CARIS ANNUAL MEETINGS 2025

Combined Presentation Slides

Monday 24th November – The National Imaging Academy, Pencoed.

Microsoft Teams - Thursday 27th November 2025,.

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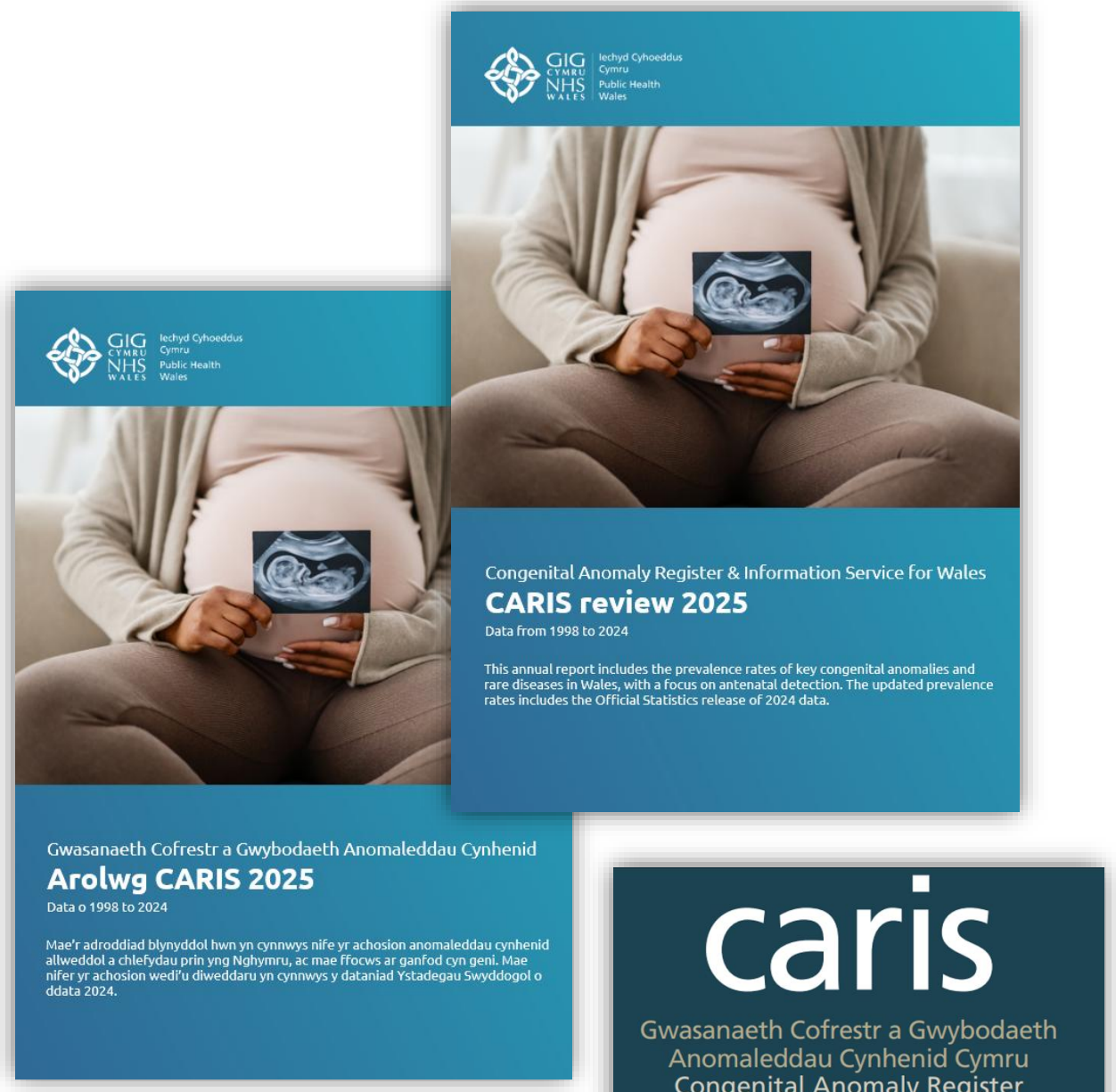
Iechyd Cyhoeddus Cymru
Public Health Wales



AGENDA

Face to Face event :

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Congenital Anomaly Register and Information Service - CARIS Annual Meeting 2025	
Date	**Monday 24th November 2025**
Time	**13:00 – 16:30**
Venue	**National Imaging Academy, Pencoed CF35 5HY**

13:00	**Welcome**	*Kinza Younas* *Consultant Obstetrician & Gynaecologist*
13:10	**CARIS update**	*Llion Davies* *Public Health Wales*
Focus session: Fetal Medicine Pathway **Chair: Kinza Younas**		
13:20	**Linking Antenatal Detection and CARIS data for quality assurance**	*Laura Macdermott* *Programme Coordinator* *Antenatal Screening Wales*
13.35	**Fetal Medicine Pathway** **&** **NT vs Cystic Hygroma**	*Armin Vandeperre* *Consultant in Obstetrics and Fetal Medicine*
14:05	**Hypoplastic left heart – prenatal diagnosis and postnatal management pathway**	*Alan Pateman* *Consultant Paediatric Cardiologist*
14:35	**CARIS Projects**	*Trainees and CARIS* *Asha and Samantha*
14:50	**Break**	
15:10	**Live scanning session**	*Armin Vanderperre* *Consultant in Obstetrics and Fetal Medicine*
16.00	**EUROCAT**	*David Tucker* *CARIS*
16:15	**Closing remarks**	

AGENDA

Virtual / Microsoft teams:
The meeting will be starting shortly...

Please make sure that your microphones are muted and cameras switched off.

This meeting will be recorded.

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Congenital Anomaly Register and Information Service - CARIS Annual Meeting 2025 Virtual Session	
Date	**Thursday November 27th 2025**
Time	**12:00 – 14:00**
Venue	**MS Teams**

12:00	**Welcome**	*Kinza Younas* *Consultant Obstetrician and Gynaecologist*
12:05	**CARIS update**	*David Tucker / Llion Davies* *Public Health Wales*
Focus session: Fetal Medicine Pathway **Chair: Kinza Younas**		
12:15	**Linking Antenatal Detection and CARIS data for quality assurance**	*Laura Macdermott* *Programme Coordinator* *Antenatal Screening Wales*
12:30	**Antenatal detection and in utero treatment**	*Asma Khalil* *Prof of Fetal Medicine* *St George's & Liverpool Womens Hospital*
13:10	**Fetal medicine pathway**	*Armin Vanderperre* *Consultant in Obstetrics and Fetal Medicine*
13:30	**Nuchal Translucency vs Cystic Hygroma**	*Mark Denbow* *Consultant in Fetal Medicine*
13:50	**Questions and closing remarks**	
14:00	**Close**	

CHAIR'S WELCOME

PROF. KINZA YOUNAS – (CLINICAL LEAD, CARIS)

MBBS, FRCOG , M.D.(UK),DFRSH.FHEA, MAS Dip(G)

EFRM (B)

CONSULTANT OBSTETRICIAN AND GYNAECOLOGIST, SBUHB.

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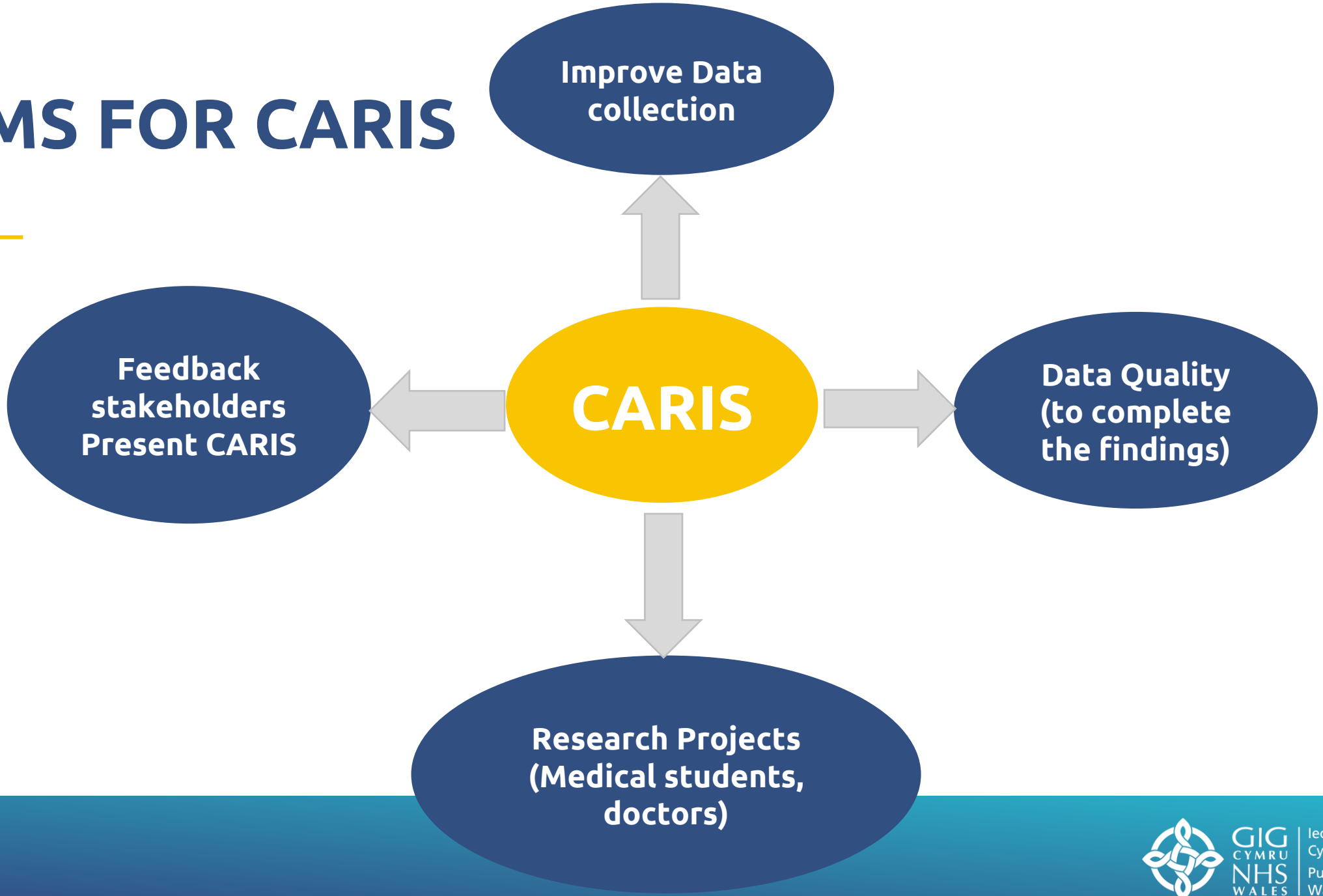
CHAIR'S WELCOME

Kind request

- Please remember to switch off cameras and microphones.
- This session is being recorded and will be published on our web page.
- There should be time at the end of each session for questions, but you can add them to the chat for us to address. If we don’t have time to respond in the meeting, we will answer any outstanding questions afterwards.
- We will provide CPD certificates upon receipt of completed feedback so please look out for the links in the chat.

AIMS FOR CARIS



DATA COLLECTION AND QUALITY

- **Clinical Governance & Quality Assurance**

Clinical oversight of case ascertainment

Ensure high standards for data accuracy, completeness, and consistency

Expert review of complex or borderline cases

- **Strategic leadership**

Advise stakeholders on trends, risks, and service implications

- Oversee policies and governance frameworks

CONTINUED.

- Clinical point of contact
- Engage colleagues' regular input via data sharing
- Annual reports and annual meetings

Measure, Evaluate, Recognize Engagement

CONTINUED.

- **•Facilitate high-quality research using CARIS data**
- •Review research proposals for clinical relevance, feasibility, and ethical considerations
- •Promote academic outputs, presentations, and multicenter collaborations.
- **Showcase CARIS projects and activates**
- •Represent CARIS in regional, National, and International networks(including congenital anomaly surveillance groups and public-health bodies.

GOALS

- Every year different themes to address important topics
- **“Antenatal Congenital anomalies detection in Wales”.**
- Engagement of participants, sharing knowledge, exploring new themes for research.
- Leading towards meaningful discussion and new collaboration
- Road leads to safe cost-effective clinical care for patients.

CARIS TEAM UPDATE

DR. LLION DAVIES - Consultant in Public Health Medicine.
DAVID TUCKER – CARIS Team Manager

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Public Health Wales

CARIS ANNUAL STATISTICS –

Published 10 November 2025

Congenital anomaly rate reduced stepwise since 1998

Years	Rate per 10,000 births
2005-2009	523.6
2010-2014	478.1
2015-2019	417.0
2020-2024	340.3*

** Expected to increase as further retrospective cases added for these most recent years*

?true reduction or under-reporting

CARIS Case Ascertainment

	1998-2024	2015-2024	Comment
Singular Anomaly % cases	61.5%	56.9%	Ascertainment proxy
Live Births % cases	84.2%	79.4%	↓ ascertainment of less serious cases?

