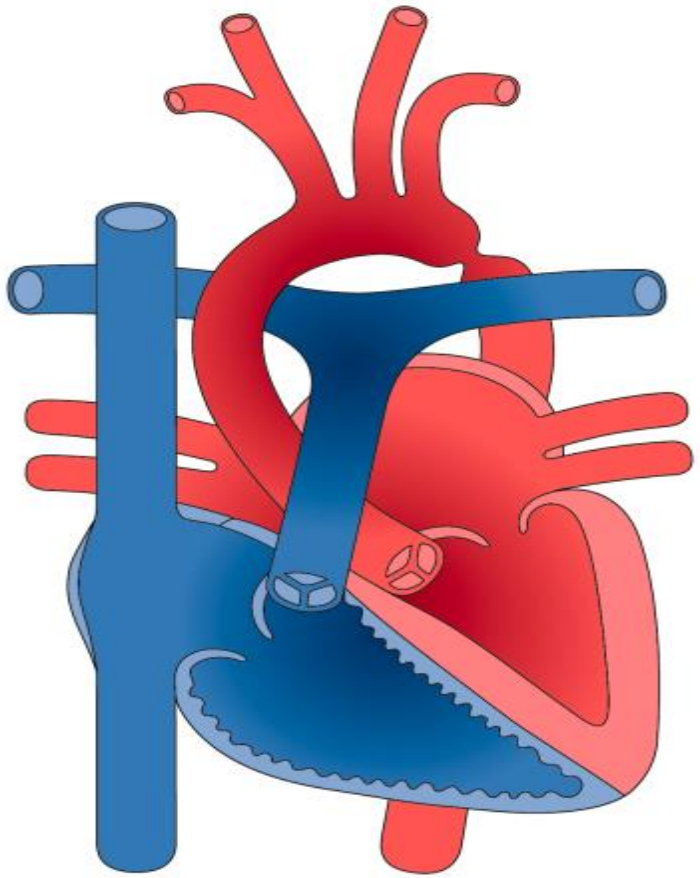
- Obstruction to ventricular inflow:

Cor triatriatum

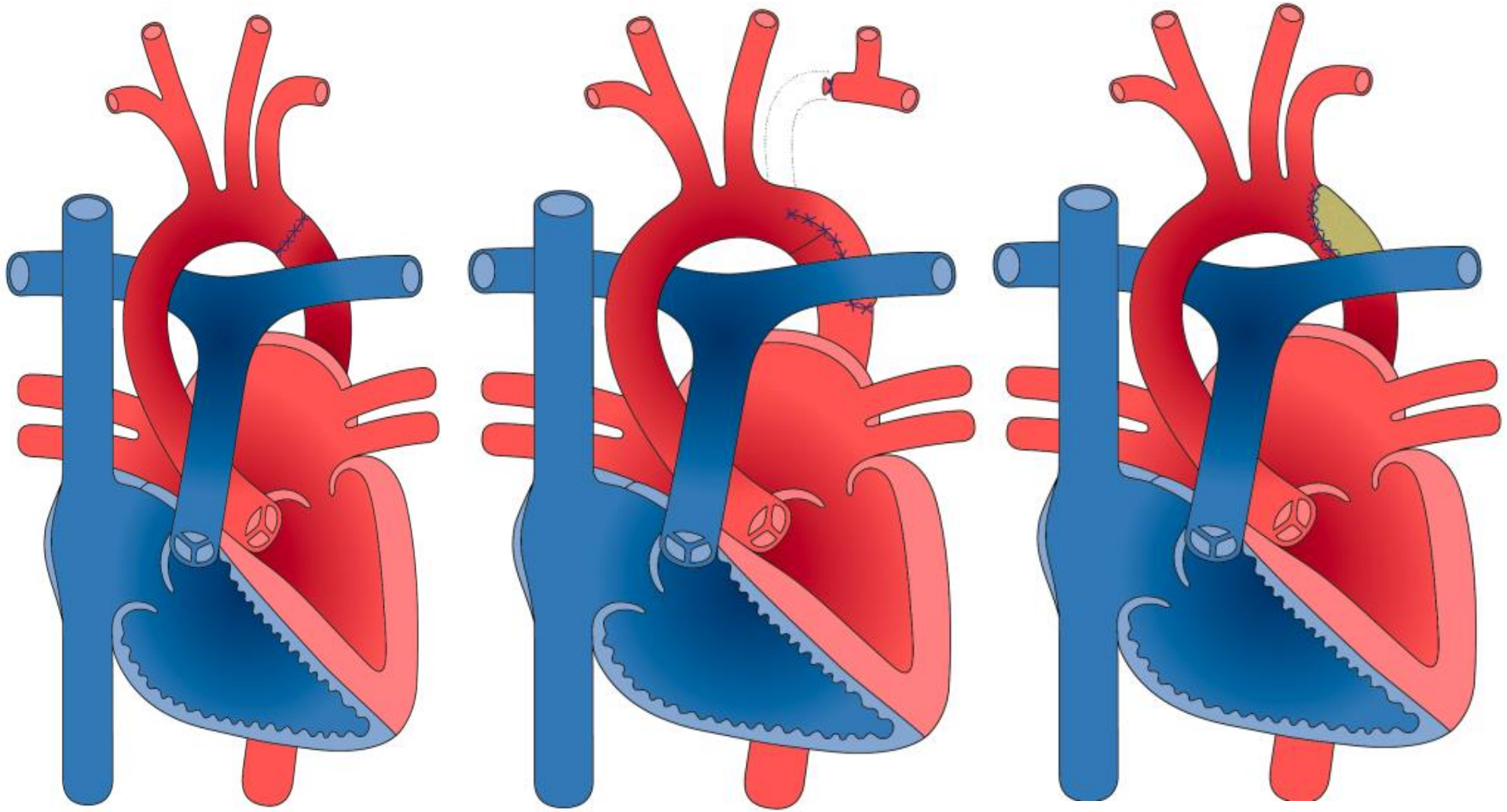
Obstruction to pulmonary veins

Shock

Coarctation of aorta



Treatment

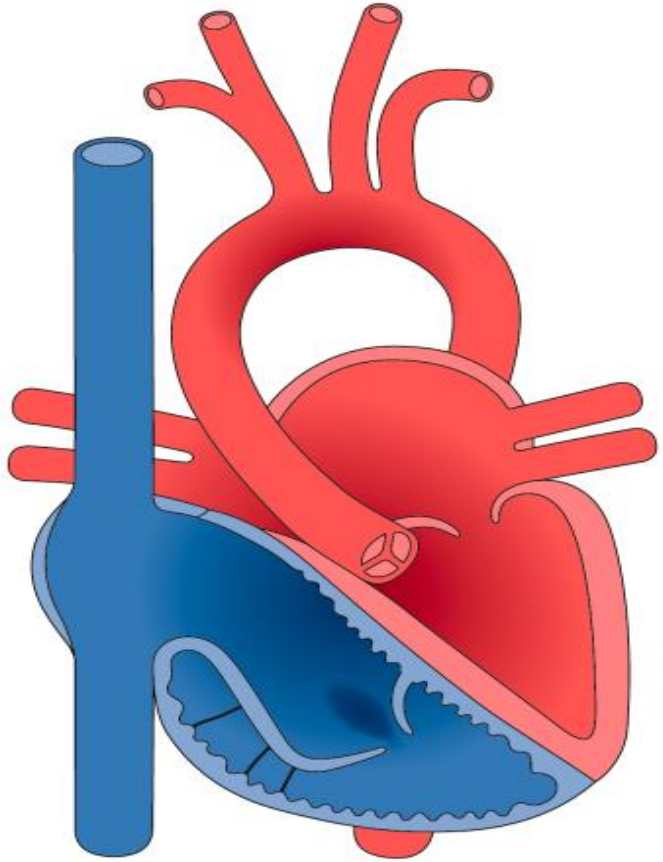


Regurgitant lesions

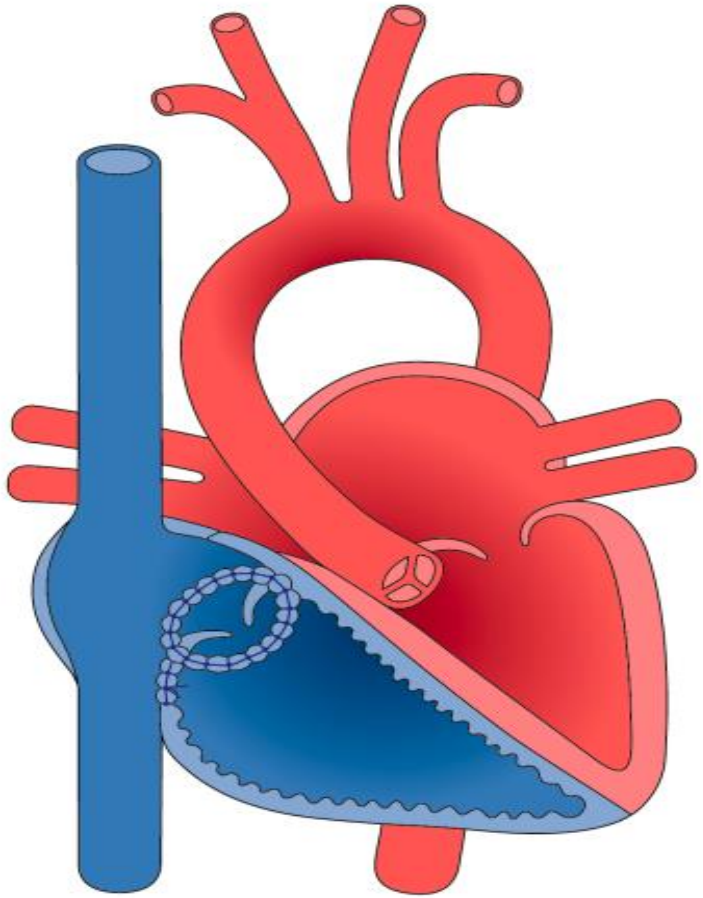
- Aortic regurgitation: Bicuspid aortic valve
- Mitral regurgitation: Mitral valve prolapse
- Tricuspid regurgitation: Ebstein’s anomaly
- Pulmonary regurgitation: Congenital absence of pulmonary valve

Heart failure

Ebstein’s anomaly



Treatment



Cyanotic CHD (decreased pulmonary blood flow)

- Obstruction to right ventricular outflow:

Tetralogy of Fallot

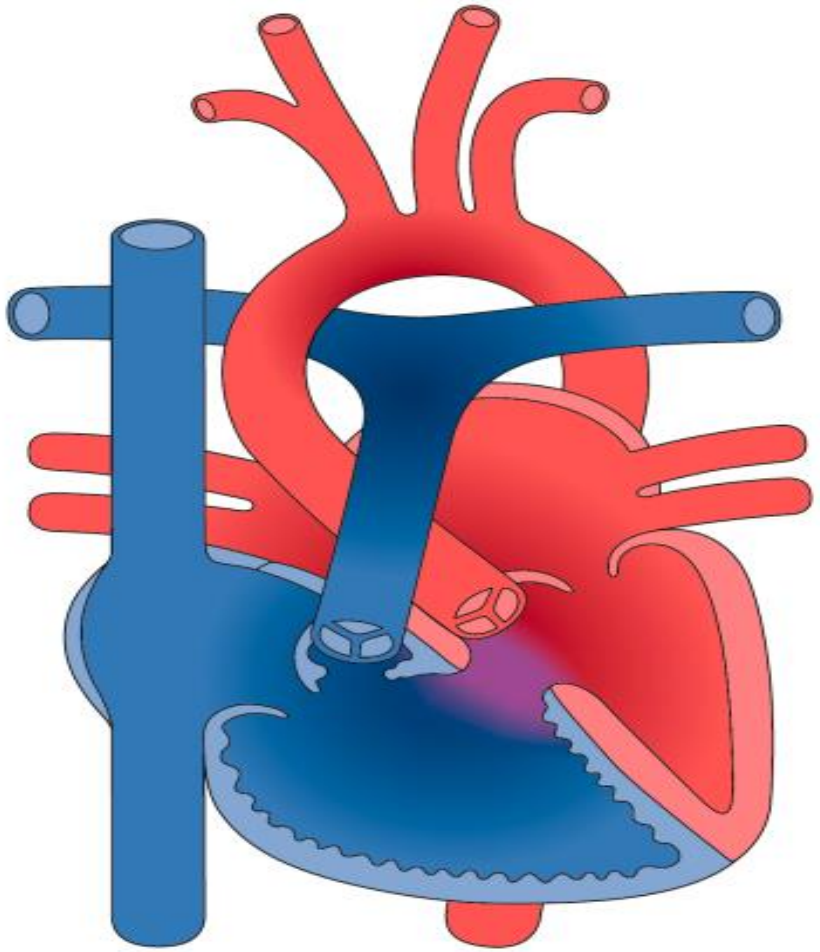
Pulmonary atresia with intact ventricular septum

- Obstruction to right ventricular inflow:

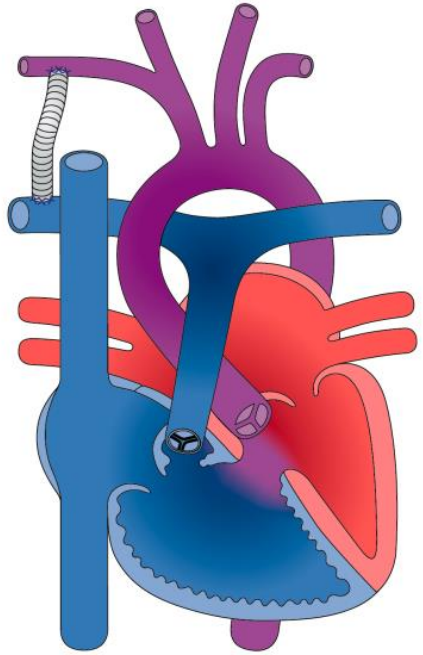
Tricuspid atresia

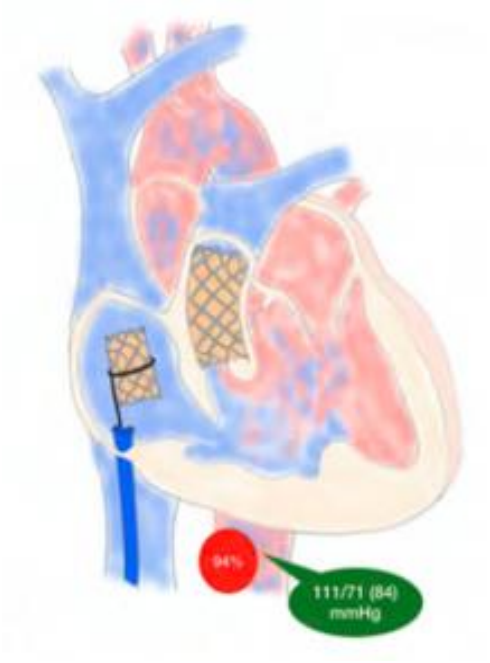
Cyanosis

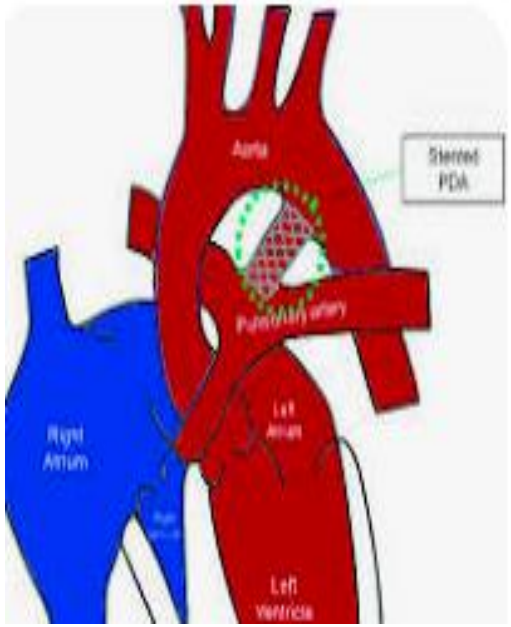
Tetralogy of Fallot



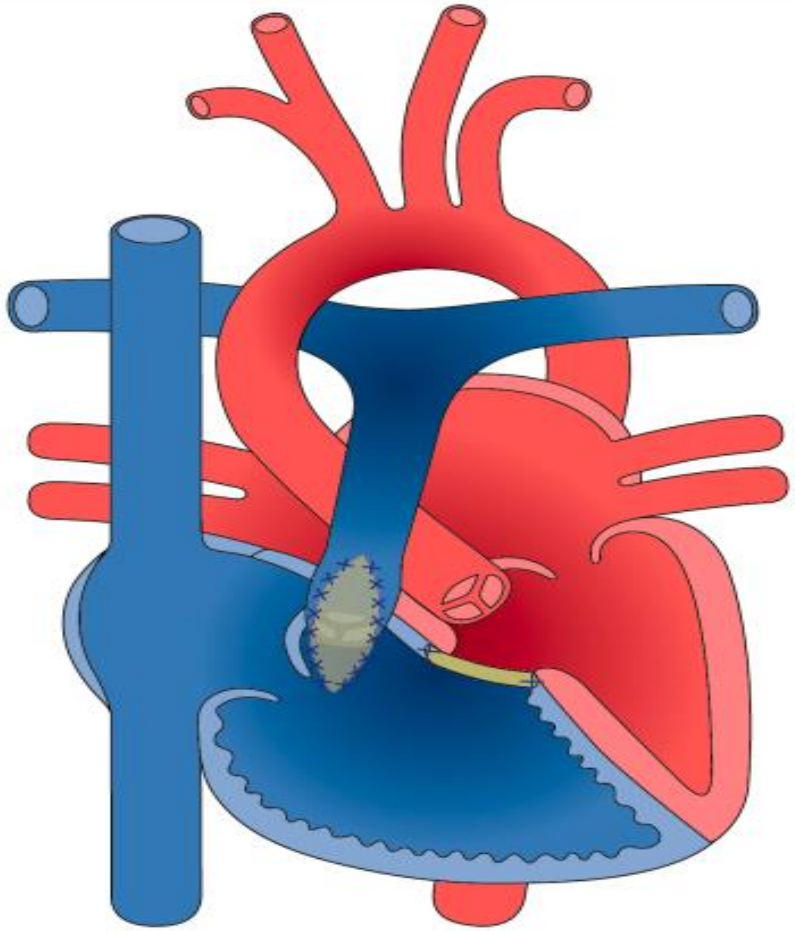
Tetralogy of Fallot treatment







Tetralogy of Fallot treatment



Cyanotic CHD (Increased pulmonary blood flow)

- Abnormal ventricular and arterial connection:

Transposition of great arteries

- Total mixing of systemic venous and pulmonary venous blood:

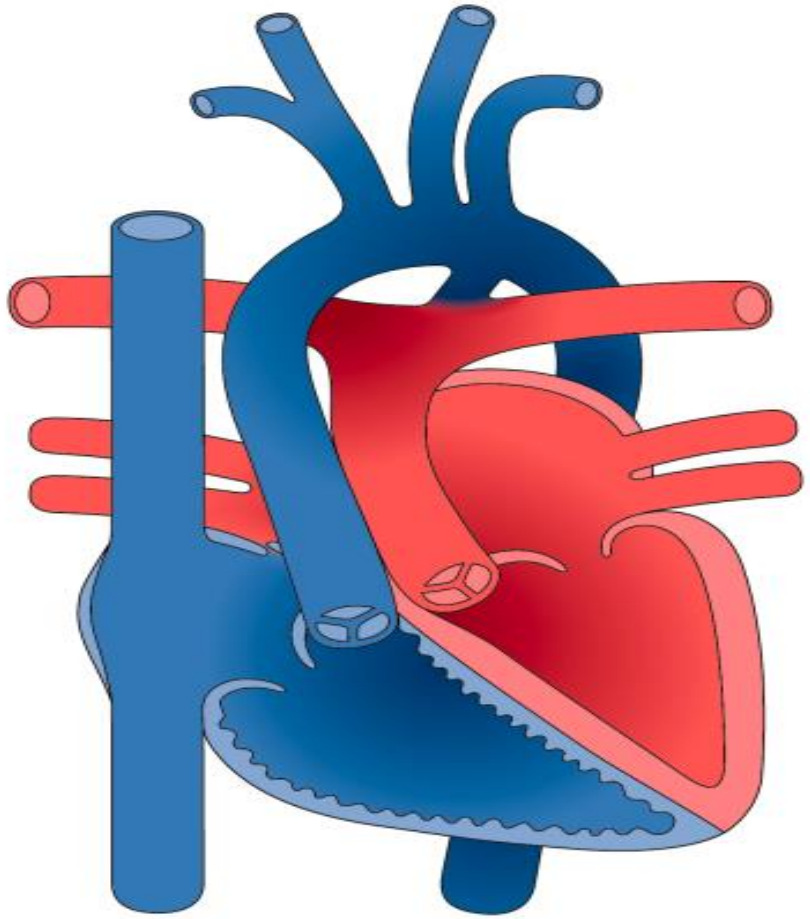
Hypoplastic left heart syndrome

Truncus arteriosus

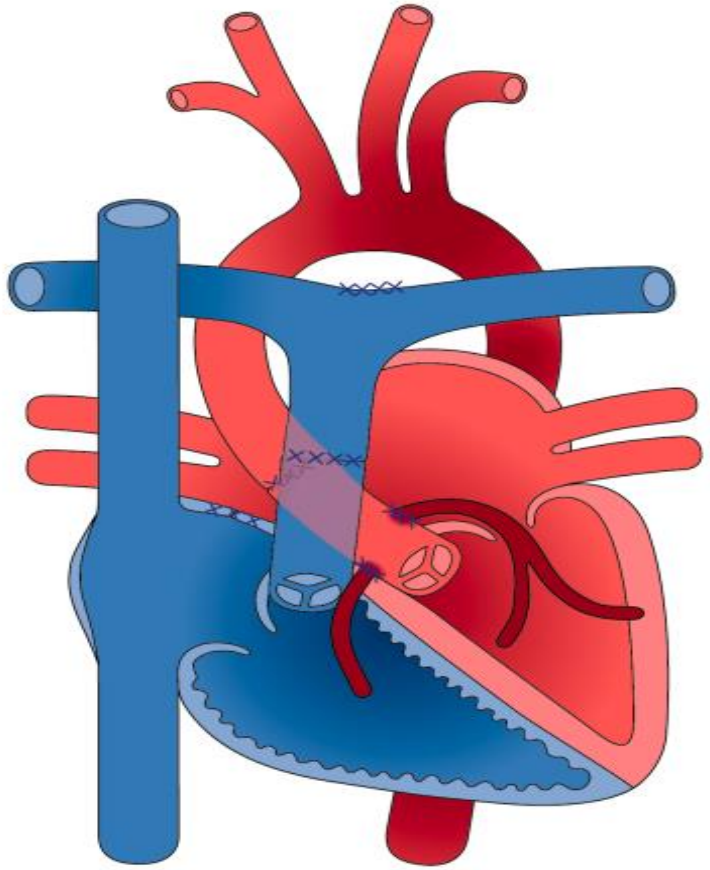
Total anomalous pulmonary venous return

Cyanosis and Heart failure

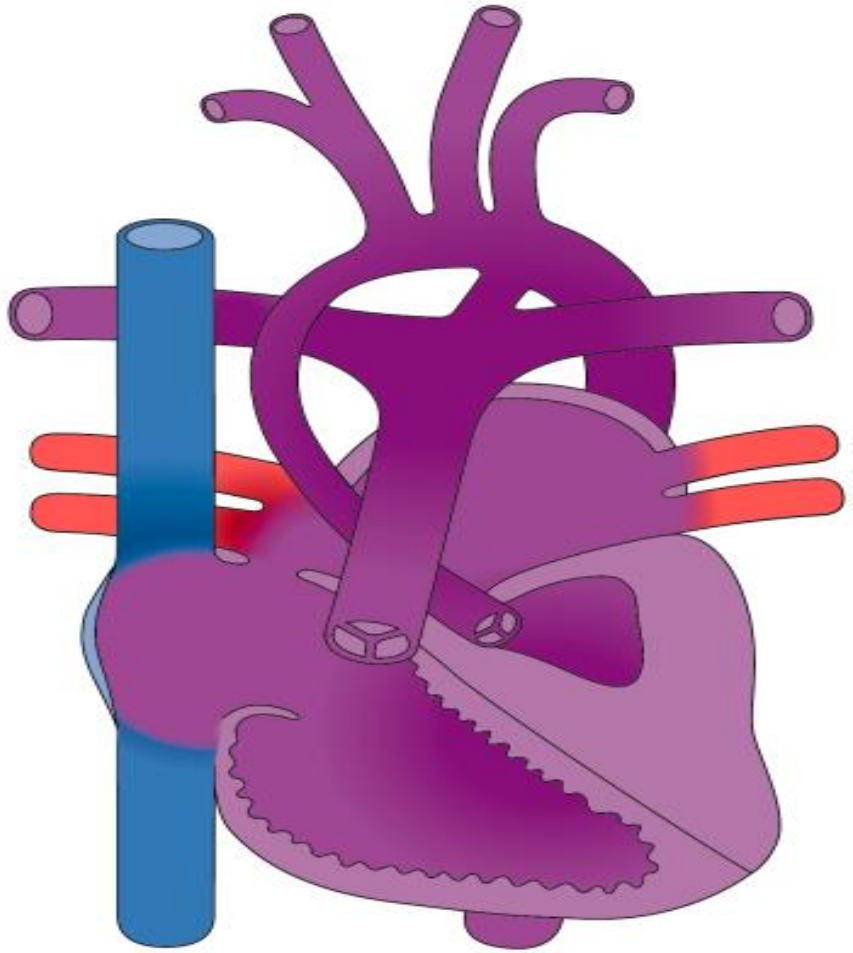
Transposition of great arteries



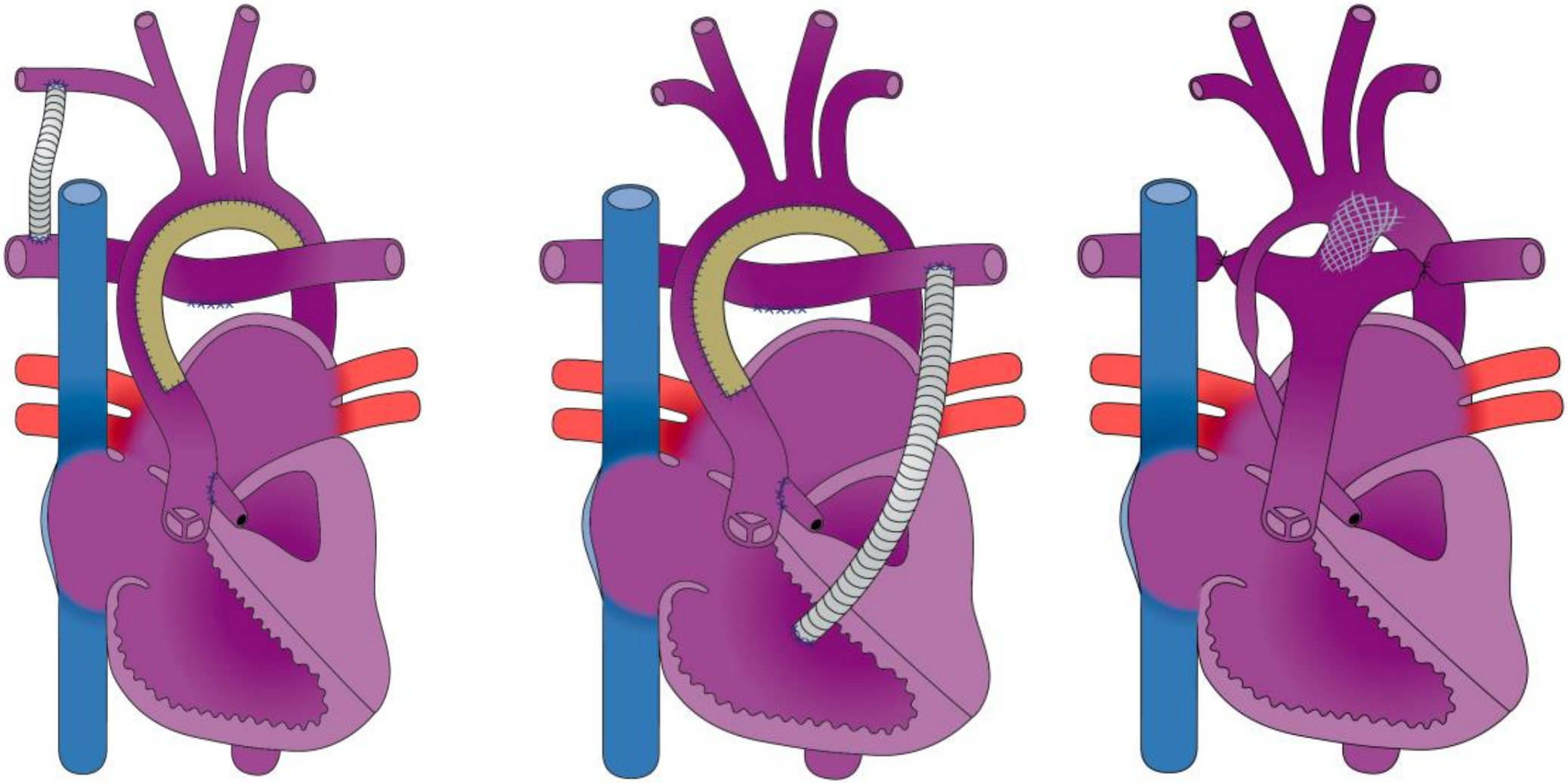
Arterial switch



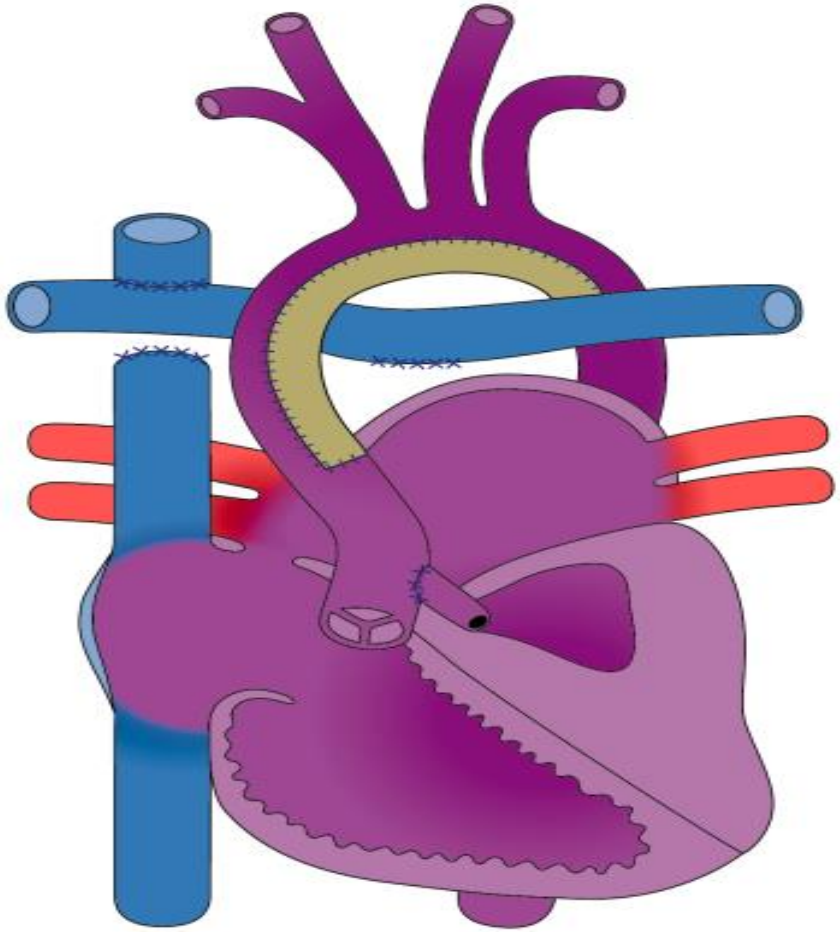
Hypoplastic Left heart syndrome



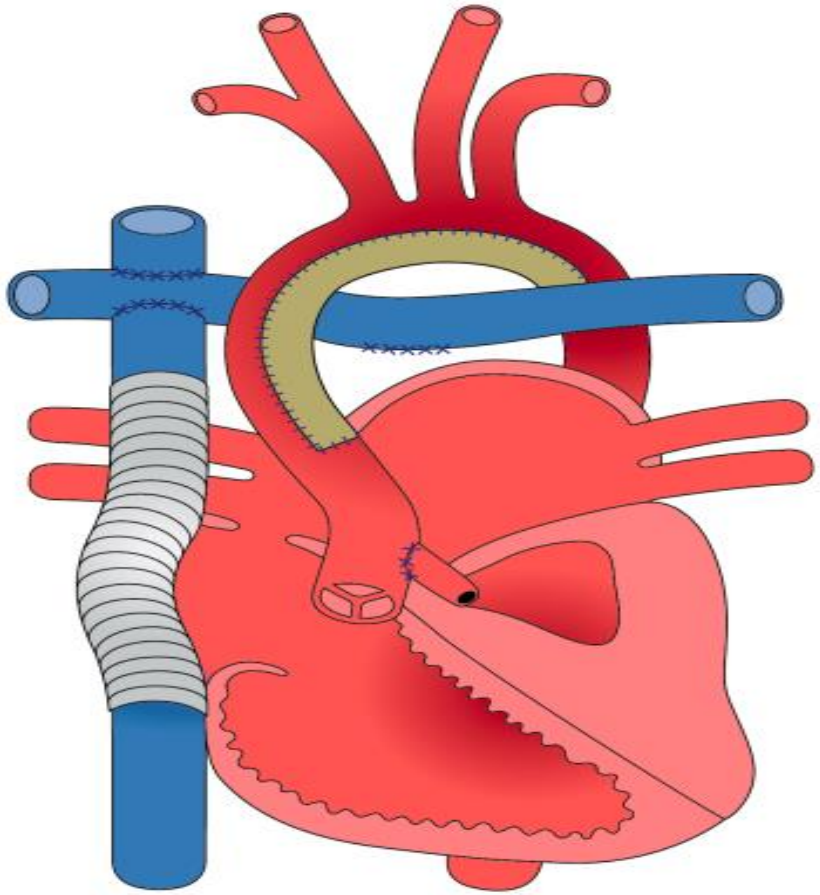
Stage 1



Stage 2 – Glenn



Stage 3 - Fontan



Classification based on Ductal dependency

- Duct dependent Systemic circulation:

Critical aortic stenosis

Hypoplastic left heart syndrome

Interrupted aortic arch

Coarctation of aorta

- Duct dependent Pulmonary circulation:

Critical pulmonary stenosis

Tetralogy of Fallot

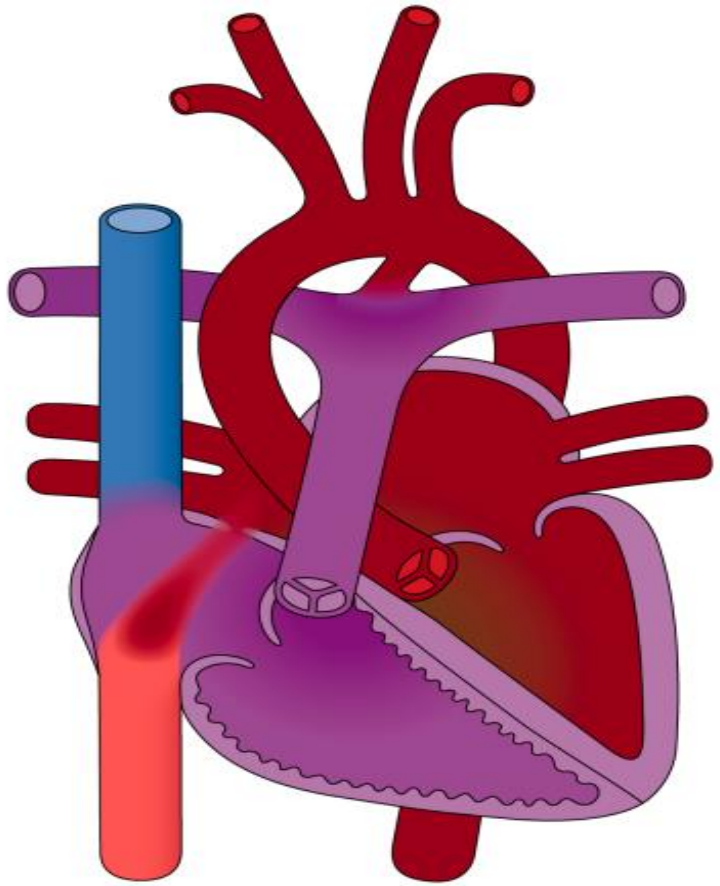
Pulmonary atresia

Tricuspid atresia

- Duct dependent Parallel circulation:

Transposition of great arteries

Foetal circulation



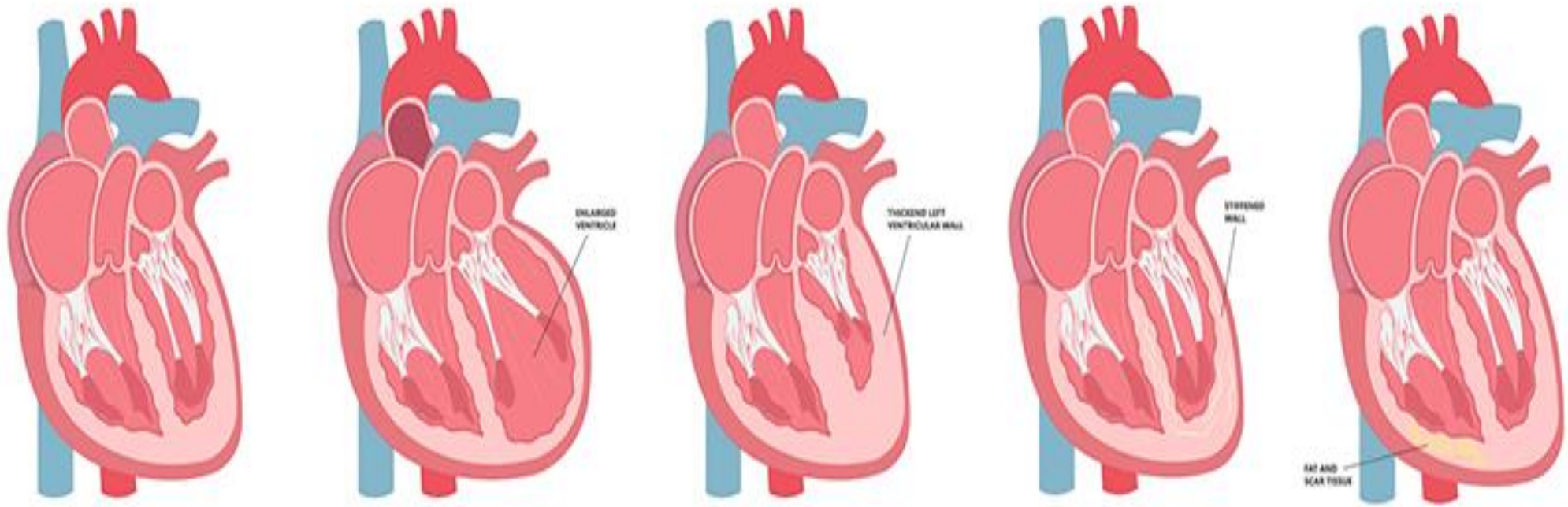
Clinical presentation	CHD		Timing of presentation Birth — 1st week — 2nd week — 1-2 months
Cyanosis (central cyanosis, hypoxia not improved by O_2)	TGA		→
	Tetralogy of Fallot		→ .....
	Tricuspid atresia		→
	Pulmonary atresia		→
	Truncus arteriosus		→
	TAPVR – obstructed		→
Shock (poor feeding, lethargy, tachycardia, pallor/pale grey skin, weak peripheral pulses, ↑ capillary refill, hypotension/decreased blood pressure in lower extremities)	Left obstructive lesions:	• Hypoplastic left heart syndrome	→
		• Critical aortic stenosis	→
		• CoA	→
	ALCAPA myocardial infarction		→
CHF (feeding difficulties, sweating, failure to thrive, difficulty breathing, tachypnea, rales, hepatomegaly)	VSD		→ .....
	Patent ductus arteriosus		→
	Atrioventricular canal		→ .....
	TAPVR – not obstructed		→ .....
	ALCAPA – recurrent ischemia		→ .....

CHF: congestive heart failure; CHD: congenital heart disease; TGA: transposition of great arteries; TAPVR: total anomalous pulmonary venous return; CoA: coarctation of the aorta; ALCAPA: anomalous left coronary artery from pulmonary artery; VSD: ventricular septal defect.

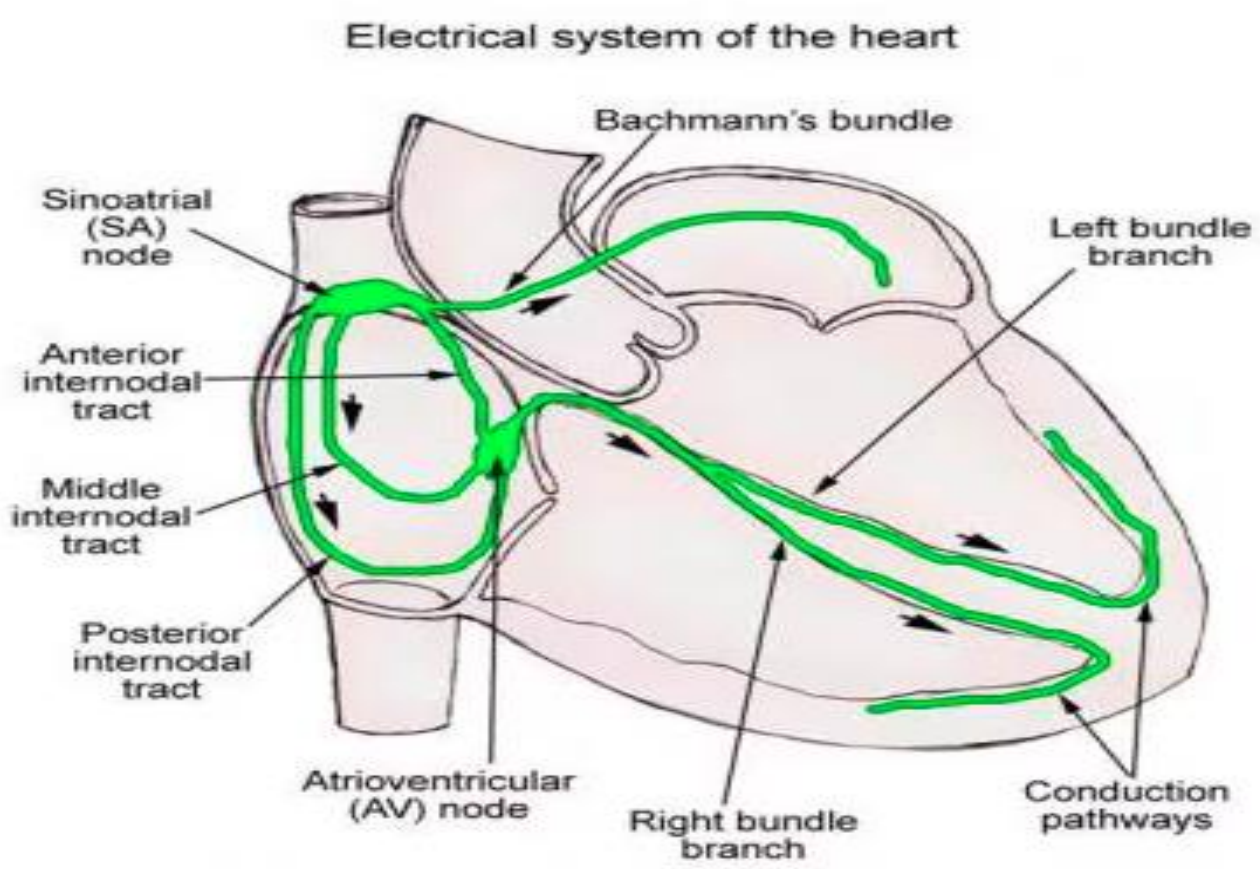
Cardiomyopathy

- Hypertrophic
- Dilated
- Restrictive
- Left ventricle non compaction
- Arrhythmogenic ventricular cardiomyopathy