- ?Covid impact
- ?Attendance impact

PLEASE REMEMBER TO…

Continue reporting **cases** to CARIS

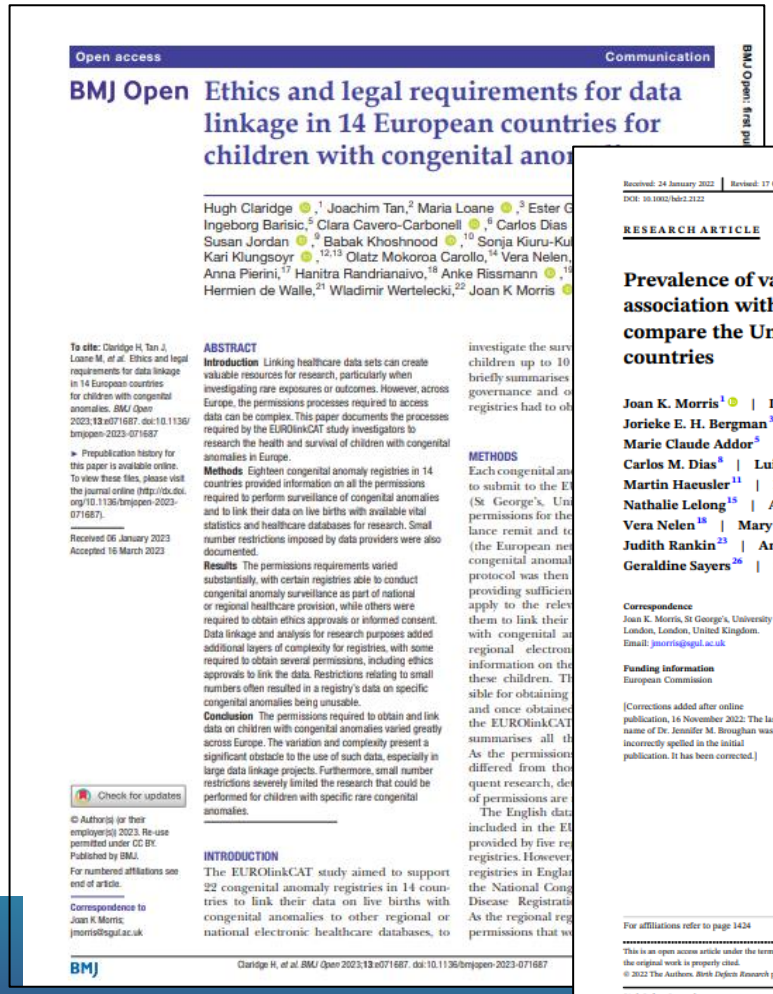
- By email to: caris.safehavenmailbox@wales.nhs.uk
- **Digitally: by visiting our report to CARIS QR links OR page**
- **https://phw.nhs.wales/services-and-teams/caris/contact-caris/**
- Traditional mail (Full case data) with downloadable forms
- E-Forms – for quick flagging of cases or full case reports
- Maternal infections – HIV and Syphilis case reporting

ENGLISH

CYMRAEG

CARIS RECENT RESEARCH CONTRIBUTION

15 articles 2024/25



Open access Communication

BMJ Open

Ethics and legal requirements for data linkage in 14 European countries for children with congenital ano

Hugh Claridge, Joachim Tan, Maria Loane, Ester G, Ingeborg Barisic, Clara Cavero-Carbonell, Carlos Dias, Susan Jordan, Babak Khoshnood, Sonja Kiuru-Ku, Kari Klungsoyr, Olatz Mokoroa Carollo, Vera Nelen, Anna Pierini, Hanitra Randrianaivo, Anke Rissmann, Hermien de Walle, Wladimir Wertelecki, Joan K Morris

ABSTRACT

Introduction Linking healthcare data sets can create valuable resources for research, particularly when investigating rare exposures or outcomes. However, across Europe, the permissions processes required to access data can be complex. This paper documents the processes required by the EUROlinkCAT study investigators to research the health and survival of children with congenital anomalies in Europe.

Methods Eighteen congenital anomaly registries in 14 countries provided information on all the permissions required to perform surveillance of congenital anomalies and to link their data on live births with available vital statistics and healthcare databases for research. Small number restrictions imposed by data providers were also documented.

Results The permissions requirements varied substantially, with certain registries able to conduct congenital anomaly surveillance as part of national or regional healthcare provision, while others were required to obtain ethics approvals or informed consent. Data linkage and analysis for research purposes added additional layers of complexity for registries, with some required to obtain several permissions, including ethics approvals to link the data. Restrictions relating to small numbers often resulted in a registry's data on specific congenital anomalies being unusable.

Conclusion The permissions required to obtain and link data on children with congenital anomalies varied greatly across Europe. The variation and complexity present a significant obstacle to the use of such data, especially in large data linkage projects. Furthermore, small number restrictions severely limited the research that could be performed for children with specific rare congenital anomalies.

INTRODUCTION

The EUROlinkCAT study aimed to support 22 congenital anomaly registries in 14 countries to link their data on live births with congenital anomalies to other regional or national electronic healthcare databases, to

METHODS

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To cite: Claridge H, Tan J, Loane M, et al. Ethics and legal requirements for data linkage in 14 European countries for children with congenital anomalies. BMJ Open 2023;13:e071687. doi:10.1136/bmjopen-2023-071687

Prepublication history for this paper is available online. To view these files, please visit the journal online (http://dx.doi.org/10.1136/bmjopen-2023-071687).

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For numbered affiliations see end of article.

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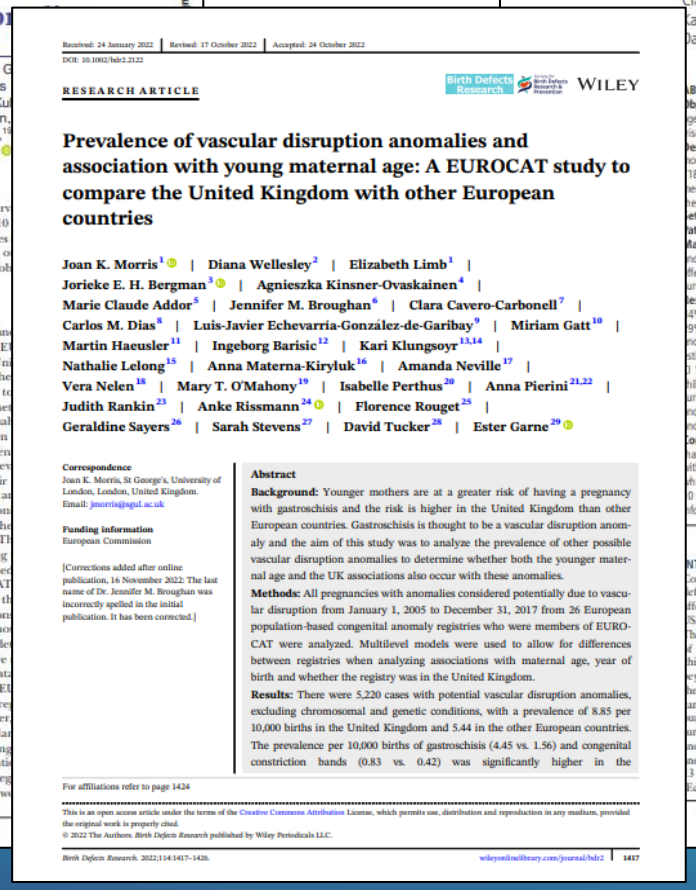

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DOI: 10.1002/bdr2.2122

RESEARCH ARTICLE

Birth Defects Research WILEY

Prevalence of vascular disruption anomalies and association with young maternal age: A EUROCAT study to compare the United Kingdom with other European countries

Joan K. Morris | Diana Wellesley | Elizabeth Limb | Jorieke E. H. Bergman | Agnieszka Kinsner-Ovaskainen | Marie Claude Addor | Jennifer M. Broughan | Clara Cavero-Carbonell | Carlos M. Dias | Luis-Javier Echevarría-González-de-Garibay | Miriam Gatt | Martin Haeusler | Ingeborg Barisic | Kari Klungsoyr | Nathalie Lelong | Anna Materna-Kiryluk | Amanda Neville | Vera Nelen | Mary T. O'Mahony | Isabelle Perthus | Anna Pierini | Judith Rankin | Anke Rissmann | Florence Rouget | Geraldine Sayers | Sarah Stevens | David Tucker | Ester Garne

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Funding information
European Commission

[Corrections added after online publication, 16 November 2022: The last name of Dr. Jennifer M. Broughan was incorrectly spelled in the initial publication. It has been corrected.]

Abstract

Background: Younger mothers are at a greater risk of having a pregnancy with gastroschisis and the risk is higher in the United Kingdom than other European countries. Gastroschisis is thought to be a vascular disruption anomaly and the aim of this study was to analyze the prevalence of other possible vascular disruption anomalies to determine whether both the younger maternal age and the UK associations also occur with these anomalies.

Methods: All pregnancies with anomalies considered potentially due to vascular disruption from January 1, 2005 to December 31, 2017 from 26 European population-based congenital anomaly registries who were members of EUROCAT were analyzed. Multilevel models were used to allow for differences between registries when analyzing associations with maternal age, year of birth and whether the registry was in the United Kingdom.

Results: There were 5,220 cases with potential vascular disruption anomalies, excluding chromosomal and genetic conditions, with a prevalence of 8.85 per 10,000 births in the United Kingdom and 5.44 in the other European countries. The prevalence per 10,000 births of gastroschisis (4.45 vs. 1.56) and congenital constriction bands (0.83 vs. 0.42) was significantly higher in the

For affiliations refer to page 1424

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Birth Defects Research. 2022;114:1417–1426. wileyonlinelibrary.com/journal/bdr2 1417

Original research

Ten-year survival of children with trisomy 13 or trisomy 18: a multi-registry European cohort study

Svetlana V Glinianaia, Judith Rankin, Joachim Tan, Maria Loane, Ester Garne, Clara Cavero-Carbonell, Hermien E K de Walle, Miriam Gatt, Mika Gissler, Kari Klungsøyr, Natalie Lelong, Amanda Neville, Anna Pierini, David F Tucker, Stine Kjaer Urhoj, Diana Gay Wellesley, Joan K Morris

ABSTRACT

Objective To investigate the survival to 10 years of age of children with trisomy 13 (T13) and children with trisomy 18 (T18), born 1995–2014.

Design Population-based cohort study that linked mortality data to data on children born with T13 or T18, including translocations and mosaicisms, from 13 member registries of EUROCAT, a European network for the surveillance of congenital anomalies.

Setting 13 regions in nine Western European countries.

Patients 252 live births with T13 and 602 with T18.

Main outcome measures Survival at 1 week, 4 weeks and 1, 5 and 10 years of age estimated by random-effects meta-analyses of registry-specific Kaplan-Meier survival estimates.

Results Survival estimates of children with T13 were 54% (95% CI 26% to 46%), 17% (95% CI 11% to 29%) and 11% (95% CI 6% to 18%) at 4 weeks, 1 and 10 years, respectively. The corresponding survival estimates were 38% (95% CI 31% to 45%), 13% (95% CI 10% to 17%) and 8% (95% CI 5% to 13%) for children with T18. The 10-year survival conditional on surviving to 4 weeks was 32% (95% CI 23% to 41%) and 21% (95% CI 15% to 28%) for children with T13 and T18, respectively.

Conclusions This multi-registry European study found that despite extremely high neonatal mortality in children with T13 and T18, 32% and 21%, respectively, of those who survived to 4 weeks were likely to survive to age 10 years. These reliable survival estimates are useful to inform counselling of parents after prenatal diagnosis.

WHAT IS ALREADY KNOWN ON THIS TOPIC

⇒ Children with trisomy 13 or trisomy 18 have extremely high neonatal and infant mortality.
⇒ A recent Canadian population-based study reported that about 13% of children with trisomy 13 and 10% with trisomy 18 may survive to age 10 years.
⇒ Long-term follow-up population-based studies of survival in children with trisomy 13 or trisomy 18 are lacking.

WHAT THIS STUDY ADDS

⇒ The majority of children born alive with trisomy 13 or trisomy 18 between 1995 and 2014 in 13 Western European regions died during the first 28 days of life: 66% of children with trisomy 13 and 62% with trisomy 18.
⇒ Survival at age 5 and 10 years was 16% (95% CI 10% to 26%) and 11% (95% CI 6% to 18%), respectively, for children with trisomy 13, and 10% (95% CI 7% to 14%) and 8% (95% CI 5% to 13%), respectively, for children with trisomy 18.
⇒ Ten-year survival conditional on surviving the first 28 days of life was 32% (95% CI 23% to 41%) and 21% (95% CI 15% to 28%) for trisomy 13 and trisomy 18, respectively.

HOW THIS STUDY MIGHT AFFECT RESEARCH, PRACTICE OR POLICY

⇒ This study demonstrates that reliable survival estimates can be obtained for children with rare anomalies by linking administrative mortality data to data on live births from European population-based congenital anomaly registries and combining results across registries. The results are important for counselling parents after prenatal diagnosis of these conditions.