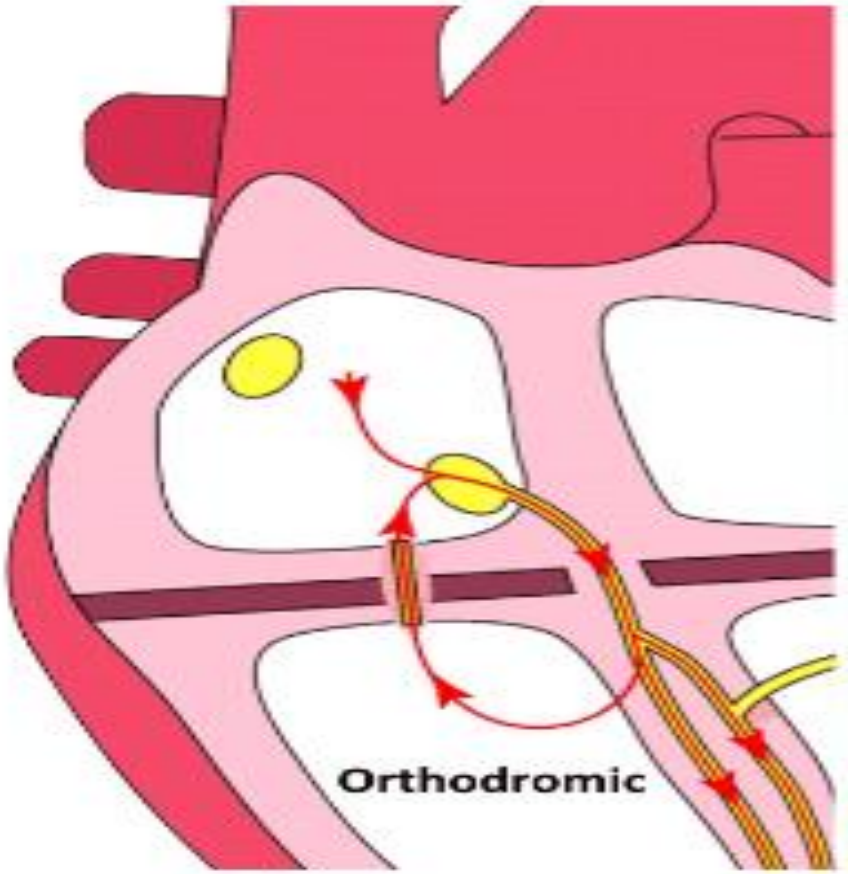
CARDIOMYOPATHY



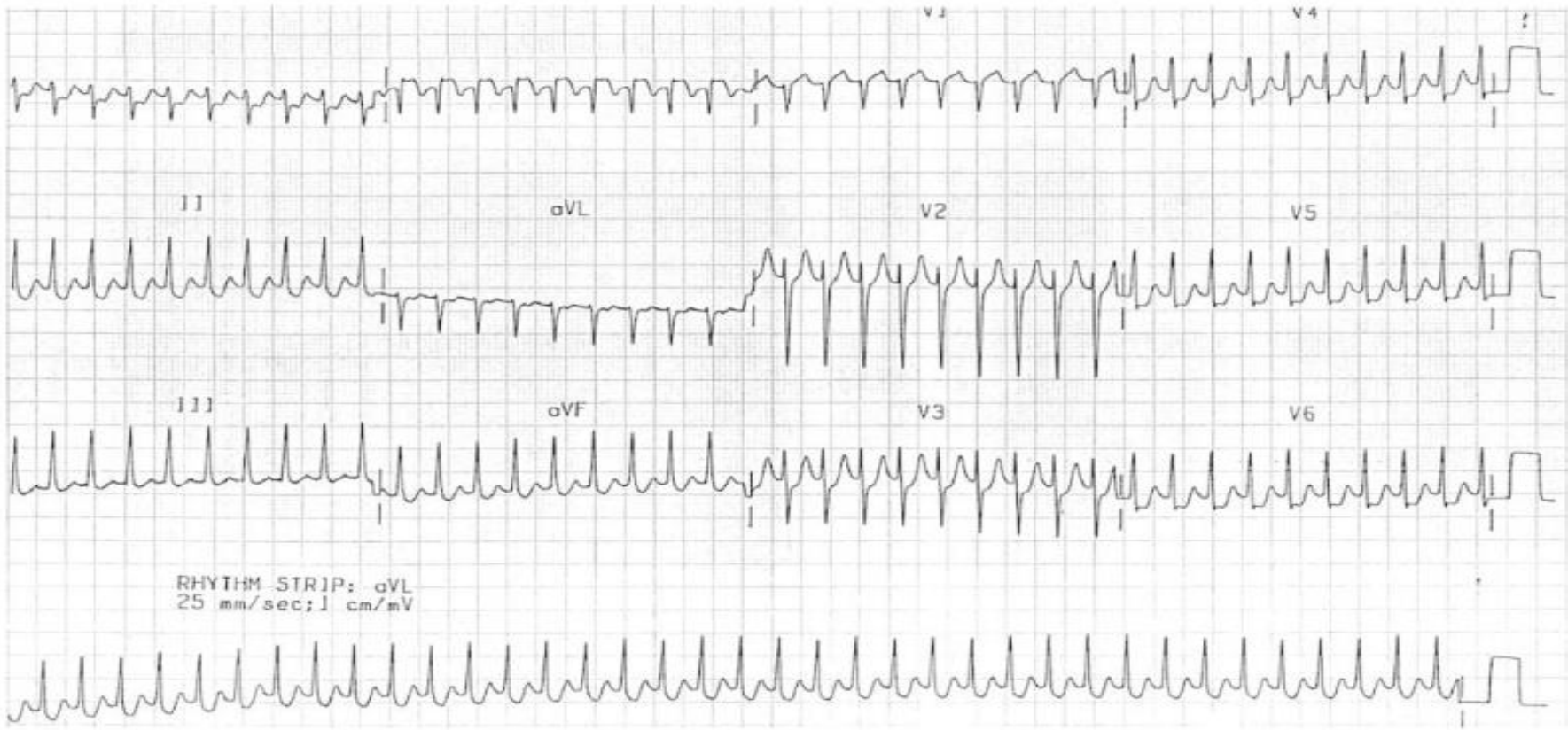
Conducting system of heart



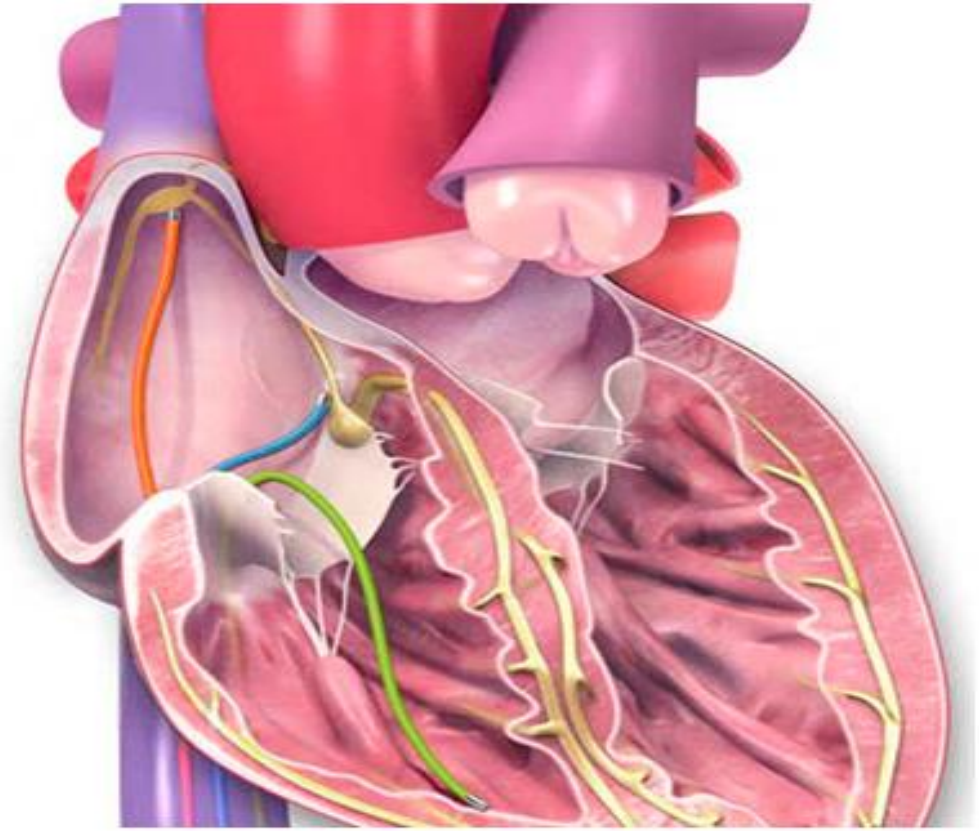
Supraventricular tachycardia (AVRT)



SVT ecg



Radiofrequency ablation



Screening and Diagnosis

- New born and infant physical examination
- Pulse oximetry testing (measuring preductal and post ductal saturations)
- Symptoms: Murmur, chest pain, breathlessness, palpitations, fainting, central cyanosis, feeding problems, failure to thrive, oedema and shock
- Family history of inherited cardiac conditions, sudden cardiac death, BAV, etc
- Genetic conditions, Congenital anomaly, Neuromuscular conditions and Connective tissue disorders

Diagnostic procedures like: CXR, Echocardiogram, standard ECG, Ambulatory ECG, Exercise ECG and Event recorder.

Initiation of treatment

- Heart failure medicines like diuretics and afterload reducing agents
- Stabilisation of sick babies and children and transfer to tertiary services
- Starting of prostin in duct dependent lesions (with input from foetal medicine and foetal cardiology)
- Medicines to control rhythm problems

Monitoring

- Single ventricle pathway
- Children with cyanosis
- Post surgery/intervention- complications
- Family history

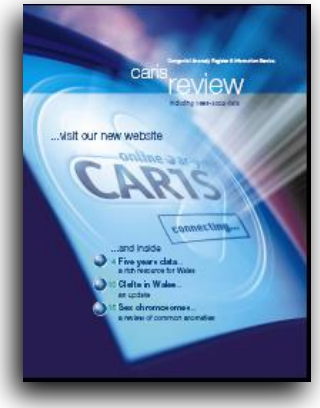
MDT support

- Open access
- Feeding problems: Dietician and speech and language team
- Community nursing team: nutrition, saturation check, provide training and education, liaising with specialist cardiac nurses, wound care
- Immunisation: RSV vaccination
- Genetics: Genetic conditions, inherited cardiac conditions (long QT)
- Cardiac Electrophysiology
- Dentist
- Clinical psychology
- Tertiary cardiology
- GP

Thank you

CARIS

Congenital Anomaly Register and Information Service



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Public Health Wales

Antenatal screening for congenital heart anomalies

Bill Taylor

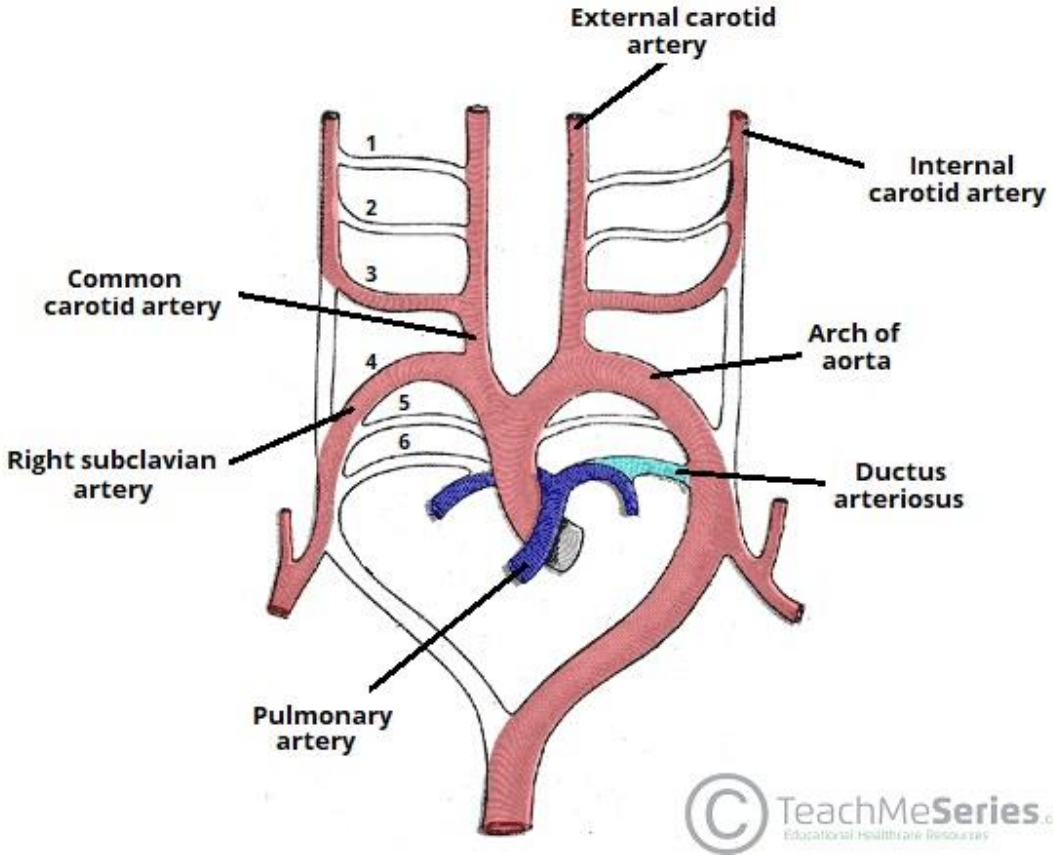
Consultant Obstetrician/Gynaecologist

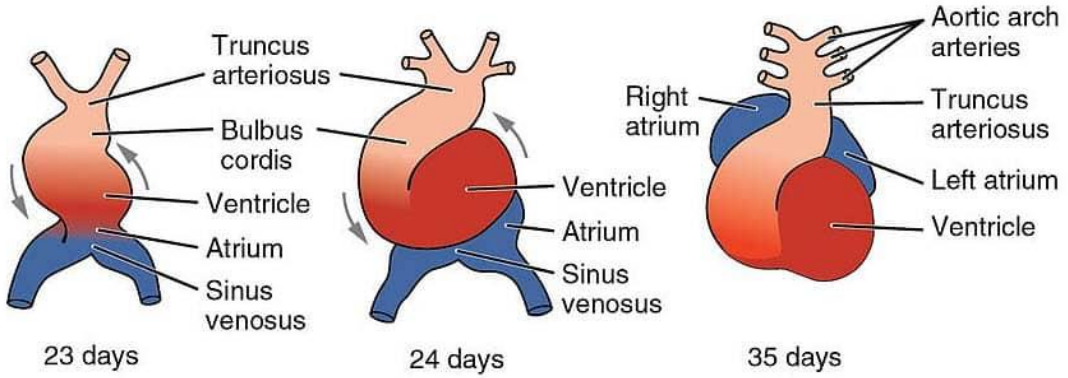
Wrexham Maelor Hospital

Antenatal care package. (Modified health screening.)
Fetal heart screening

- Accurate dating scan in the range 11 to 14 weeks
- Combined screening (NT + chemistry or Quad test for Trisomy screening)
- 20 week Fetal Anomaly scan. (18 – 22 weeks) 99.9 % acceptance.
- Fetal “Echo” – second look at 22 – 23 weeks

Embryology of the heart is complicated. Many stages, folding and remodelling, much to achieve, plenty to go wrong.
First heart beat is on day 21 of Embryonic life.

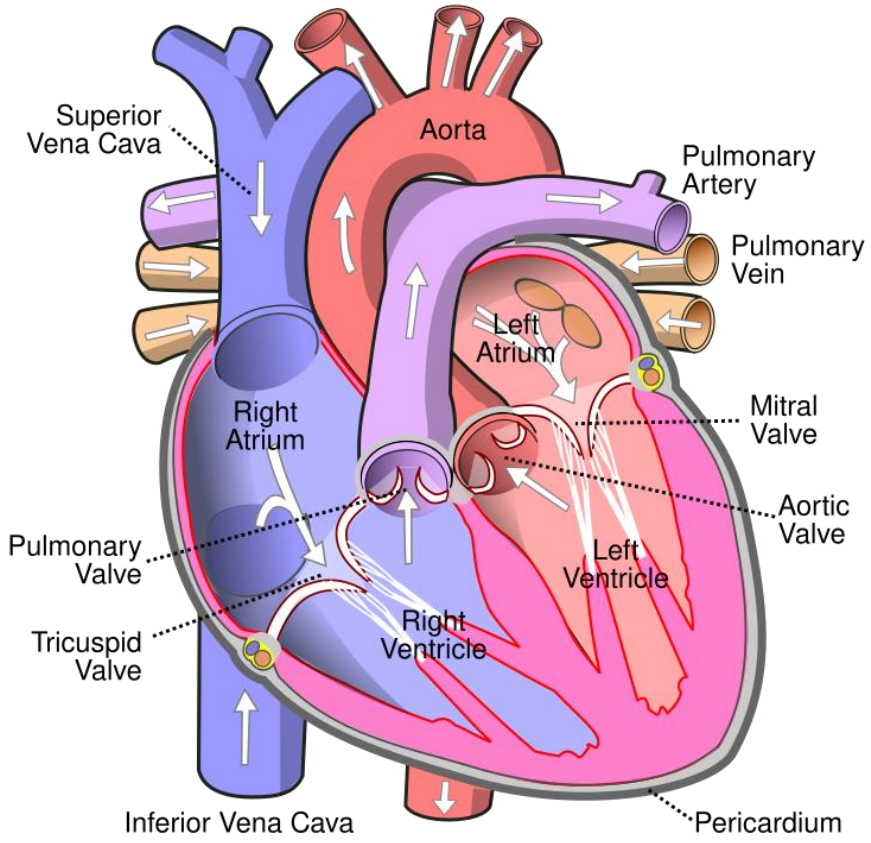




Who gets a “second look” fetal heart screen ?

- Something abnormal seen on FAS / FAS scan not completed
- Raised nuchal translucency > 3.5 mm, and all other tests normal
- Direction from Clinical Genetics service based on previous history
- Maternal / paternal history of CHD
- Previously affected child with CHD , 22q 11 deletion
- Onging medication e.g. lithium , Lamotrigine in high dosage. (“BUMPS” Best Use of Medicines in Pregnancy website)
- Twins – at a later gestation if one isn’t thriving.
- Poorly controlled diabetes especially periconception and first trimester.
- *However 90% of Heart anomalies occurs in pregnancies with no risk factors *

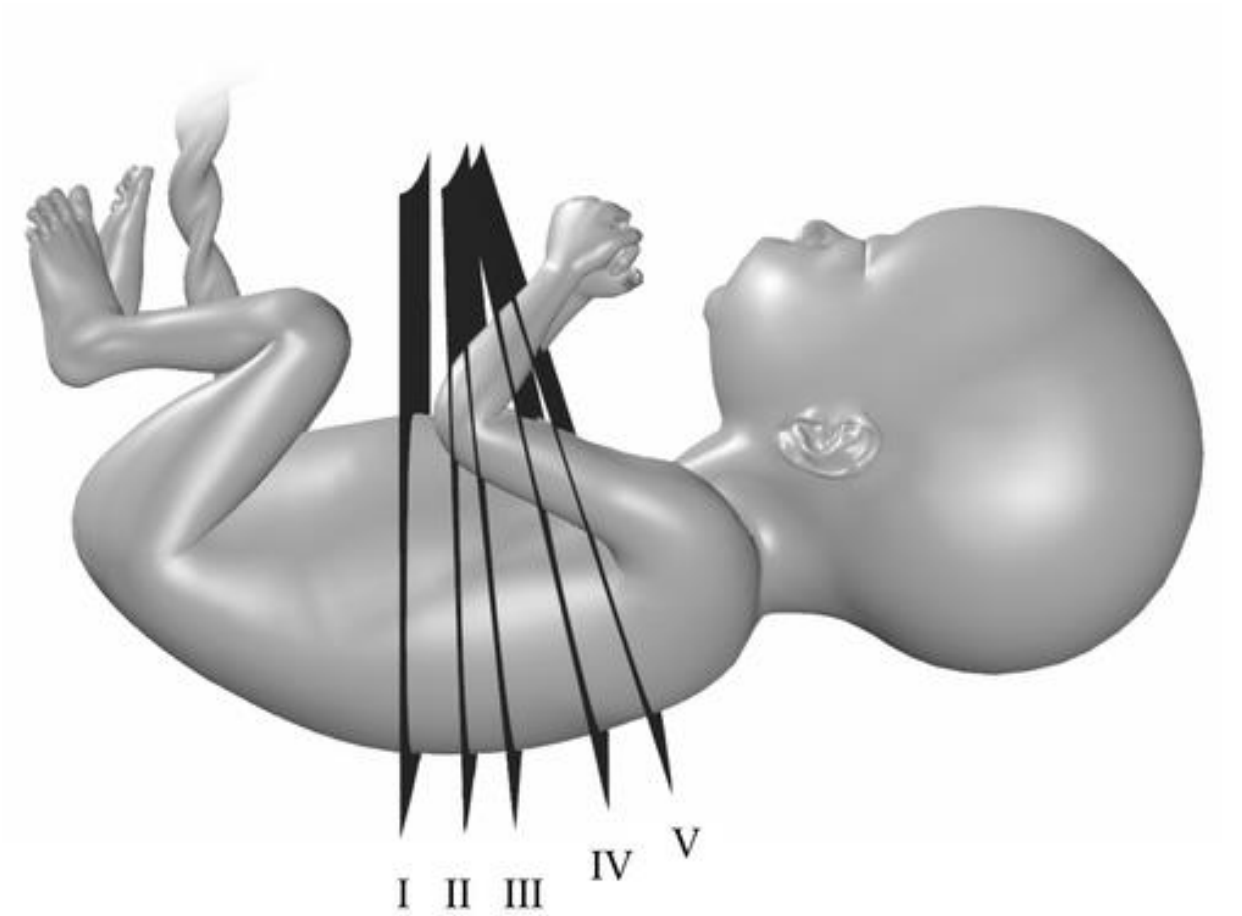
Textbook picture of a human heart.
Almost always drawn from the front of the chest.



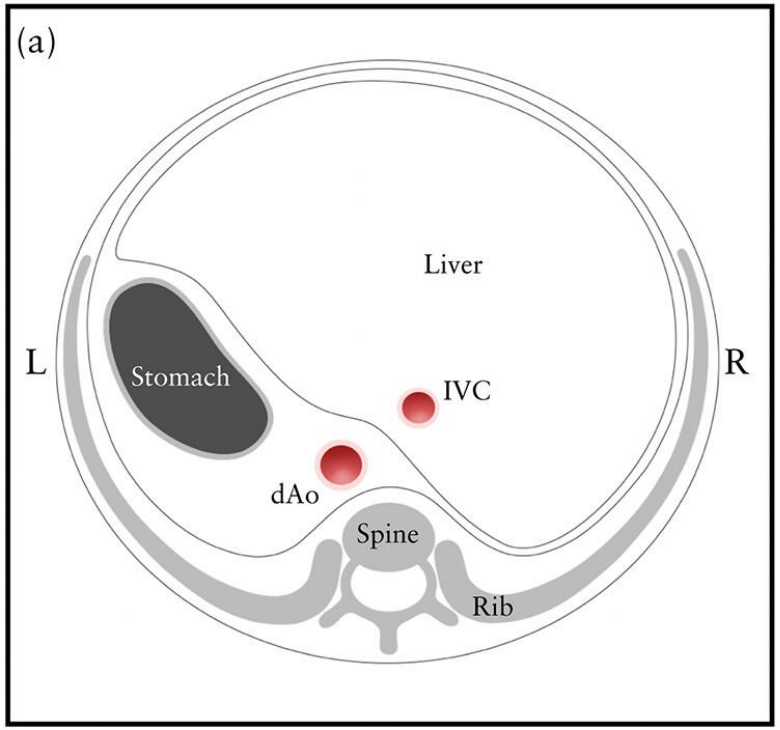
- Can you draw the heart viewed from the left side of the chest, upside down ?

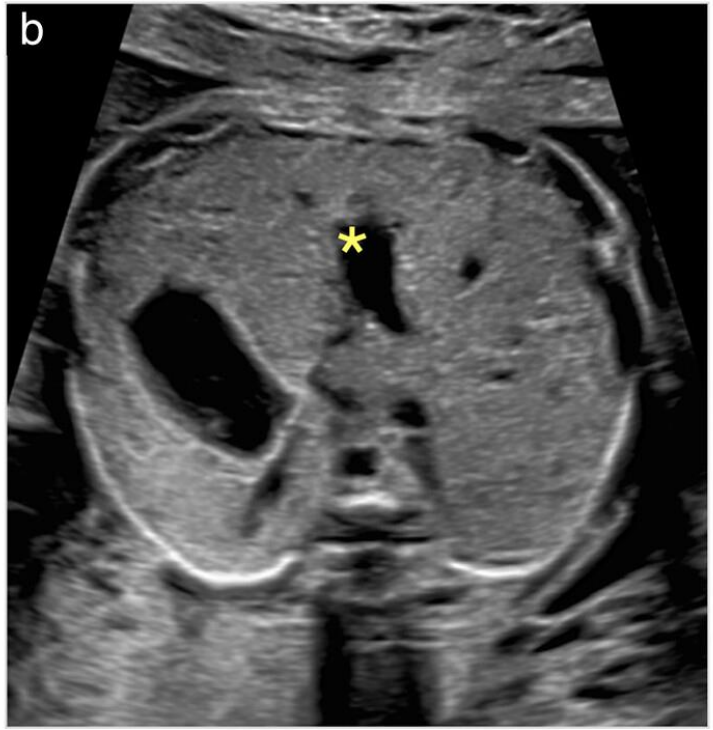
- What is behind the heart ?

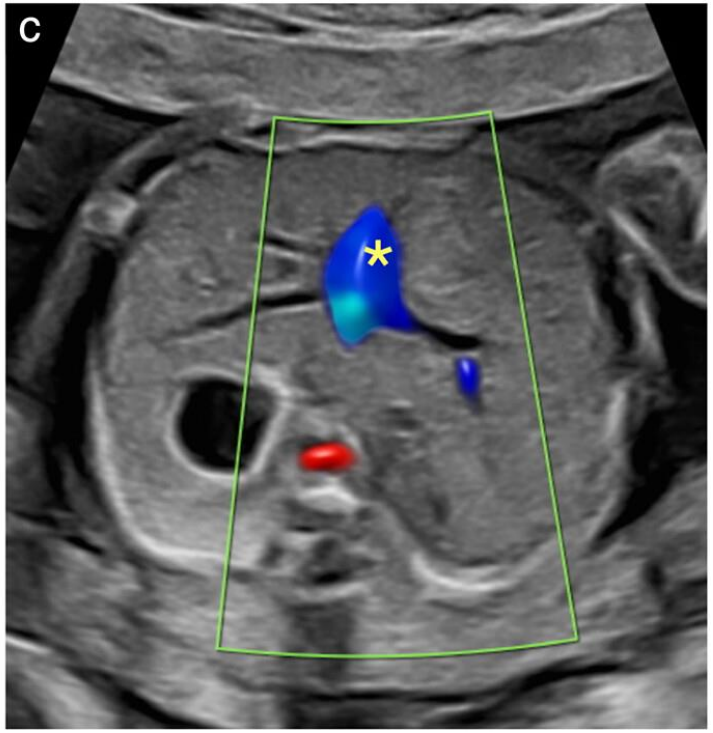
Ultrasound - position and technique



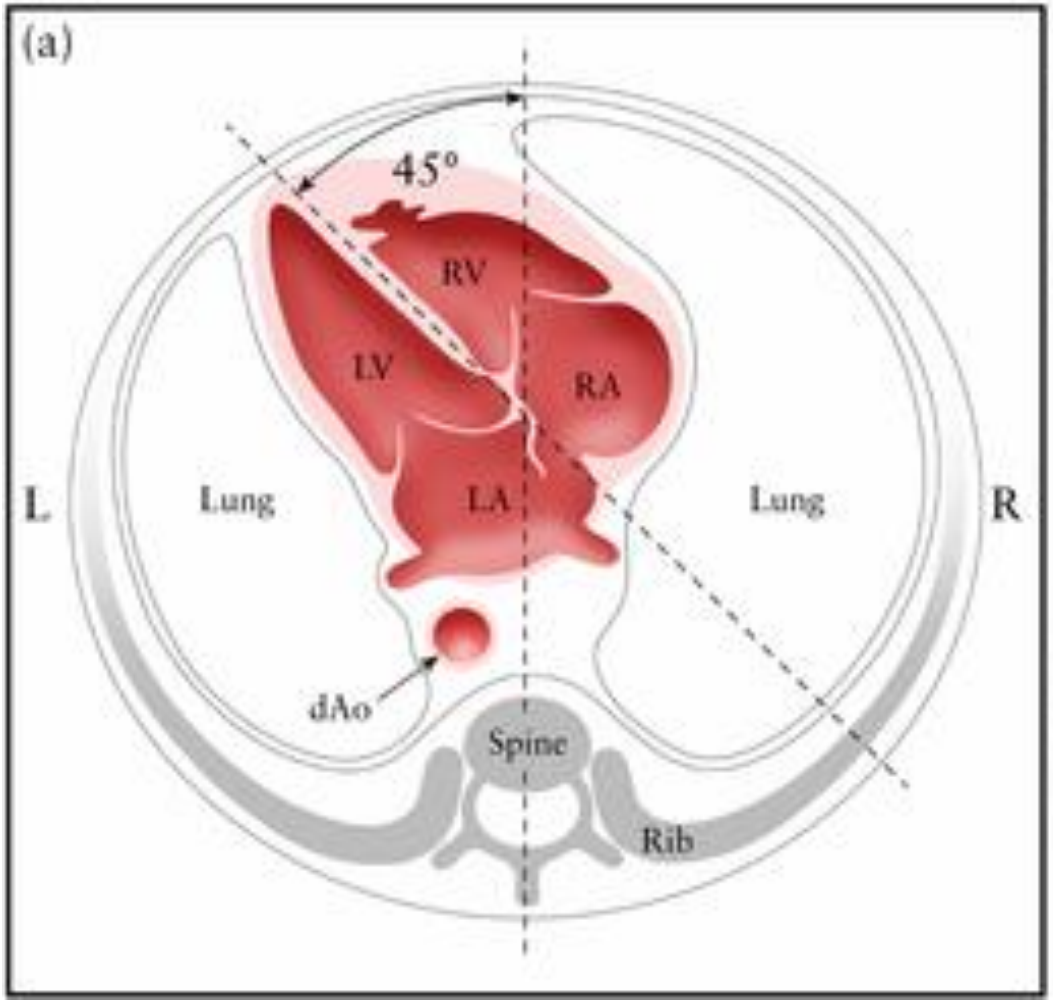
Establish orientation. Situs solitus.