INTRODUCTION

Congenital anomalies (CAs), including structural defects, chromosomal and genetic syndromes, affect about 2%–3% of births in Europe[1] and in the USA,[2] and are a leading cause of infant mortality.[3 4] They are also a growing contributor to mortality of children under 5 years of age[5] and of older children.[6] Survival of children with major CAs beyond 1 year has substantially improved during the last few decades due to advances in neonatal care and surgical interventions.[7 8] As shown in our recent multi-centre European study, 10-year survival exceeded 90% for most major structural anomalies and the most common chromosomal anomaly, Down syndrome (trisomy 21).[9] Trisomy 13 (T13) (Patau syndrome) and trisomy 18 (T18) (Edwards syndrome) are the most common autosomal trisomies after Down syndrome and are characterised by multiple structural anomalies and intellectual disability in survivors. The combined total prevalence including pregnancies resulting in a termination of pregnancy for fetal anomaly (TOPFA), stillbirths and live births varies from 5 to 10 per 10 000 births.[10–12] Children with T13 or T18 have a high mortality risk during the first weeks of life and the majority die during the first year.[11 13–17] Recent population-based US and Canadian studies

Glinianaia SV, et al. Arch Dis Child 2023;108:461–467. doi:10.1136/archdischild-2022-325068 461

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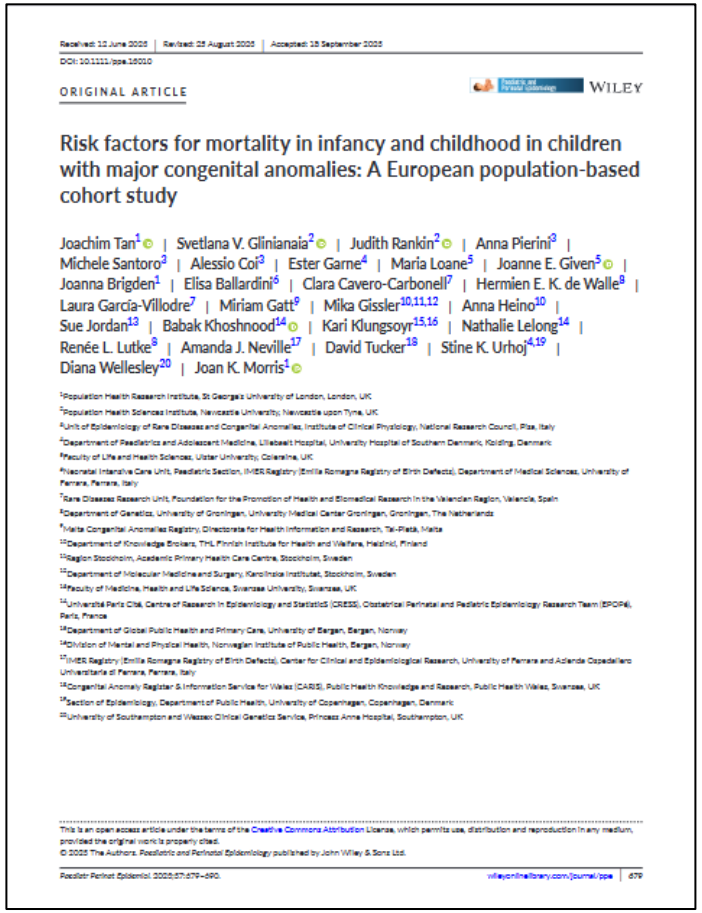

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DOI: 10.1111/ppe.13010

ORIGINAL ARTICLE

Paediatric and Perinatal Epidemiology WILEY

Risk factors for mortality in infancy and childhood in children with major congenital anomalies: A European population-based cohort study

Joachim Tan | Svetlana V. Glinianaia | Judith Rankin | Anna Pierini | Michele Santoro | Alessio Coi | Ester Garne | Maria Loane | Joanne E. Given | Joanna Brigden | Elisa Ballardini | Clara Cavero-Carbonell | Hermien E. K. de Walle | Laura García-Villodre | Miriam Gatt | Mika Gissler | Anna Heino | Sue Jordan | Babak Khoshnood | Kari Klungsoyr | Nathalie Lelong | Renée L. Lutke | Amanda J. Neville | David Tucker | Stine K. Urhoj | Diana Wellesley | Joan K. Morris

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CARIS ACTIVE PROJECTS

Academic

- Gastroschisis paper with Cardiff University

eurolinkcat
Establishing a linked European Cohort of Children with Congenital Anomalies

Health Protection

- Maternal Covid-19 and congenital anomalies
- Parvovirus

Improvement Work

- HIV and Syphilis – Microbiology, CARIS, CDSC

4 Nations - Newborn blood spot screening engagement

PARVOVIRUS IN WALES

PRESENTED BY: SAMANTHA FISHER –
CARIS SENIOR REGISTRATION OFFICER

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Public Health Wales

Parvovirus

Infection

- Also known as slapped cheek syndrome/fifth disease and is common in children
- General infection signs like fever and headache 7-10 days before classic rash. Generally non-infectious once rash appears.
- 50% adults have already had parvovirus infection and have immunity against it.

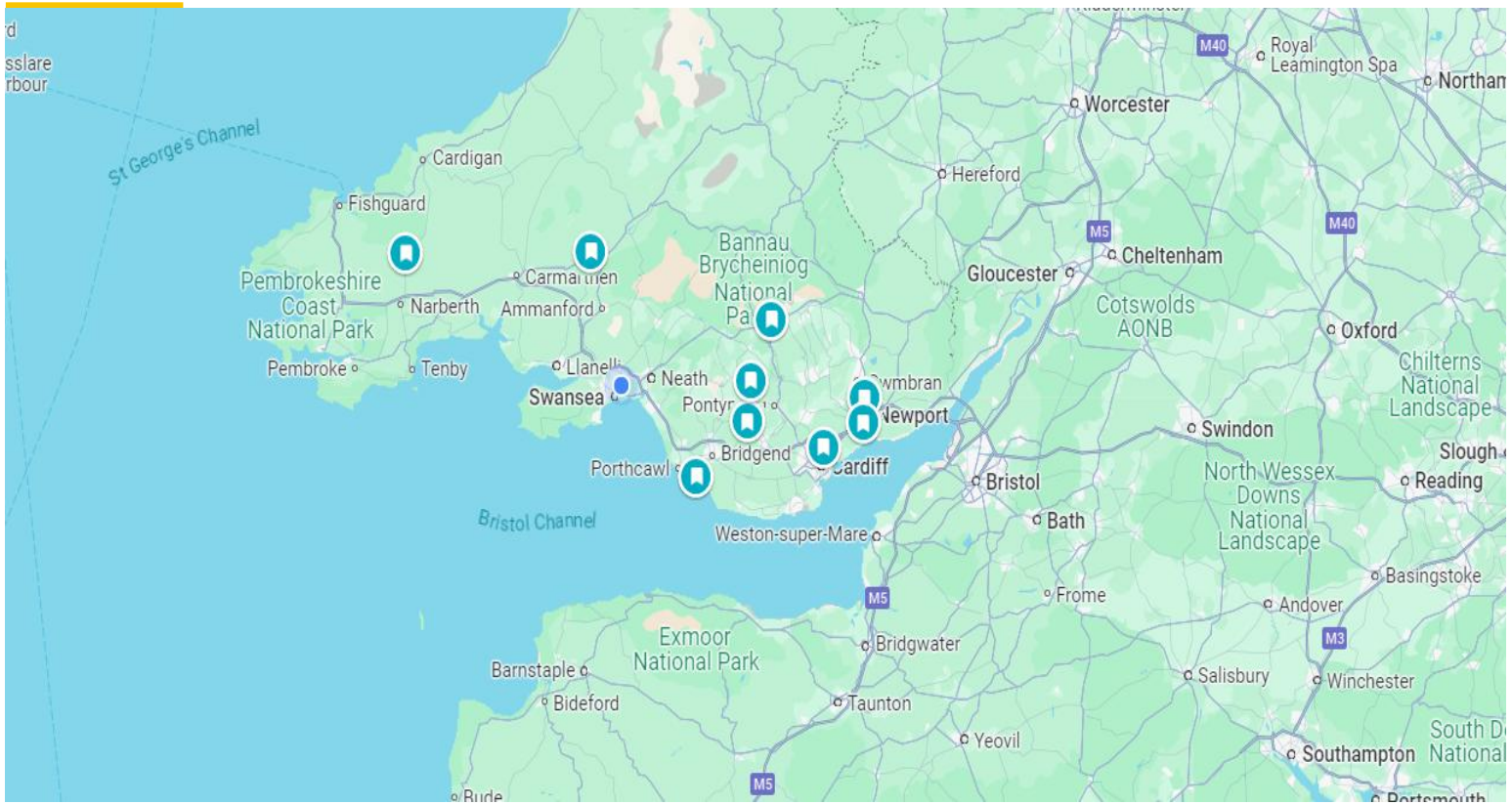
If a pregnant woman is infected there is a 30% risk of transmission to foetus

Infection in pregnancy is of most concern **before 26 weeks**, where it can be associated with:

- Fetal hydrops, congenital anaemia, pleural/pericardial effusions, intrauterine growth restriction
- Miscarriage or still birth

Parvovirus in Wales

Case monitoring



- We recognized an increase in Parvovirus cases during pregnancy in 2024, including some instances of spontaneous fetal loss.
- The geographic locations were plotted to see if a cluster emerged.
- The cases are distributed, however, are all in South Wales.
- We needed to check previous years data to confirm if cases were higher than usual

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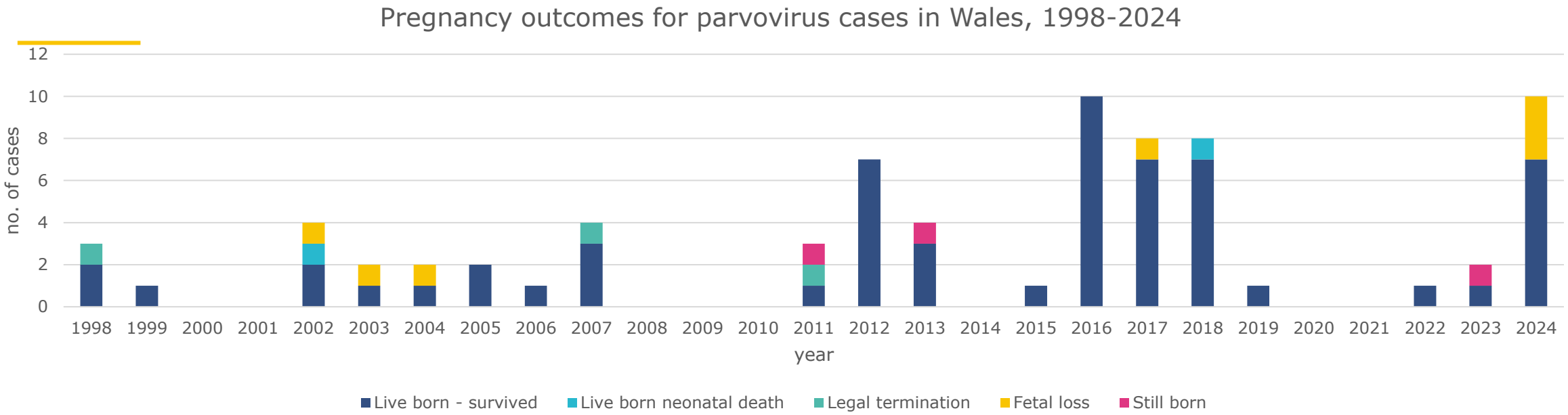
Parvovirus in Wales

Case numbers and outcomes



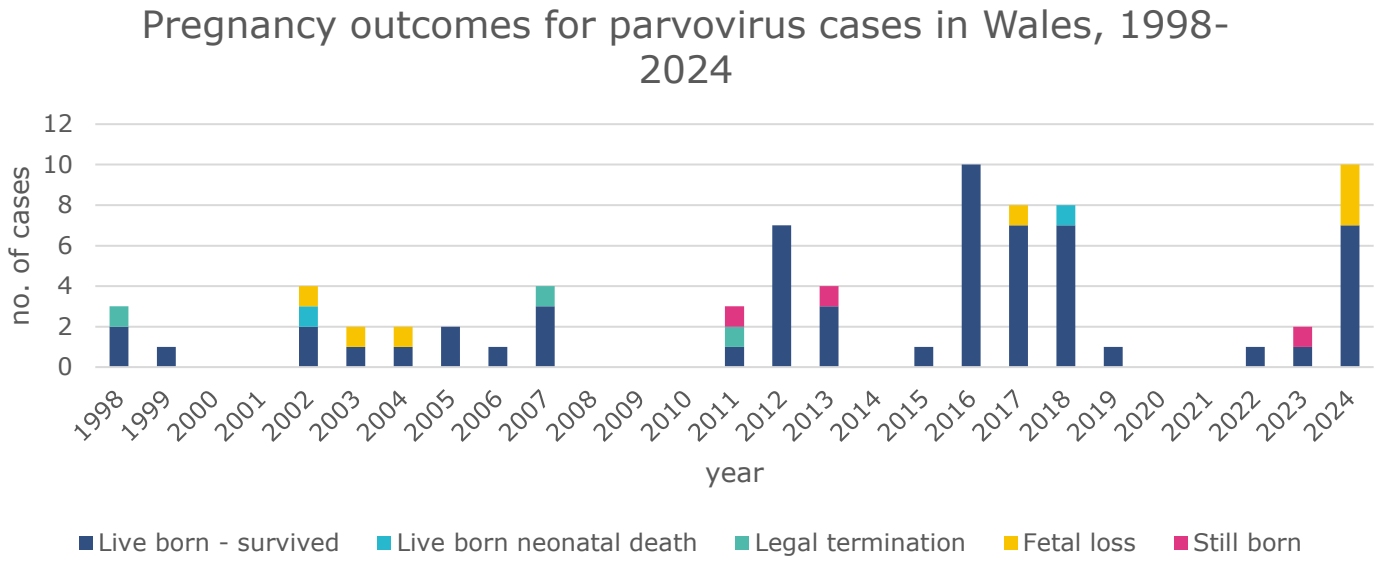
- 74 cases since CARIS began in 1998, 10 of these were in 2024
- The increase in cases over recent years could be due to improved reporting and recording in CARIS
- No cases in 2020 and 2021 but this is likely due to pandemic shielding and a natural decline in transmission
- Three spontaneous fetal losses reported in 2024 to date, which is higher than data from previous years

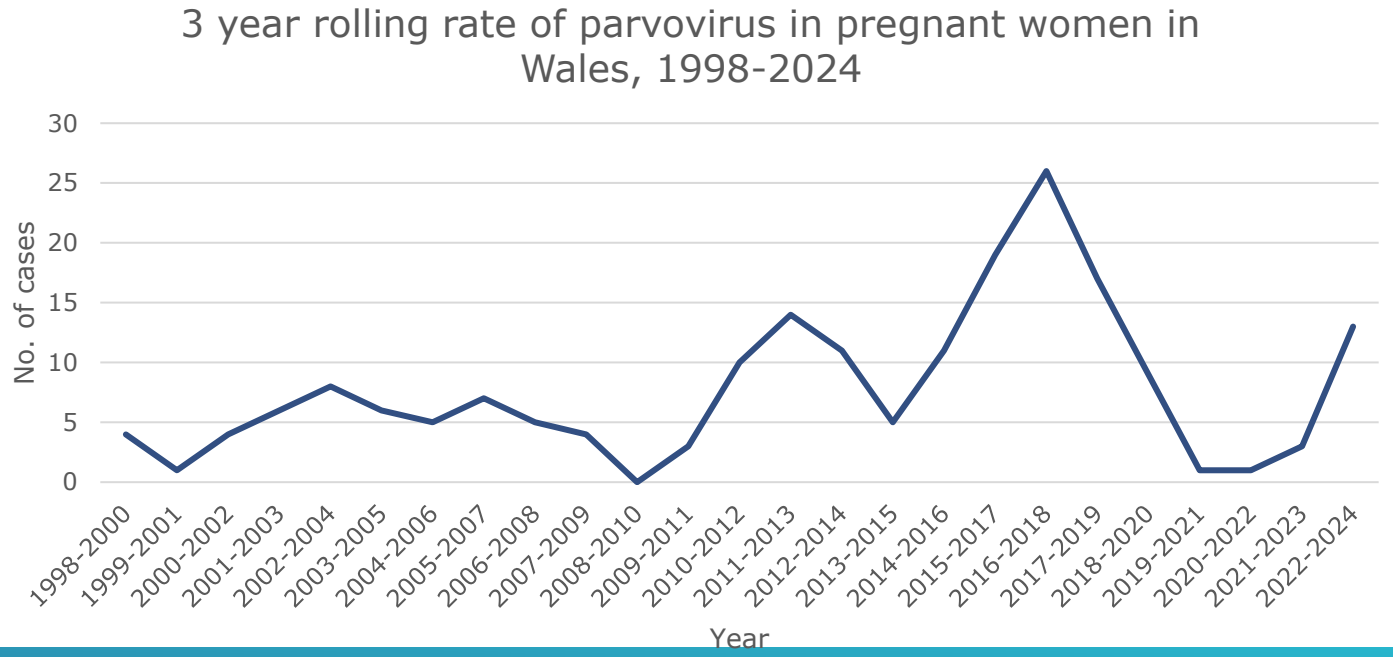
Parvovirus in Wales

3-year rolling rate

The 3-year rolling rate shows a trend where cases spike around every 4 years. This coincides with evidence found globally.

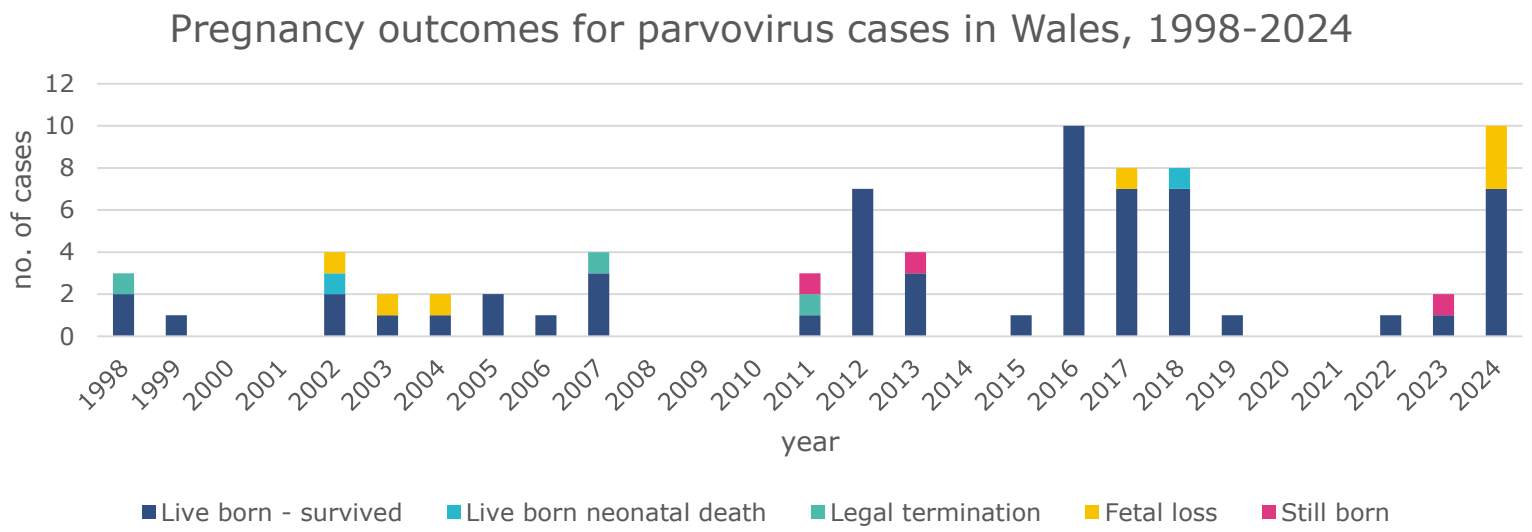
Pandemic shielding meant no cases in 2020-21, but a rebound of higher case numbers in 2024 has been noticed globally. Meaning we could see high case numbers again this spring before they decline again





Parvovirus in Wales

Summary of findings



- Contact with young children who are susceptible to contracting Parvovirus is a risk factor pregnant women
- 49/74 had another child
- 18 had a job that involved contact with children
- Health Protection advice should be to remind pregnant women to avoid contact with parvovirus if possible and be vigilant of symptoms.

- Only 4 of the 74 cases were in North Wales and none from Mid Wales. We need to find a way of accessing Mid and North Wales data
- Welsh Clinical Portal enables us to see lab results for Parvovirus testing which could be increasing our case numbers.

ASSESSING THE ASSOCIATION BETWEEN ARTIFICIAL REPRODUCTIVE TECHNOLOGIES AND CARDIAC AND GASTROINTESTINAL CONGENITAL ANOMALIES

AUTHOR: ASHA MAHAMED – TRAINEE IN PUBLIC HEALTH, CARDIFF UNIVERSITY

PRESENTED BY: PROF. KINZA YOUNAS

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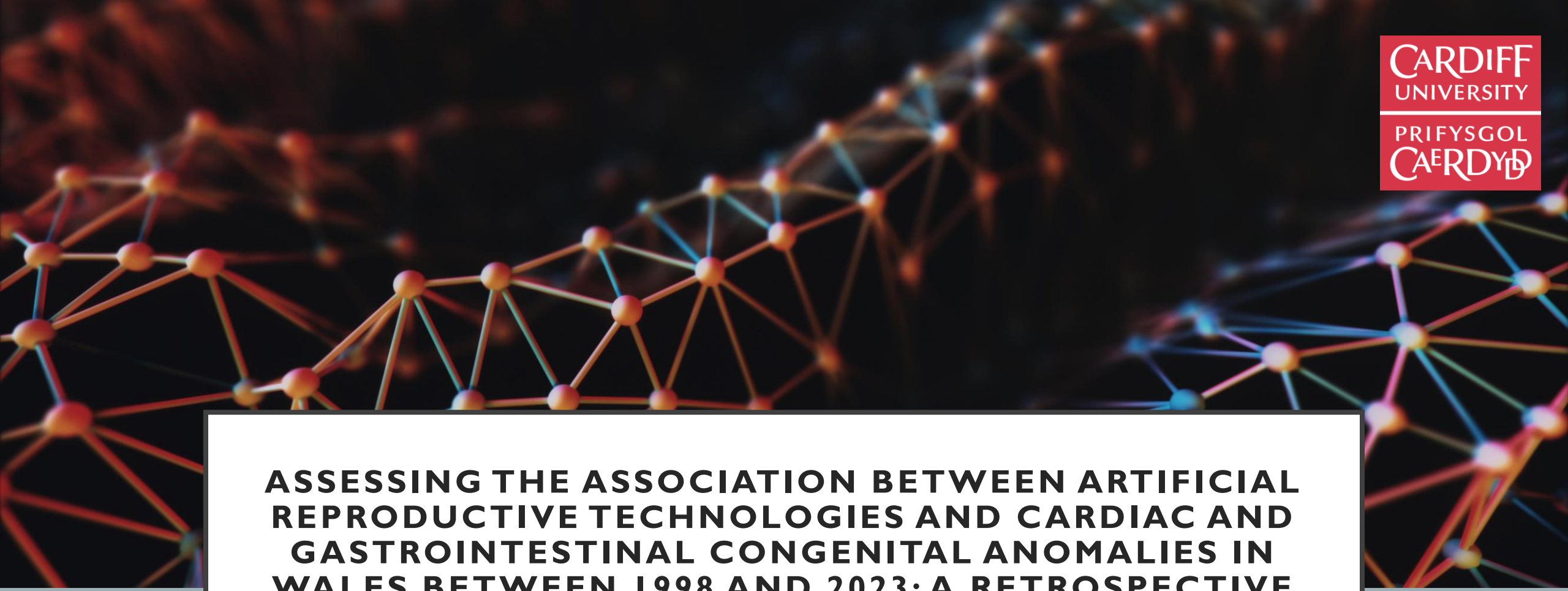


ASSESSING THE ASSOCIATION BETWEEN ARTIFICIAL REPRODUCTIVE TECHNOLOGIES AND CARDIAC AND GASTROINTESTINAL CONGENITAL ANOMALIES IN WALES BETWEEN 1998 AND 2023: A RETROSPECTIVE COHORT STUDY

Authors : Mahamed, A. , Tucker, D. , Hurt, L., Davies, L., Younas, K.

BACKGROUND

1

Subfertility affects 1 in 6 couples in the UK (Harris 2023)

2

6,700 IVF cycles were recorded in 1991 while in 2022, 77,000 cycles + 6,000 DI (Thornton 2024)

3

21% of IVF user are 40 years old and over in 2019 while in 1991 it is only 10% (HFEA 2021)

4

2019 the multiple birth rate reached 6%, falling from 28% in the 1990s (HFEA 2021)

CONGENITAL ANOMALIES AND ART

Research shows congenial anomaly risks are higher in ART pregnancies than conventional conception (Hoorsan et al. 2017)

In the UK, about 2 % or 1 in 59 live births have a congenital anomaly (NCARDRS 2021)

In Wales, 4.9% of both live and still births had at least one congenital anomaly (CARIS 2022)

Congenital anomalies have immediate, long term and wide-reaching impacts:

Trauma for both parents and child

Physical, Developmental, Social

Economic

Population	Women in Wales who used ART to conceive a pregnancy that resulted in a congenital anomaly that was reported to CARIS between 1998 and 2023
Intervention/exposure	ART focusing on risk factors including: ▪ Age (mother and father): under 35 years old and 35 years old and over ▪ Deprivation: Welsh Index of Multiple Deprivation (WIMD)
Comparison/control	Comparing between ART and non-ART cases: ▪ IUI- Intrauterine insemination ▪ GIFT- Gamete intrafallopian transfer ▪ IVF- invitro fertilisation ▪ ICSI- Intra cytoplasmic sperm injection
Outcomes	Congenital anomalies classified by ICD-10

METHODOLOGY

Baseline- Women in Wales who didn't use ART but had a pregnancy that resulted in a congenital anomaly that was reported to CARIS between 1998 and 2023