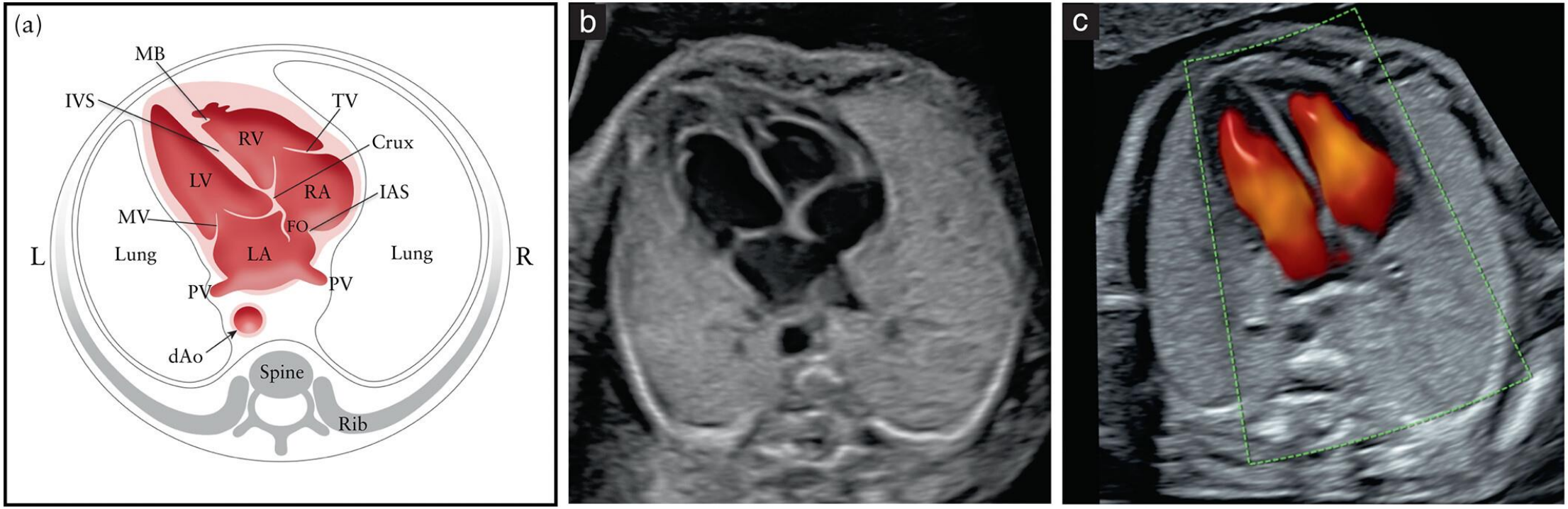




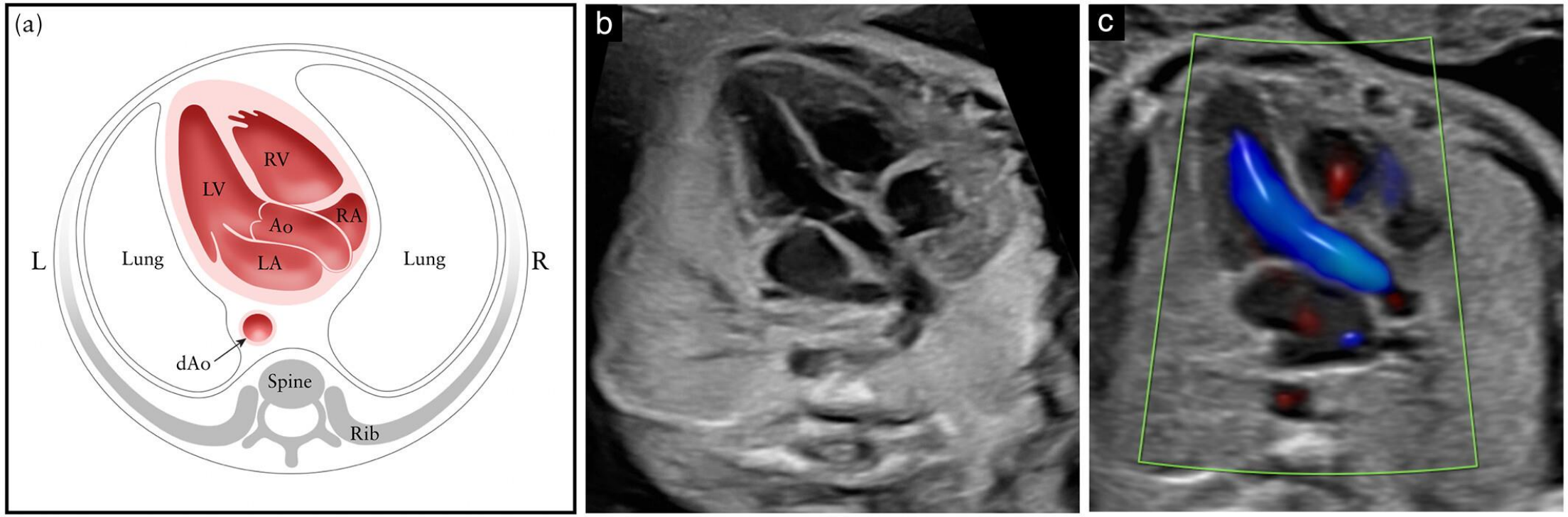
Four chamber view



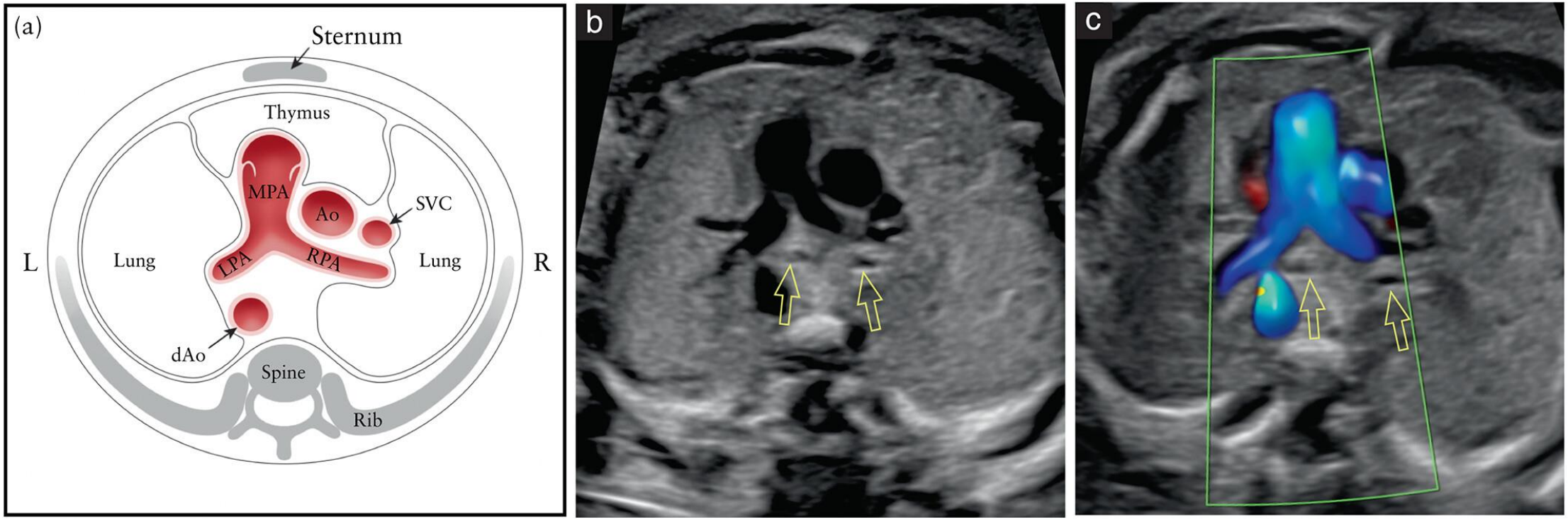
Further assessment of 4 chambers



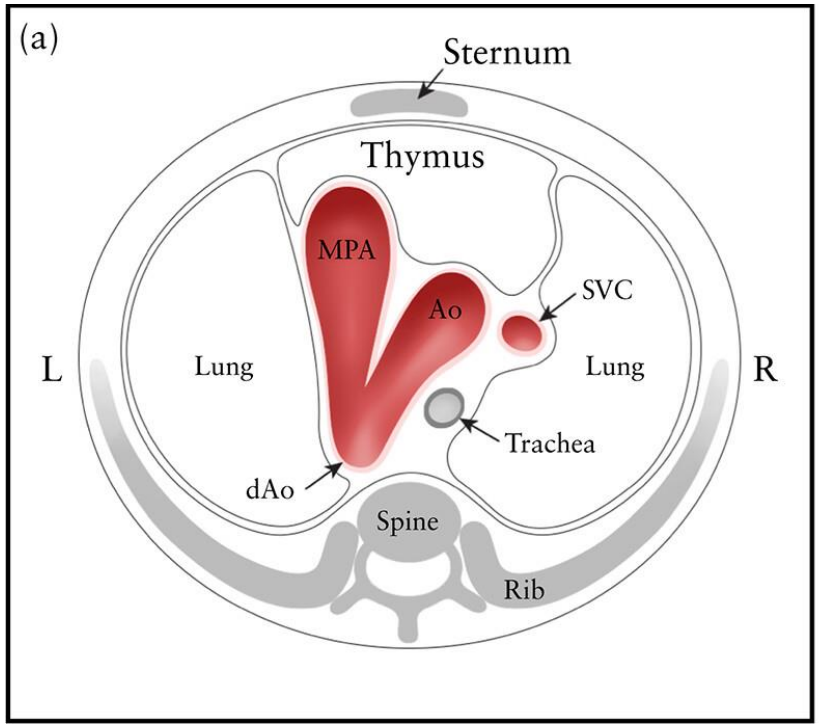
Left outflow tract.

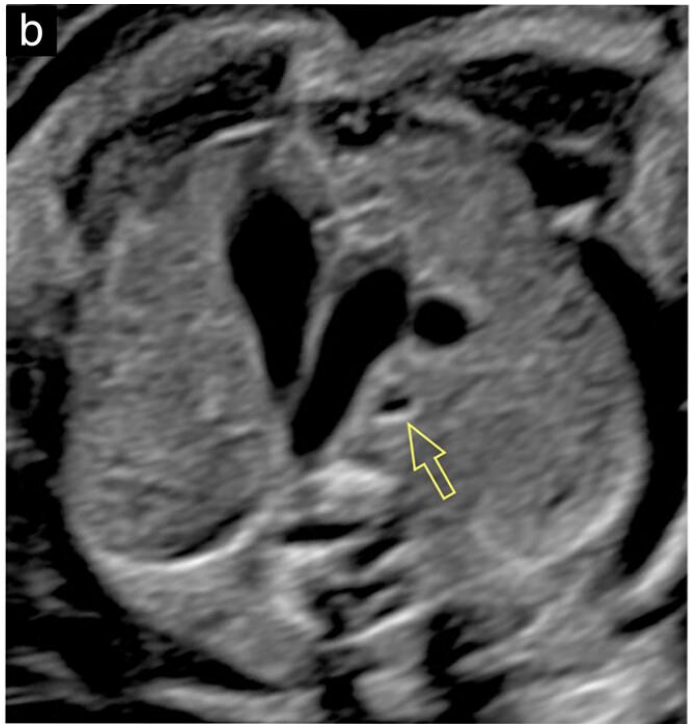


Right Outflow Tract (ROT). 3 vessel view (3VV)



ROT / 3VV


(a)
Sternum
Thymus
MPA
Ao
SVC
L
Lung
Lung
R
Trachea
dAo
Spine
Rib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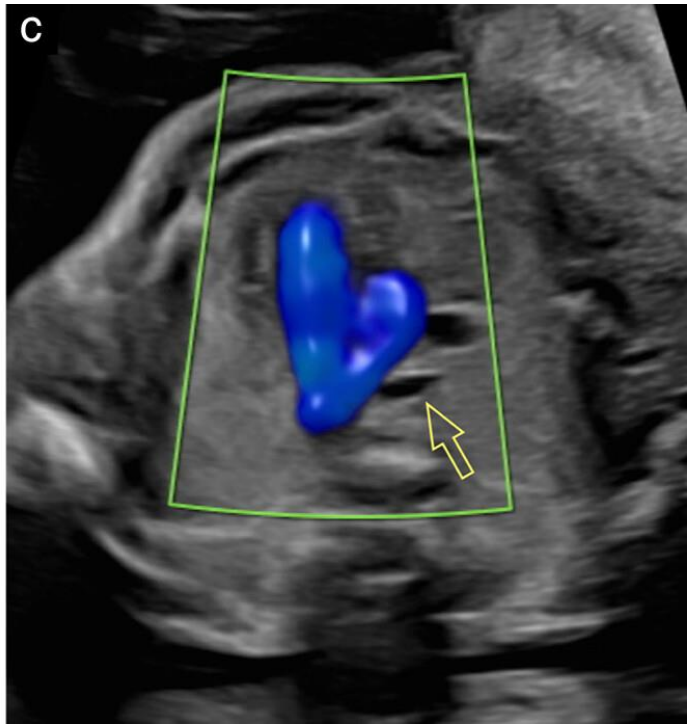
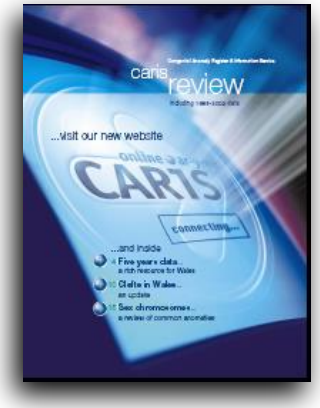

c

How to manage an anomaly in the DGH.