METHODOLOGY

RETROSPECTIVE COHORT STUDY

Data from CARIS was anonymised

Ethical approval was not required under CARIS governance permissions

Exclusion criteria used to reduce bias

IBM SPSS Version 29.0.2.0

Maternal and paternal age and level of deprivation

Descriptive

Logistic Regression

Unadjusted and adjusted

RESULTS

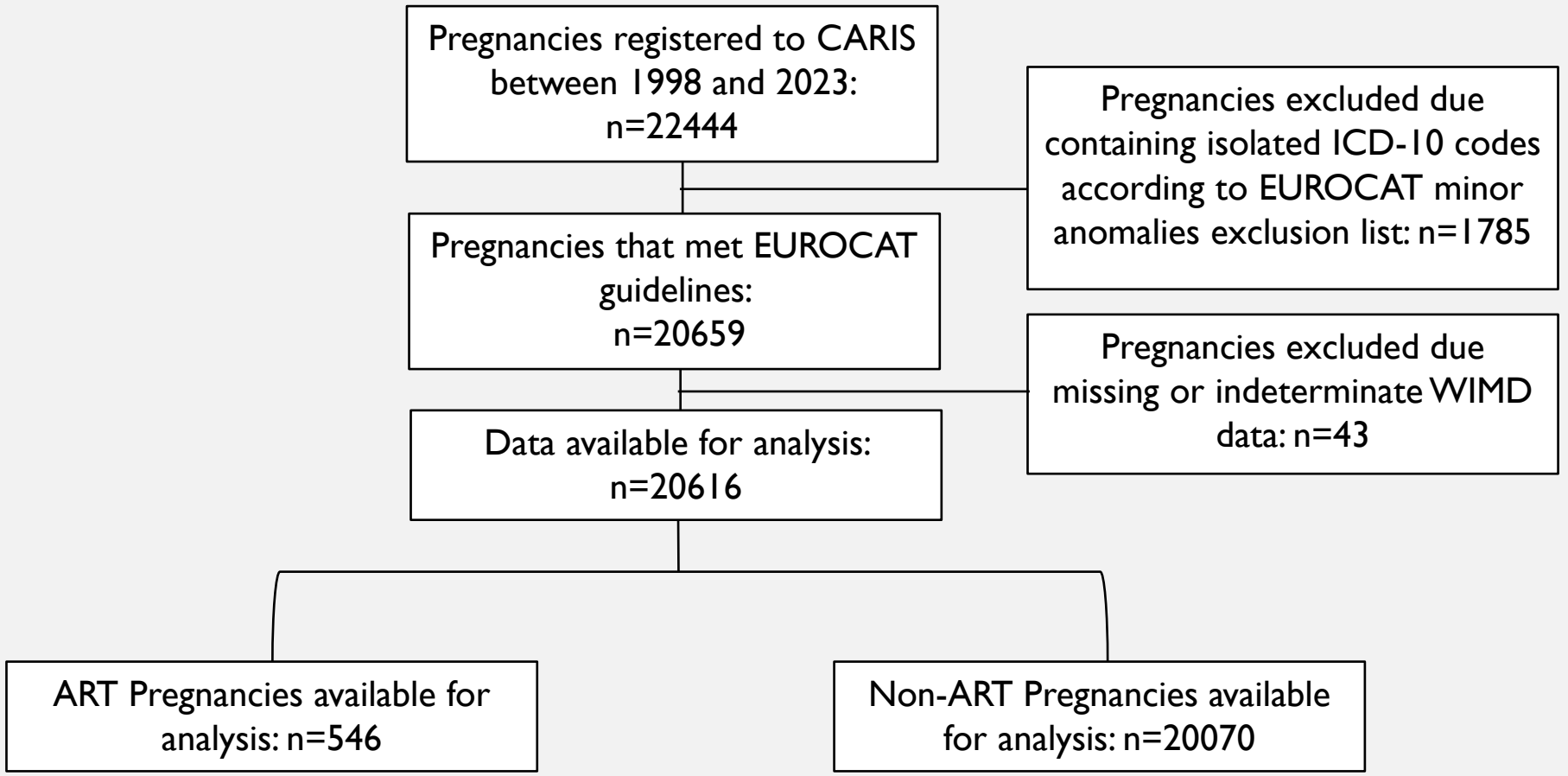
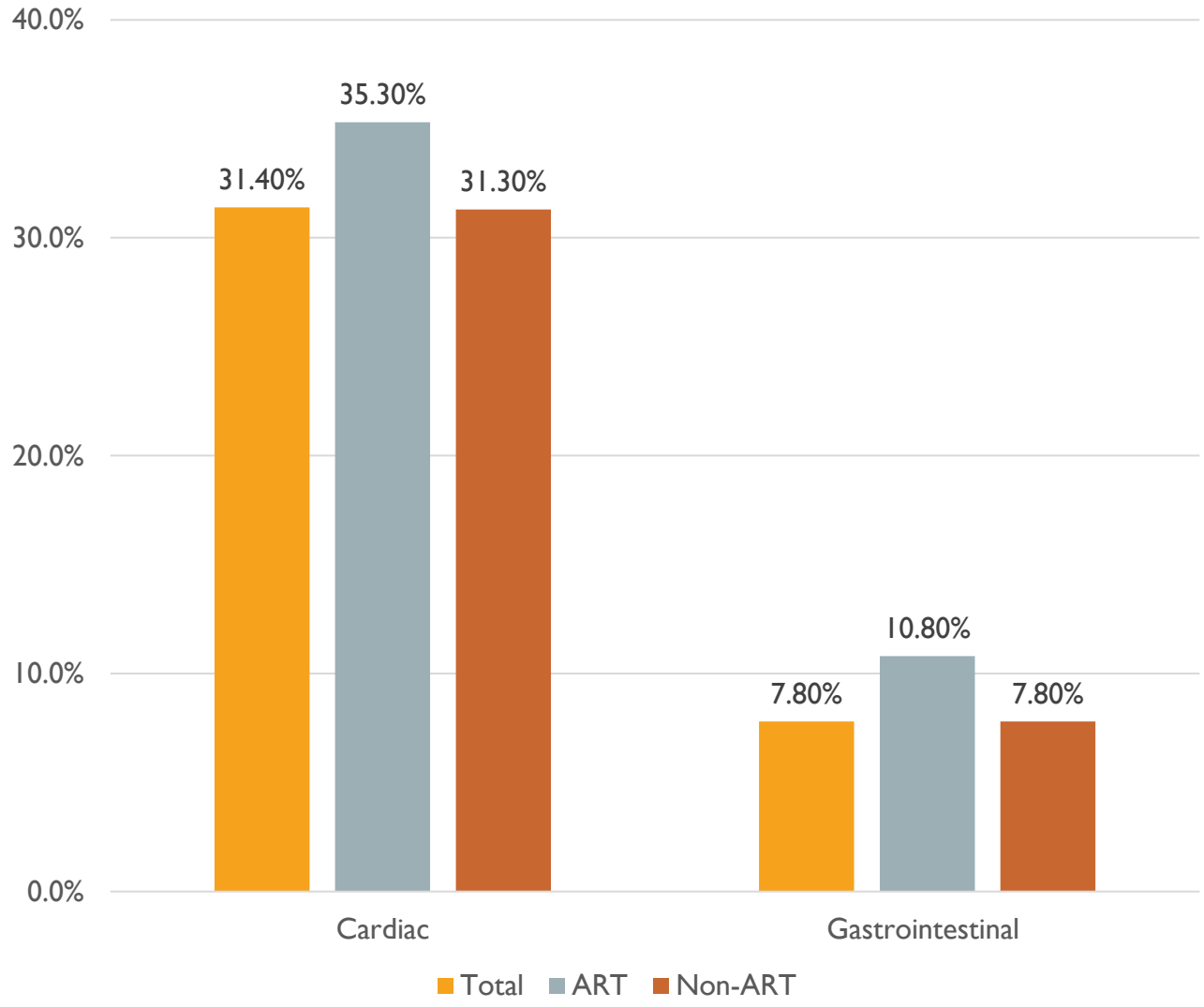
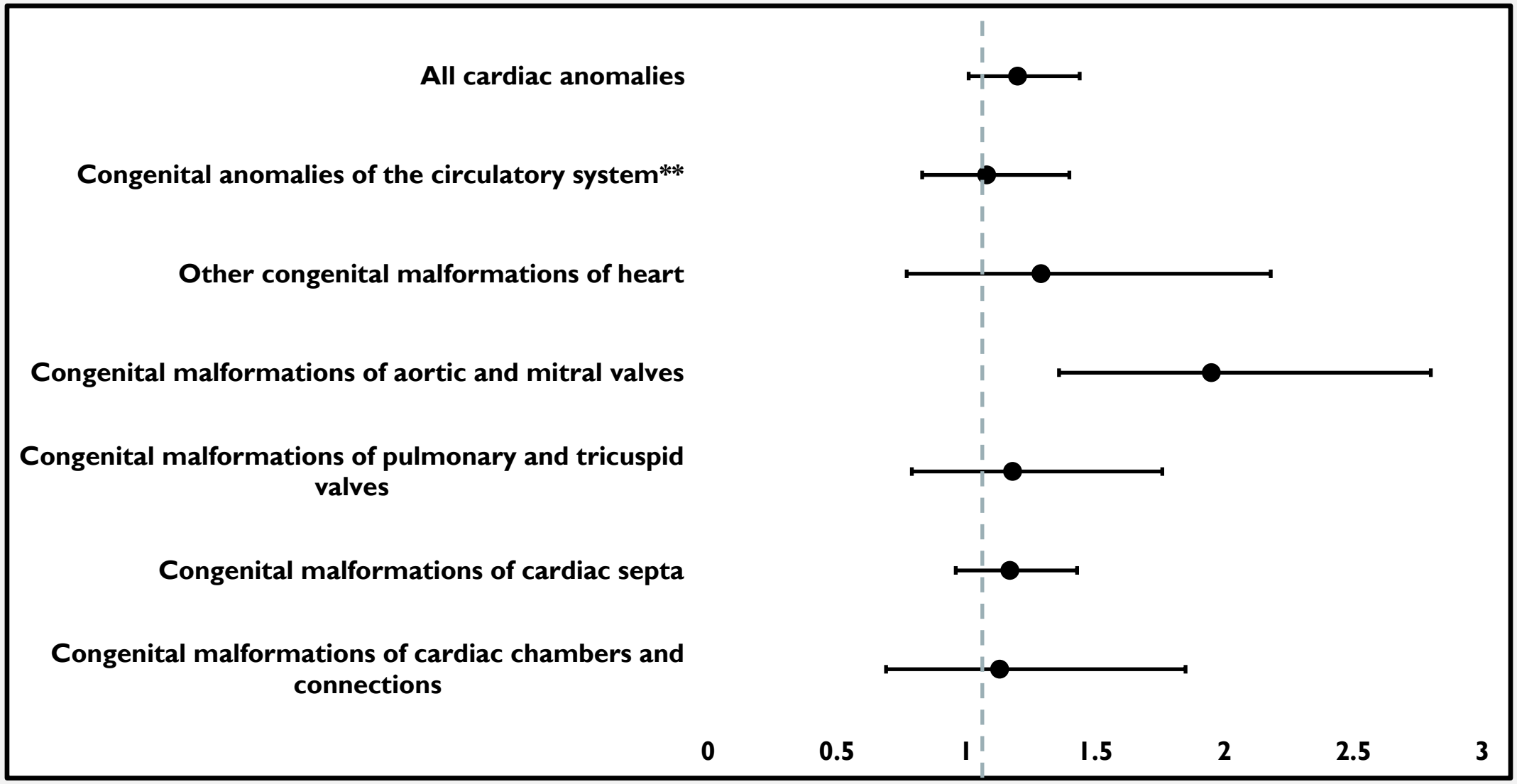


Figure 1- Cohort flow diagram.

CARDIAC AND GASTROINTESTINAL ANOMALY PREVALENCE

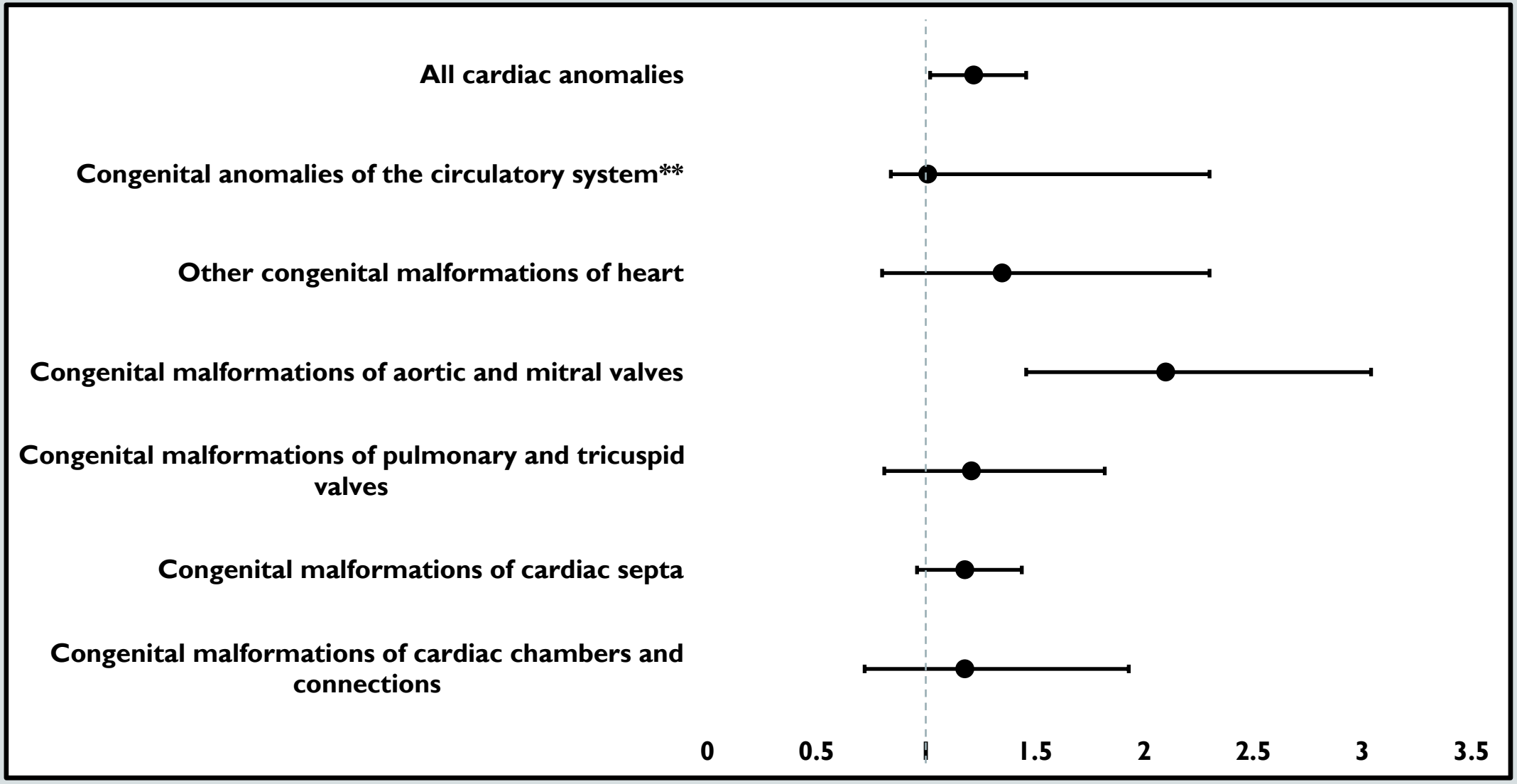


UNADJUSTED ASSOCIATIONS BETWEEN ART USE AND CARDIAC ANOMALIES



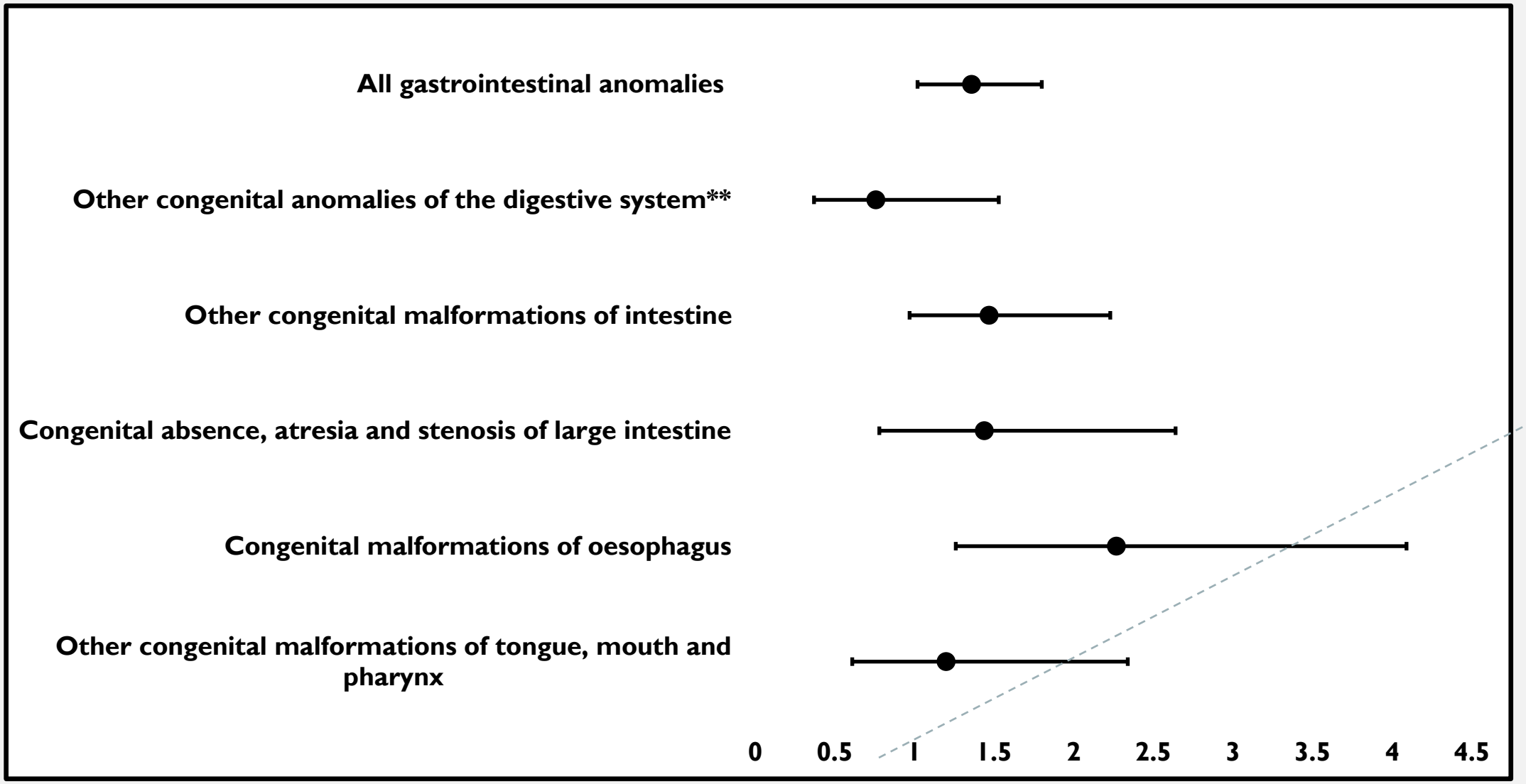
OD (95% CI)
1.20 (1.01, 1.44)
1.08 (0.83, 1.40)
1.29 (0.77, 2.18)
1.95 (1.36, 2.80)
1.18 (0.79,1.76)
1.17 (0.96, 1.43)
1.13 (0.69, 1.85)

ADJUSTED ASSOCIATIONS BETWEEN ART USE AND CARDIAC ANOMALIES



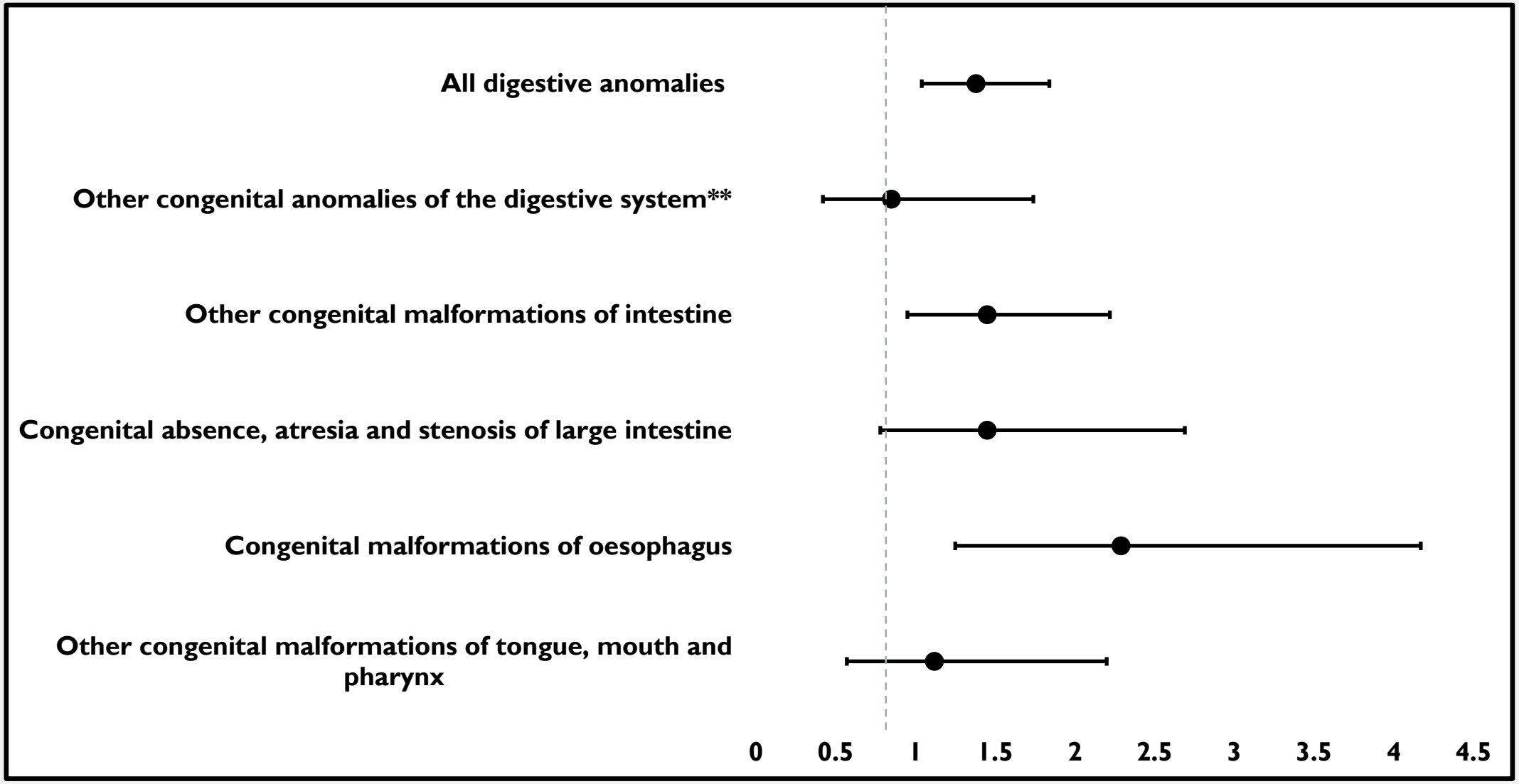
OD (95% CI)
1.22 (1.02, 1.46)
1.01 (0.84, 1.43)
1.35 (0.80, 2.30)
2.10 (1.46, 3.04)
1.21 (0.81,1.82)
1.18 (0.96, 1.44)
1.18 (0.72, 1.93)

UNADJUSTED ASSOCIATIONS BETWEEN ART USE AND GASTROINTESTINAL ANOMALIES



OD (95% CI)
1.36 (1.02, 1.80)
0.76 (0.37, 1.53)
1.47 (0.97, 2.23)
1.44 (0.78, 2.64)
2.27 (1.26, 4.09)
1.20 (0.91, 2.34)

ADJUSTED ASSOCIATIONS BETWEEN ART USE AND GASTROINTESTINAL ANOMALIES



OD (95% CI)
1.38 (1.04, 1.84)
0.85 (0.42, 1.74)
1.45 (0.95, 2.69)
1.45 (0.78, 2.69)
2.29 (1.25, 4.17)
1.12 (0.57, 2.20)

CONCLUSIONS

01

ART use and cardiac and gastrointestinal anomalies appear to be associated in Wales

02

Parental age and deprivation do not appear to influence risk

03

The associations within different types of ART such as IVF and ICSI need to be researched further in Wales

04

To allow clinicians and patients to make more informed decisions

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FOCUS SESSION

CHAIR – KINZA YOUNAS

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

LINKING ANTENATAL DETECTION AND CARIS DATA FOR QUALITY ASSURANCE

LAURA MACDERMOTT – Programme Coordinator – Antenatal Screening Wales.

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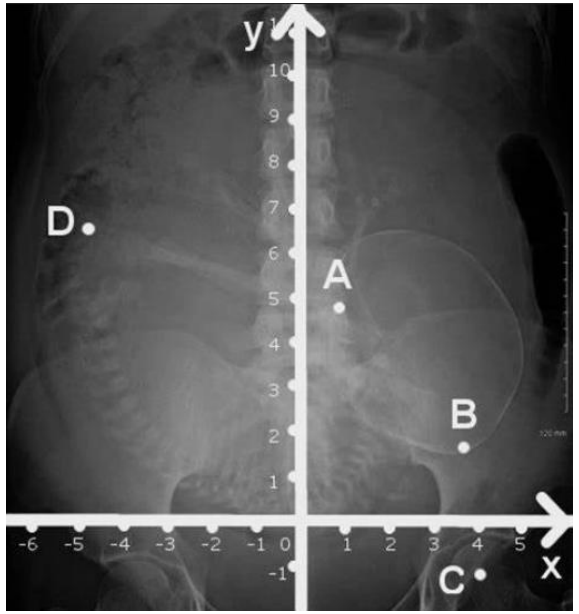
Public Health Wales

Looking back

Where were we?

1970s

- AFP
- Rubella
- X-ray



1970s

Looking back

Where were we?

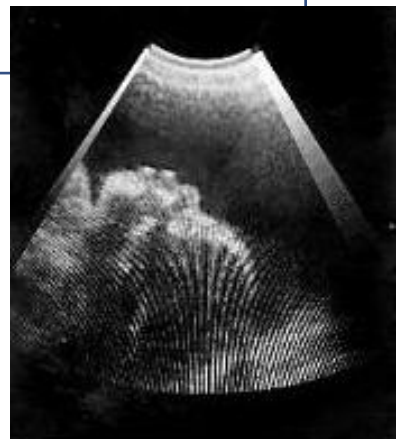
1970s

- AFP
- Rubella
- X-ray

1980s

- Blood Group and Antibodies
- Ultrasound

1980s



Looking back

Where were we?

1970s
- AFP
- Rubella
- X-ray

1980s
- Blood Group and Antibodies
- Ultrasound

1990s
- Hepatitis B and Syphilis
- Sickle Cell and Thalassaemia

Looking back
UKNSC recommendation

- UK NSC est. 1996

"Screening is the process of identifying apparently healthy people who may have an increased chance of a disease or condition.

Individuals can then be offered more information, further tests or treatment as appropriate."

National Screening Committee

UK NSC screening recommendation

Based on the last UK NSC review of this condition that occurred in March 2017.

Screening is not currently recommended for this condition.

Screening pregnant women for GBS is not recommended by the UK NSC because:

- a woman may have a positive result a few weeks before labour and a negative result when she gives birth
- GBS does not cause an infection in every baby- there is no way of telling which babies will be affected
- screening may result in giving many women antibiotics when they do not need them
- it is not known if the benefits of screening outweigh the harms for most of the population
- the proportion of babies affected by disease in countries where screening is carried out is similar to that in the UK

Supporting documents from the 2017 review

Evidence summary Group B Streptococcus (2017) compressed
This document provides the evidence on which the current UK NSC recommendation is based.

UK NSC coversheet and consultation responses GBS (2017)
This document summarises the review process including the public consultation comments.

Looking back

The start of change

- Wales Antenatal Screening Project 2002
  - Absent policies and protocols
  - Inequality
  - Lack of audit mechanisms
  - Recommended screening not implemented
  - Invalid consent
  - Ad hoc training

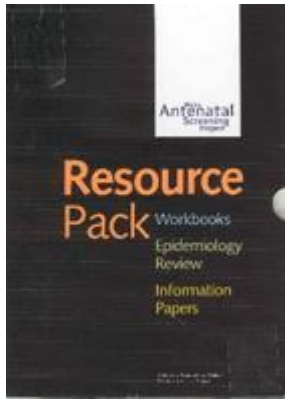
Screening Test / Trust Name	Early pregnancy scan
Bro Morgannwg Trust	
Cardiff and Vale Trust	
Carmarthenshire Trust	
Ceredigion and Mid Wales Trust	
Conwy and Denbighshire Trust	
Gwent Trust	
North East Wales Trust	
North Glamorgan Trust	
North West Wales Trust	
Pembrokeshire and Derwen Trust	
Pontypridd and Rhondda Trust	
Powys Healthcare Trust	
Swansea Trust	

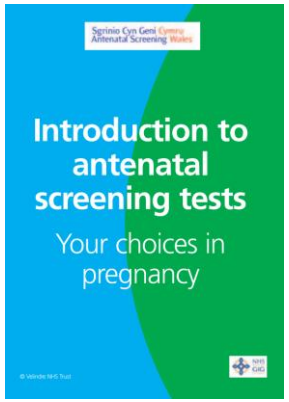
Screening Test / Trust Name	HIV
Bro Morgannwg Trust	
Cardiff and Vale Trust	
Carmarthenshire Trust	
Ceredigion and Mid Wales Trust	
Conwy and Denbighshire Trust	
Gwent Trust	
North East Wales Trust	
North Glamorgan Trust	
North West Wales Trust	
Pembrokeshire and Derwen Trust	
Pontypridd and Rhondda Trust	
Powys Healthcare Trust	
Swansea Trust	

Looking back

Working towards equality

- Agreed All Wales Policy, Standards and Protocols
- All Wales education strategy
- All Wales Information for women