- Is amniocentesis indicated ?
- Assess the anomaly or fail to reach a diagnosis.
- Referral to a tertiary centre
- Fetal medicine MDT
- Is the conditional treatable / survivable ? Life limiting ? Lethal ?
- Serial assessment of anomaly and fetal growth. Shared follow up
- Where to deliver ? How to deliver ?
- Plan of care shared with both referring and receiving units.

CARIS

Congenital Anomaly Register and Information Service



Annual Meeting 2024

GIG CYMRU NHS WALES

Iechyd Cyhoeddus Cymru

Public Health Wales

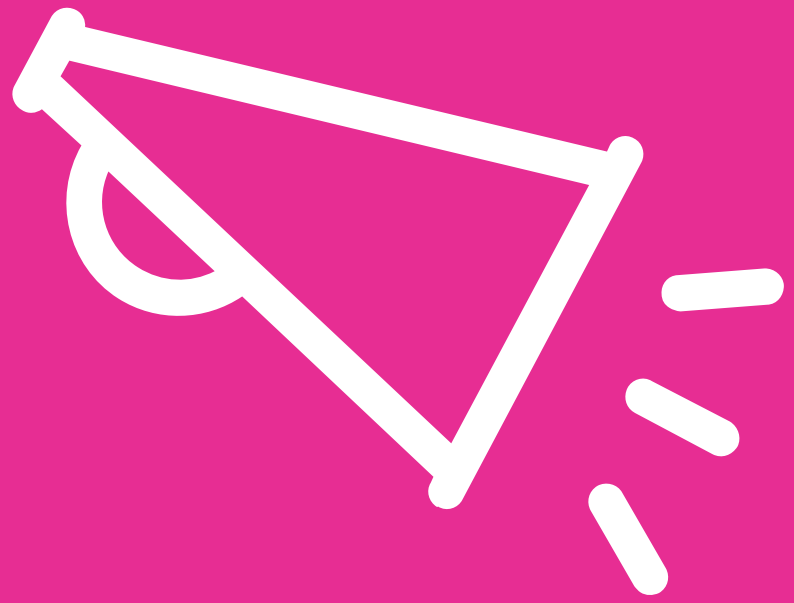
tiny tickers

a better start for tiny hearts

CARIS – Annual Meeting
26th November 2024

Anne Rhodes

Improving the early detection and care of babies with serious heart conditions



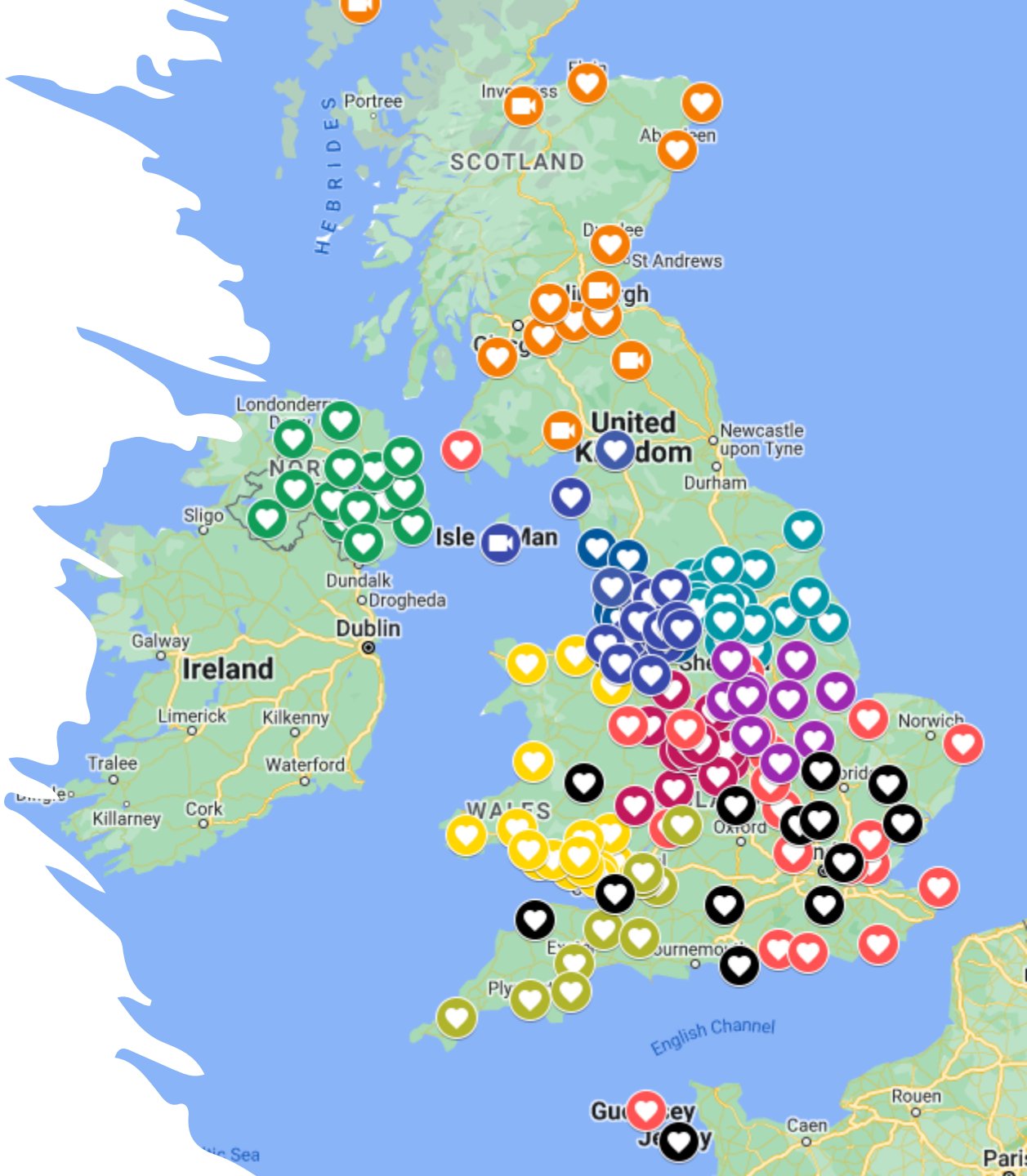
Our mission is

to help babies with serious heart conditions – giving them a better start in life.

Early detection means babies get the treatment they need from the first opportunity…

National prenatal detection rates of CHD are currently at 50%

Together, we want to get these rates up to 75%



Elements of training

FACE TO FACE

ONLINE

WEBINARS

UNIVERSITY LECTURES AND CONFERENCES

MATERIALS

HP NEWSLETTERS

We train sonographers and other health professionals

giving them the skills and confidence to spot heart defects during pregnancy or as soon as possible after birth.

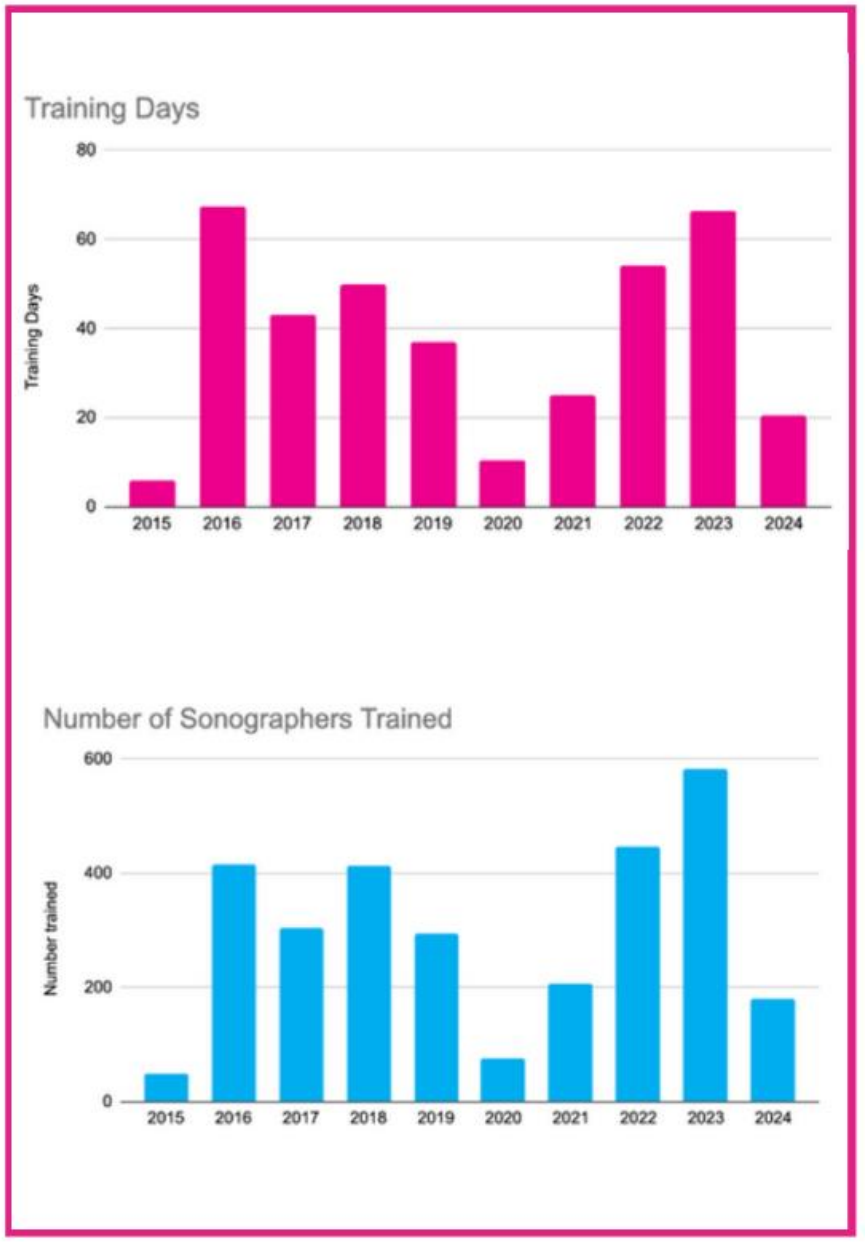
Face to Face training

- Regional projects
- National Training Fund
- Ad Hoc training

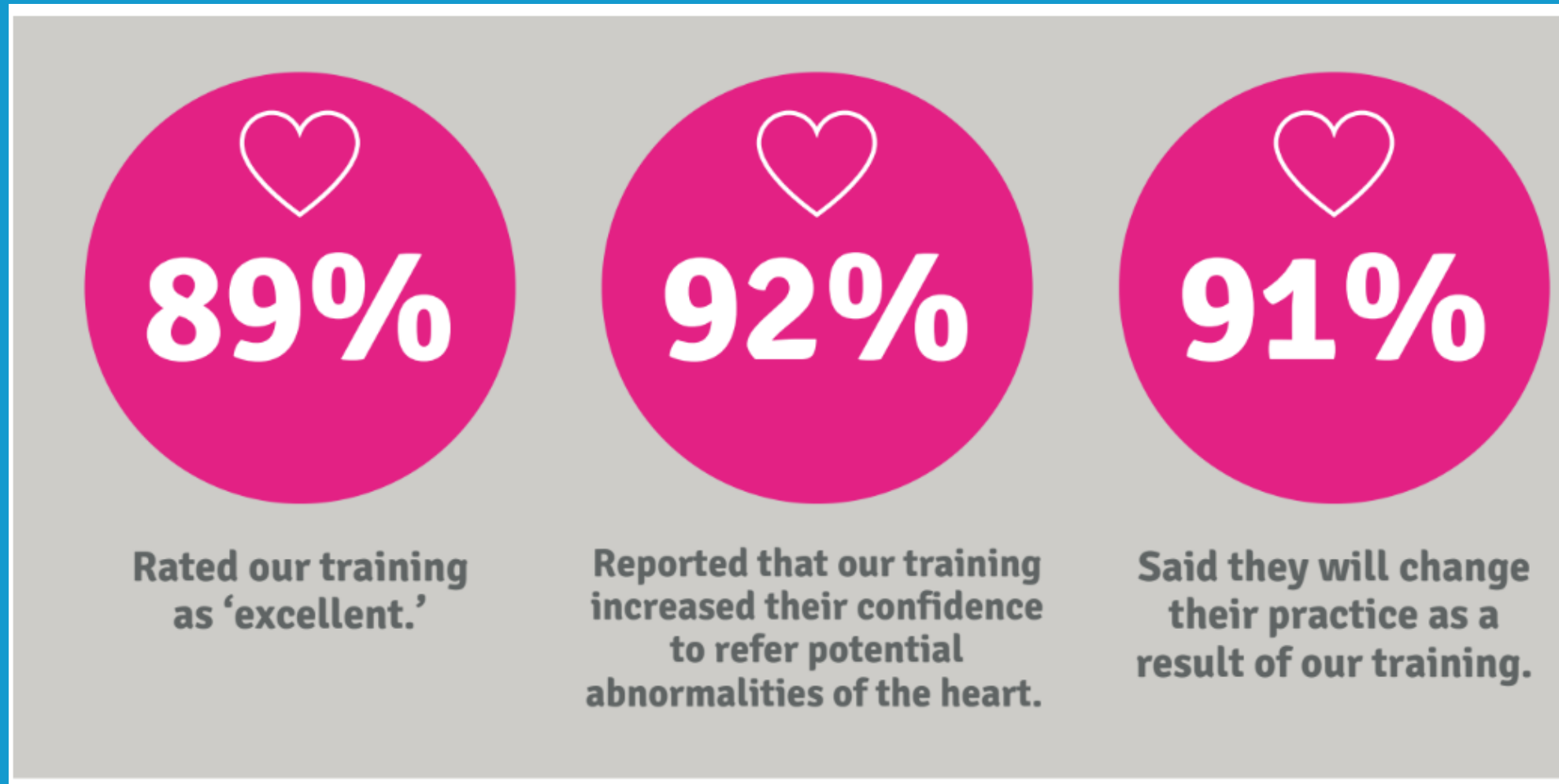
Materials

https://tinytickers-shop.square.site/s/shop


Hospitals that have received
Tiny Tickers training
Dublin
Ireland
Paris


Training Days
Training Days
80
60
40
20
0
2015
2016
2017
2018
2019
2020
2021
2022
2023
2024
Number of Sonographers Trained
Number trained
600
400
200
0
2015
2016
2017
2018
2019
2020
2021
2022
2023
2024

TRAINING FEEDBACK



We offer families the information and support they need

We spread awareness and give parents vital information about the signs of an undetected heart problem in their new baby.