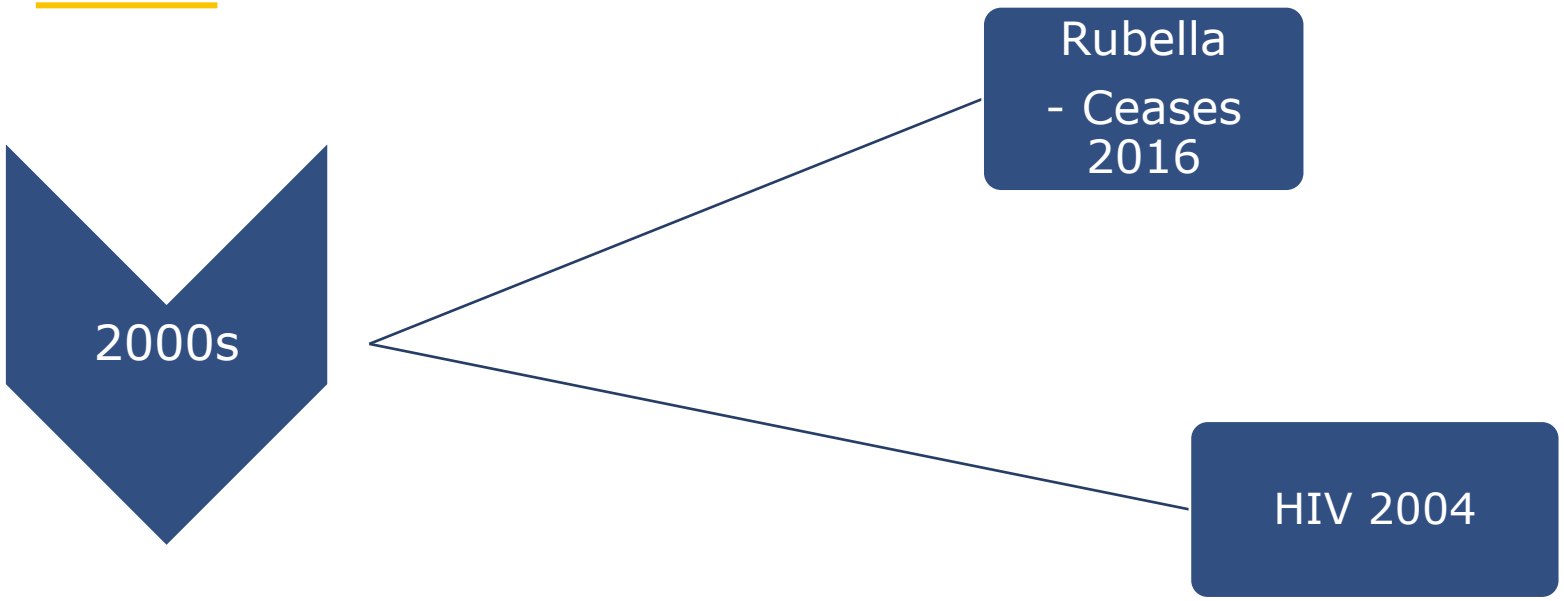


Where are we today?
Current practice

- Policy, Standards and Protocols Version 6
- The ASW Programmes are:
  - Blood group and antibody screening
  - HIV screening
  - Hepatitis B screening
  - Syphilis screening
  - Down’s syndrome, Edwards' syndrome and Patau's syndrome screening
  - Sickle cell and thalassaemia screening
  - Early pregnancy dating scan
  - Fetal anomaly ultrasound scan.