FAMILY SUPPORT

Virtual Peer Support Groups

- Parent to be group
- Parent group
- Drop in sessions

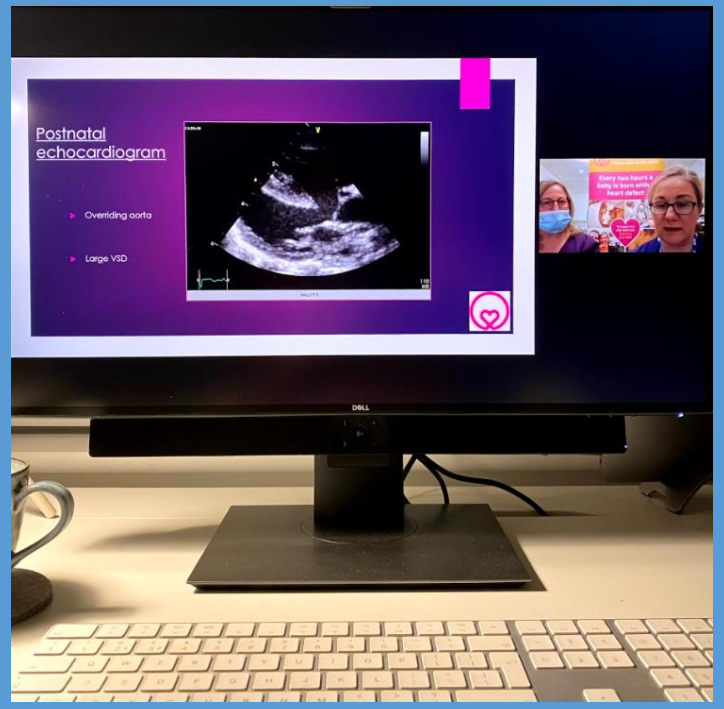
CARDIAC HUB FOR HEALTH PROFESSIONALS

- Medical abbreviations
- Detection rate data
- FASP leaflet
- Patient journey
- Information on conditions

CARDIAC HUB FOR FAMILIES

- How the heart develops
- Information on conditions
- UK paediatric heart units
- Visiting your specialist heart centre
- Family experience videos
- Dental care for children with CHD
- Bronchiolitis
- Mental health information for parents

WEBINARS



350

Health professionals registered for one or more condition-specific webinar

tiny tickers
a better start for tiny hearts

Think HEART

Know the signs
Think HEART

HEART RATE
Too fast or slow (normally 100-160 beats per min)?

ENERGY
Sleepy, quiet, floppy, tired or falling asleep feeding?

APPEARANCE
Pale, waxy, dusky, blue, purple, mottled or grey colour?

RESPIRATION
Breathing fast or slow (normally 40-60 breaths per min)?

TEMPERATURE
Persistently cold to touch - particularly hands and feet?

Think HEART was created by Dr Joan LaRovere, with the Royal Brompton, a specialist children's heart hospital.

609

Pulse oximetry machines placed in UK hospitals since 2017

PULSE OXIMETRY

We lobby decision-makers in the NHS

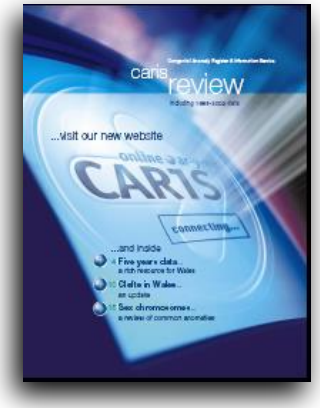
THANK YOU

Stay in touch with us on social media - we're on Twitter, Instagram, Linkedin, Facebook and YouTube!

@tinytickers

CARIS

Congenital Anomaly Register and Information Service



Annual Meeting 2024

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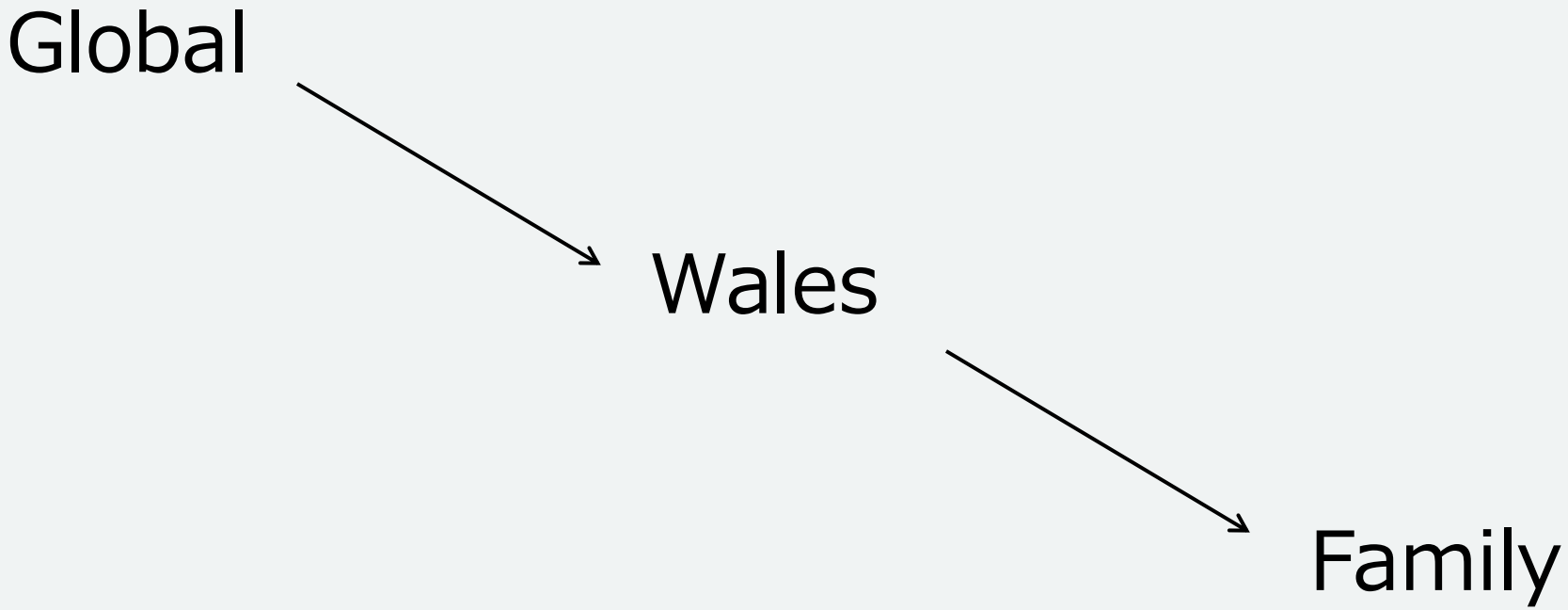
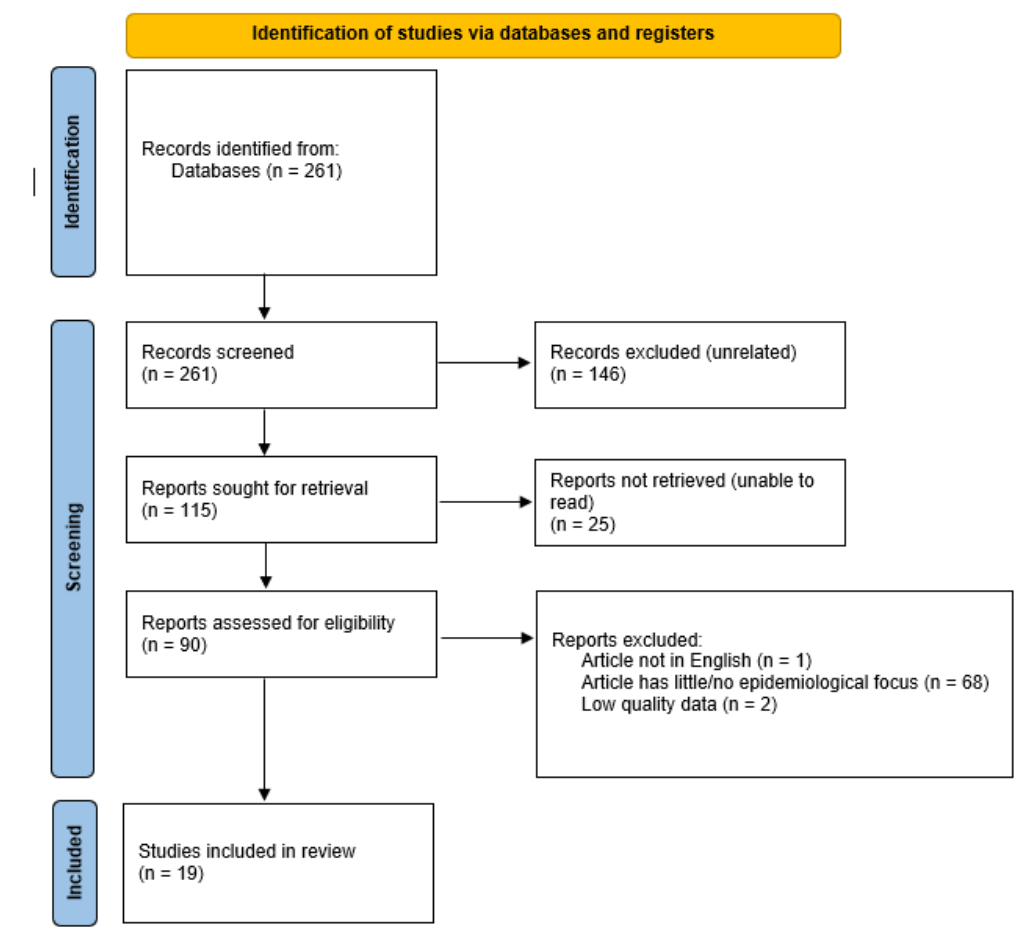
Public Health Wales


Epidermolysis
Bullosa
-John Helo, Cardiff University

Perspectives…




Global



PRISMA FLOW DIAGRAM

Incidence and Prevalence

Author	Country	Time period	Incidence (per 1 million live births)	Prevalence (per 1 million people)
Petrof et al.	England and Wales	2002-2021	67.8	34.8
Farokhforghani et al.	Iran	2021	Unavailable	6.72
Baardman et al.	Netherlands	1988-2018	41.30	22.4
Fine et al.	United States	1999-2016	19.57	11.07
Kho et al.	Australia	2010	Unavailable	10.3
Has et al.	Germany	2023	45.00	54.03
Gear et al.	New Zealand	2019	Unavailable	19.5
Raboei et al.	Saudi Arabia	2003-2020	24.51	Unavailable
Hernandez et al.	Spain	2013	6.00	Unavailable


Wales

Familial Characteristics

Category	Frequency	Percentage (%)
Maternal age at birth:<30	16	70
Maternal Age at birth:>=30	7	30
Consanguinity: Applicable	<5	-
Family History: Applicable	9	39
Family History: Not applicable	14	61
Number of previous pregnancies: <=2	18	78
Number of previous pregnancies: >2	5	22
Number of cases with co-occurring congenital anomalies	5	22

Child characteristics

Category	Frequency	Percentage (%)
Sex: Male	11	48
Sex: Female	12	52
Birth Year: 1998-2008	13	57
Birth Year: 2009-2018	10	43
Singleton Births	23	100
Live Births	23	100
Birth Weight: >=2500 grams	21	91
Birth Weight: <2500 grams	2	9
Gestational age: <37 weeks	3	13
Gestational age: >=37 weeks	20	87
1 Year Survival	22	96
10 Year Survival	21	91
When the case was first suspected: At birth	9	39
When the case was first suspected: Within 1 year of birth	8+9 (Cumulative 17)	74
When the case was first suspected: Over 1 year after birth	5+8+9 (Cumulative 22)	96


Family

Background

Patient: 1 year old M

Born 6 weeks early

Diagnosed with Recessive Dystrophic Epidermolysis Bullosa 10 days after birth by skin biopsy

Born with fused toes on the right foot + deformity causing right foot to be plantarflexed

Significant birth trauma on both hands, right knee and both lower extremities

Thrush on bottom at birth – 3 months to resolve

Mother is very involved in care

Complications

Pt NG tube fed from day one – led to an oesophageal stricture that was identified 3 months after birth

Episodes of haematemesis daily w/ dysphagia and SOB

Gastrostomy to replace NG tube in November 2023 – Primary source of nutrition

Eye abrasions – pruritic nature of EB. Approx. 10 total

Surgery in January 2024 for oesophageal stricture

All nails had fallen off by 2 months.

Adaptations

Only wears seamless clothing

Soft sheepskin lining play area and all other surfaces patient will touch

Conti sheets used to line nappy- ensured to not have direct nappy contact with skin

Dressings replaced by mother every day

Mother uses bio-oil over pts body – reduces pruritus

Baby loves swimming (with dressings on!)

Avoidable issues

Very rare condition – not often encountered clinically

ECG stickers taken off pts body left patches of missing skin

Strong adhesive tape to stabilise intubation resulted in lesions on pts face

Skin pulled during venepuncture, causing a lesion at the site

Take-away points

Children with EB have extremely fragile skin – what is ‘usual procedure’ for other patients can very easily cause significant trauma and subsequent scarring

Parents are very likely to be extremely involved in the care of the child – they are a very valuable source of information on how to care for the child, and including them in the care will reassure them


Thank you
Any questions?

Please remember to:

Continue reporting cases

- **By email to:** Caris.**Safehavenmailbox**@wales.nhs.uk
- **By internet:** nww2.nphs.wales.nhs.uk:8080/CARISWarningCard.nsf/WarningCardForm?OpenForm
- **By CARIS Cards or Data Forms**

Visit our website for prevalence data:

https://phw.nhs.wales/services-and-teams/caris/

Minimum of: NHS Number, date of birth & postcode

Thank you for attending

Please remember to complete the short feedback form that will be dropped in the chat shortly.

A certificate of attendance will be issued on request upon completion of this form.

CARIS

Congenital Anomaly Register and Information Service

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Iechyd Cyhoeddus Cymru

Public Health Wales