ASW Screening

Changes over the years



ASW Screening

Changes over the years

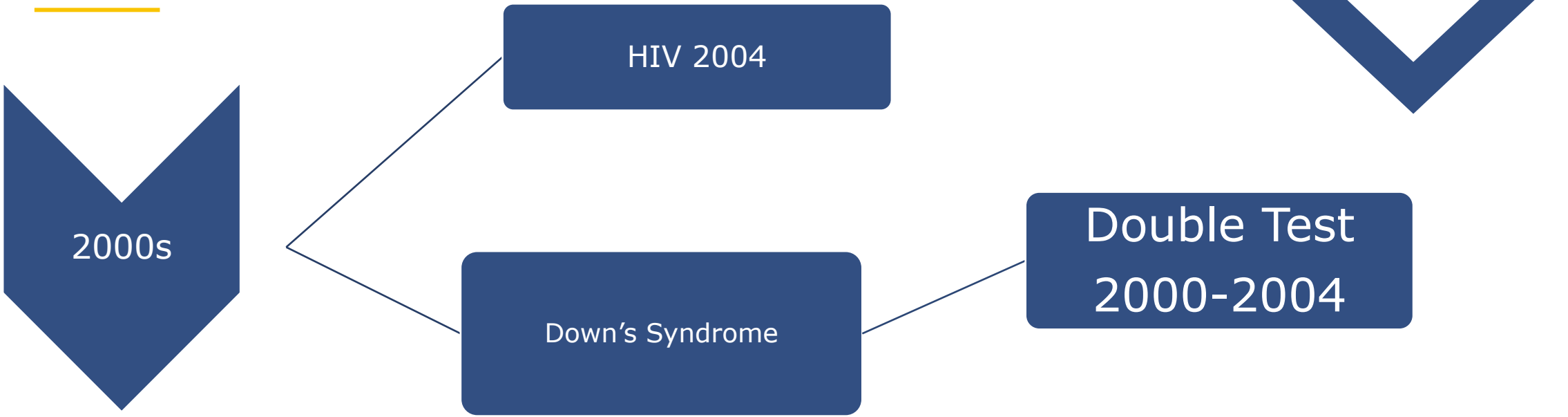
2000s

HIV 2004

Down's Syndrome

ASW Screening

Changes over the years



ASW Screening

Changes over the years

2000s

HIV 2004

Down's Syndrome

Double Test
2000-2004

Triple Test
2004-2011

ASW Screening

Changes over the years

2000s

HIV 2004

Down's Syndrome

Double Test
2000-2004

Triple Test
2004-2011

Quadruple Test
2011-2014

ASW Screening

Changes over the years

2000s

HIV 2004

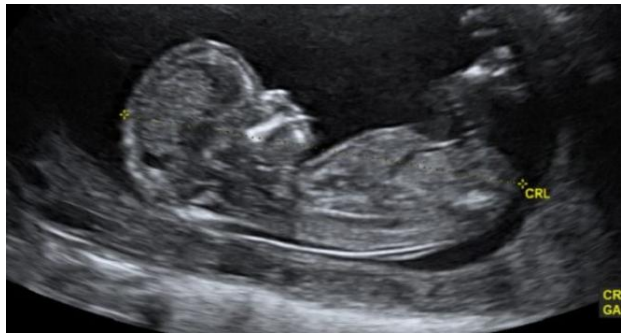
Down's Syndrome

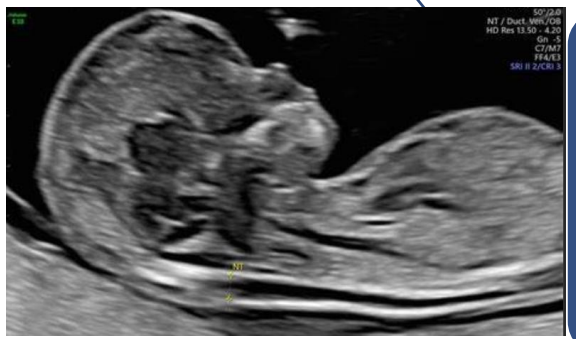
2000s

Double Test
2000-2004

Triple Test
2004-2011

Quadruple Test
2011-2014

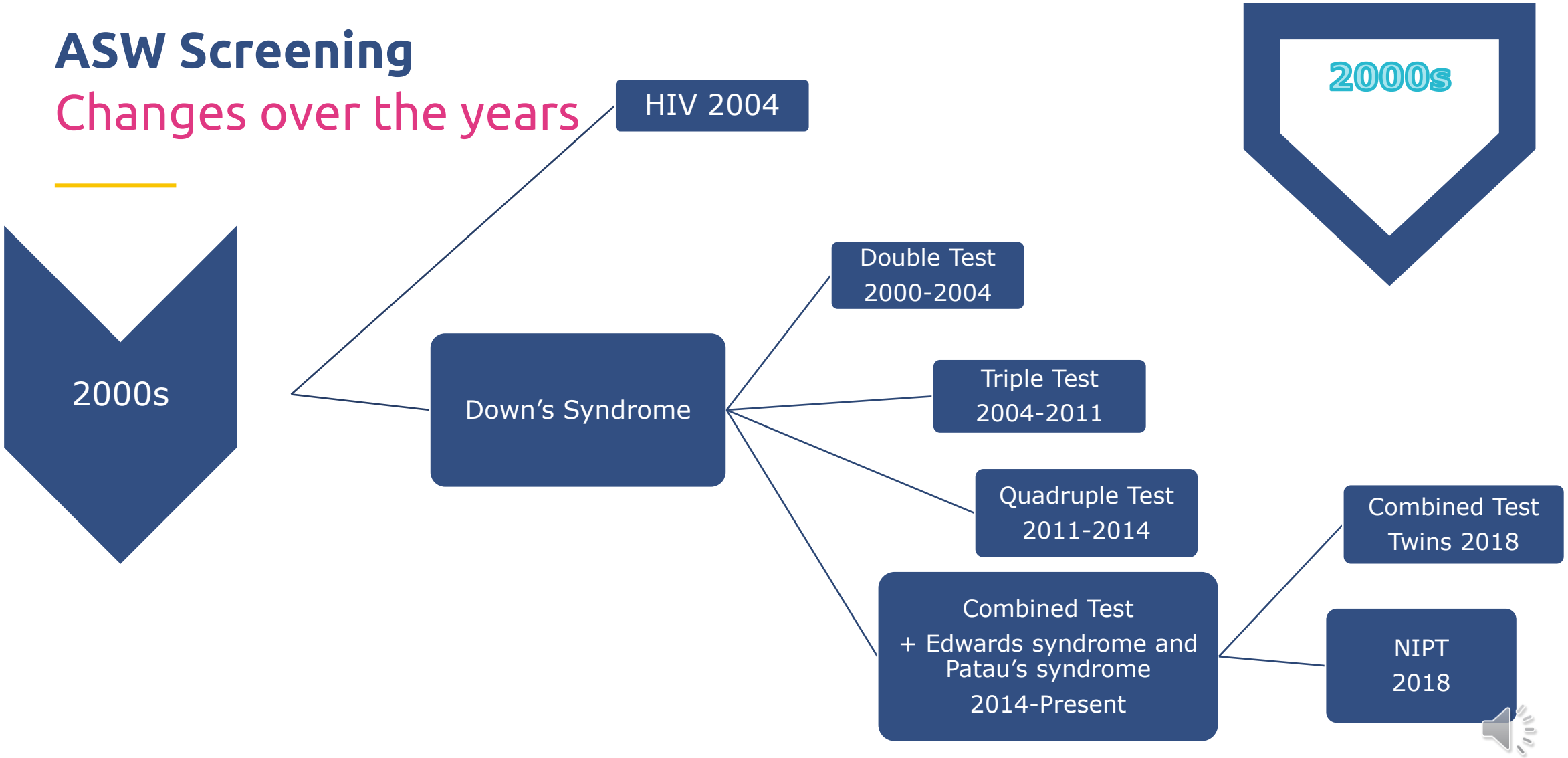




Combined Test
+ Edwards syndrome and Patau's syndrome
2014-Present

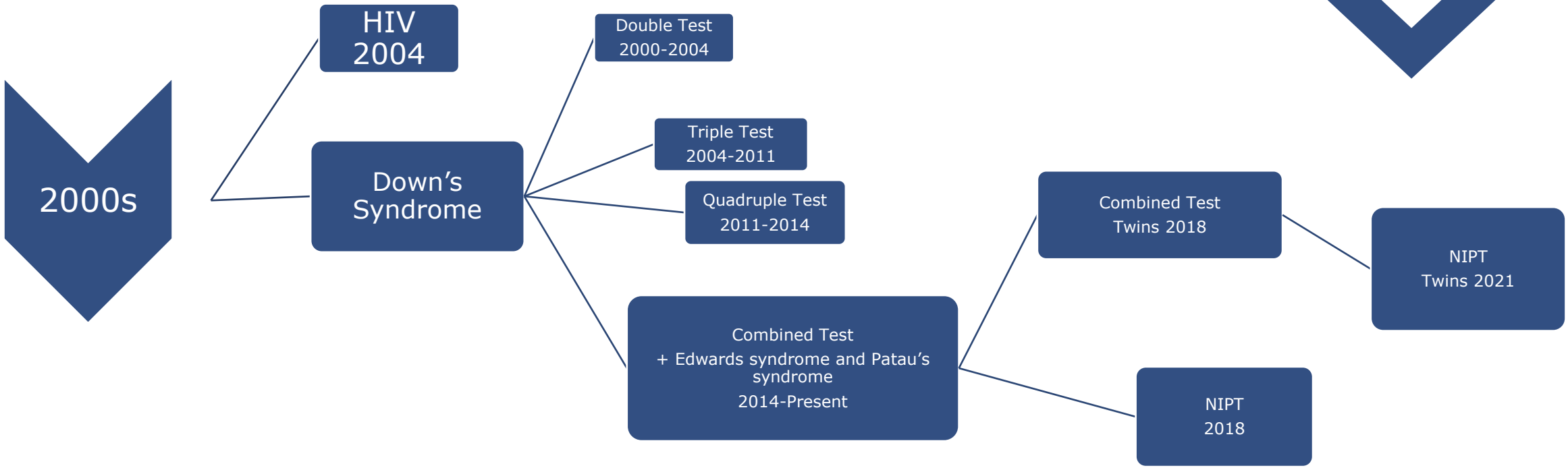
ASW Screening

Changes over the years



ASW Screening

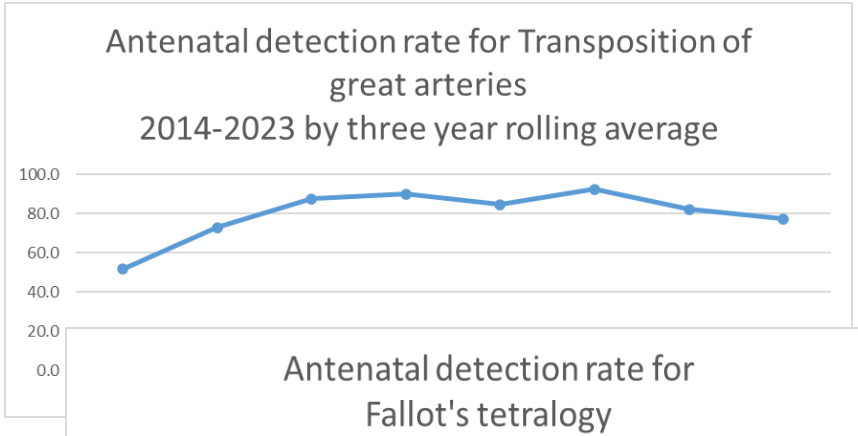
Changes over the years

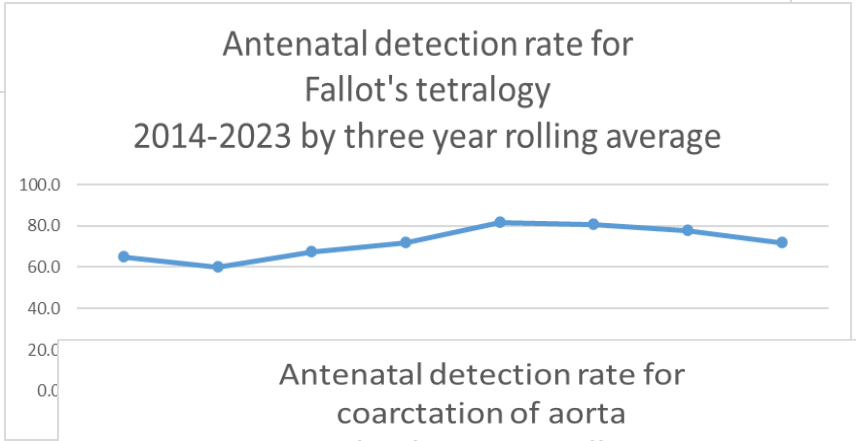


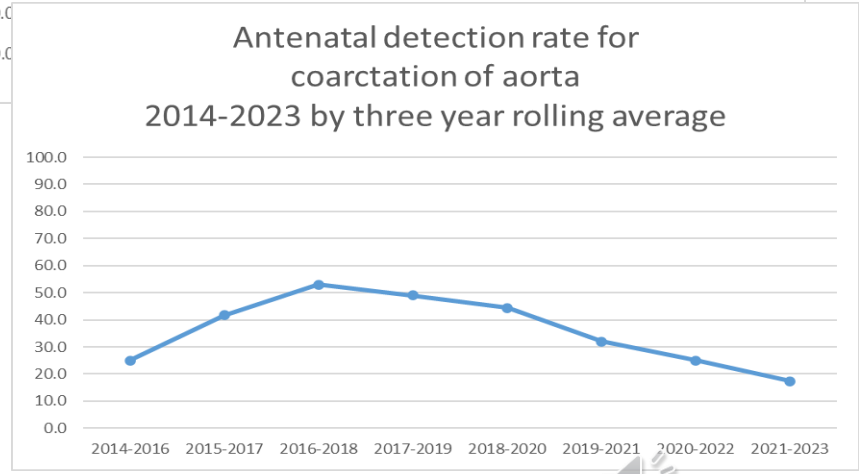
Programme Quality Assurance

Monitoring and Quality Assurance

- CARIS, DQASS, clinical experts & stakeholders
- CARIS annual report 2024 suggest a downward trend in antenatal detection
- CARIS & ASW to investigate detection rates
  - Detected at anomaly = sonographer suspicion of any type of cardiac anomaly at the anomaly scan
  - Undetected by screening = cases that would not have been detected without growth scans







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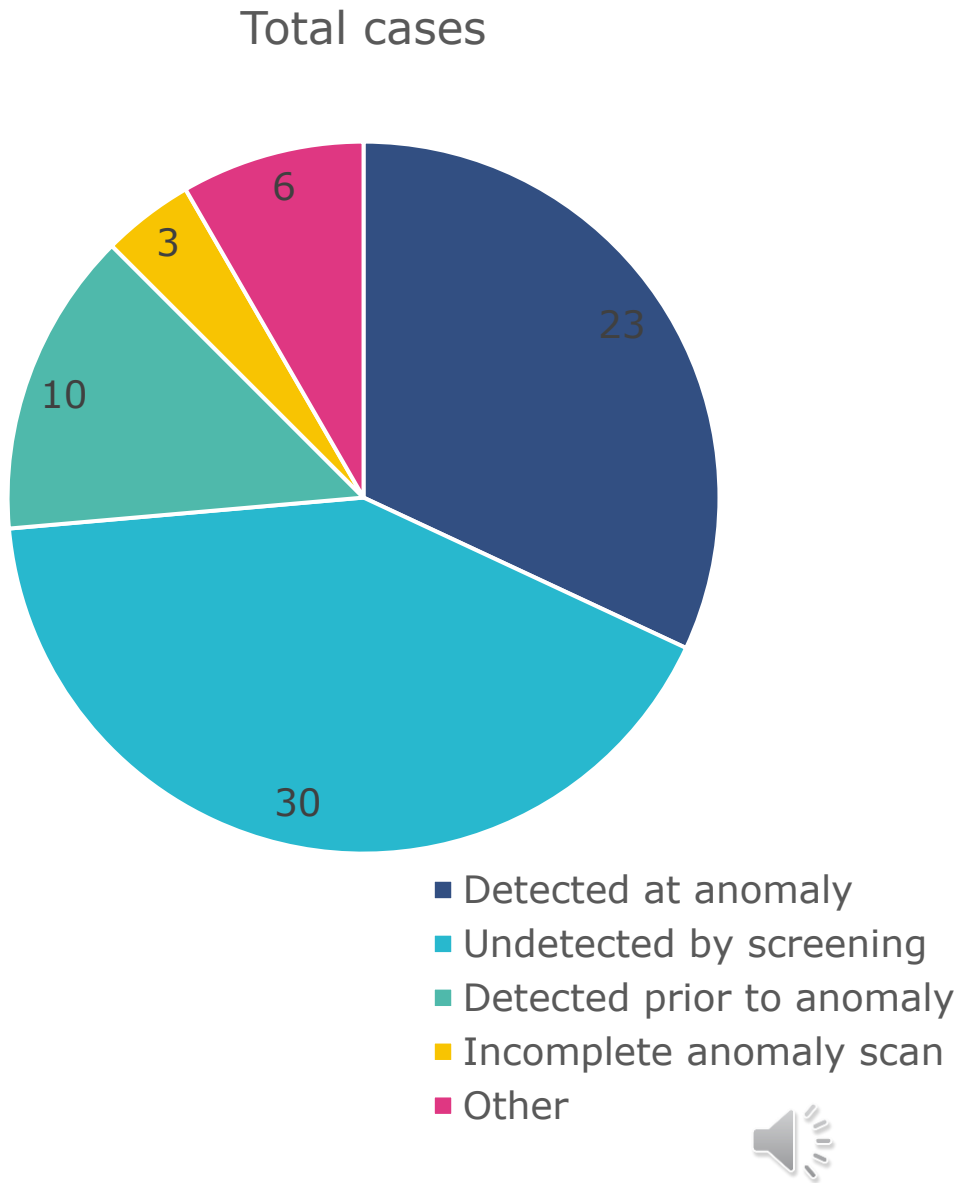
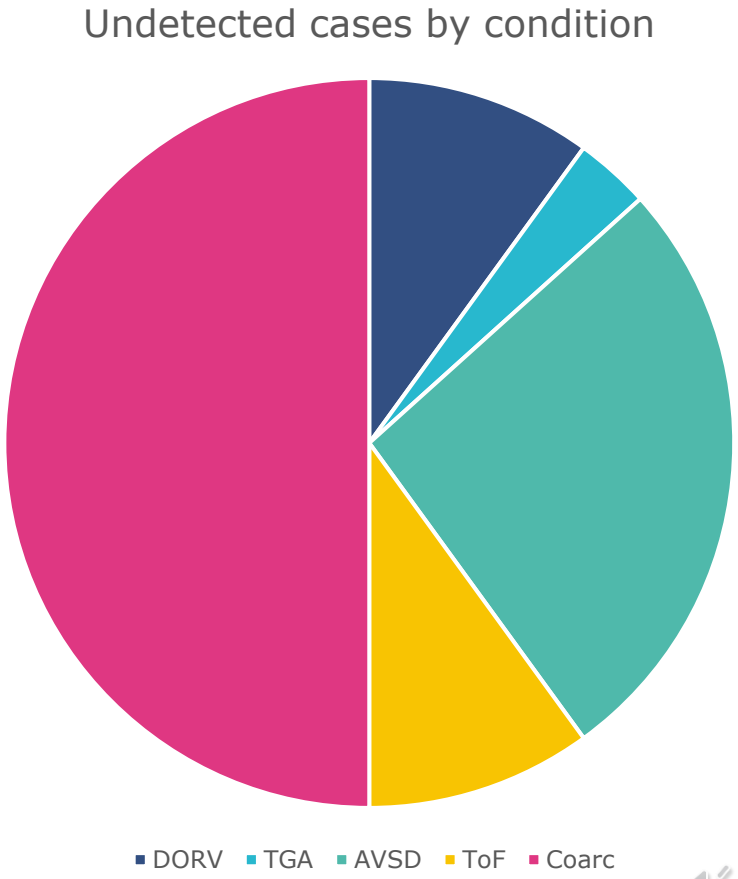
Programme Quality Assurance
CARIS Report

- 72 cases examined
  - 33 were detected by screening
    - 23 at the anomaly scan – 5 of these were reclassified as normal on tertiary scan
    - 10 prior to the anomaly scan – mostly because of findings on the dating scan
  - 3 from 'other' category detected because of non-cardiac anomalies instigating tertiary referral



Programme Quality Assurance
CARIS Report

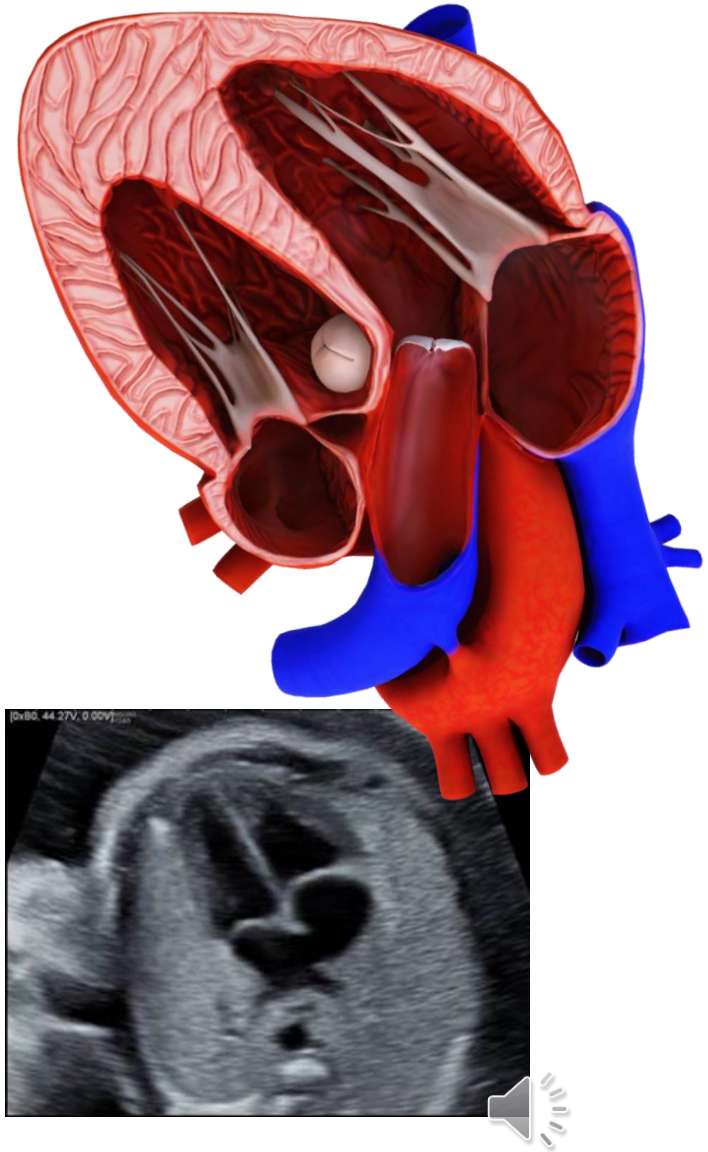
- 72 cases examined
  - 30 cases undetected by screening
    - 50% Coarctation of the aorta
  - No obvious correlation between undetected cases and maternal BMI, however data is limited
  - 6 cases not detected by screening were seen on growth scans



Programme Quality Assurance

CARIS report - outcomes

- Reassured by findings
- Of the cases undetected by screening, 50% were coarctation
- Remind professionals of pathways
  - NT ≥3.5mm at dating - cardiology referral
  - Incomplete anomaly checklist
    - Offer rescan prior to 22w+6d
    - Incomplete checklist not to be referred to FMU unless anomaly suspected
- Findings discussed with WFCVN representatives for consideration at future training events



ASW & CARIS Final Thoughts

Strengthening quality through collaborative leadership

- Midwifery
  - Governance lead
  - Screening coordinator
- Ultrasound
  - Obstetric lead
  - NT lead
  - Cardiac lead
- Obstetricians
- DQASS
- CARIS

ANTENATAL DETECTION AND IN-UTERO TREATMENT

PROF. ASMA KHALIL

MD, MBBCh, FRCOG, MSc.(Epi), DFSRH, Dip.(GUM)

Prof. of Fetal Medicine at St. George's University of London

Director of Fetal Medicine Unit

Liverpool Women's Hospital.

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Public Health Wales

NHS

St George's University Hospitals

NHS Foundation Trust

Liverpool Women's

NHS Foundation Trust

Please see the recorded version of this presentation Thank You

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Public Health Wales

THE FETAL MEDICINE PATHWAY

DR. ARMIN VANDEPERRE – Consultant in Obstetrics and Fetal Medicine, UHW.

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

Fetal Medicine Department C+V:

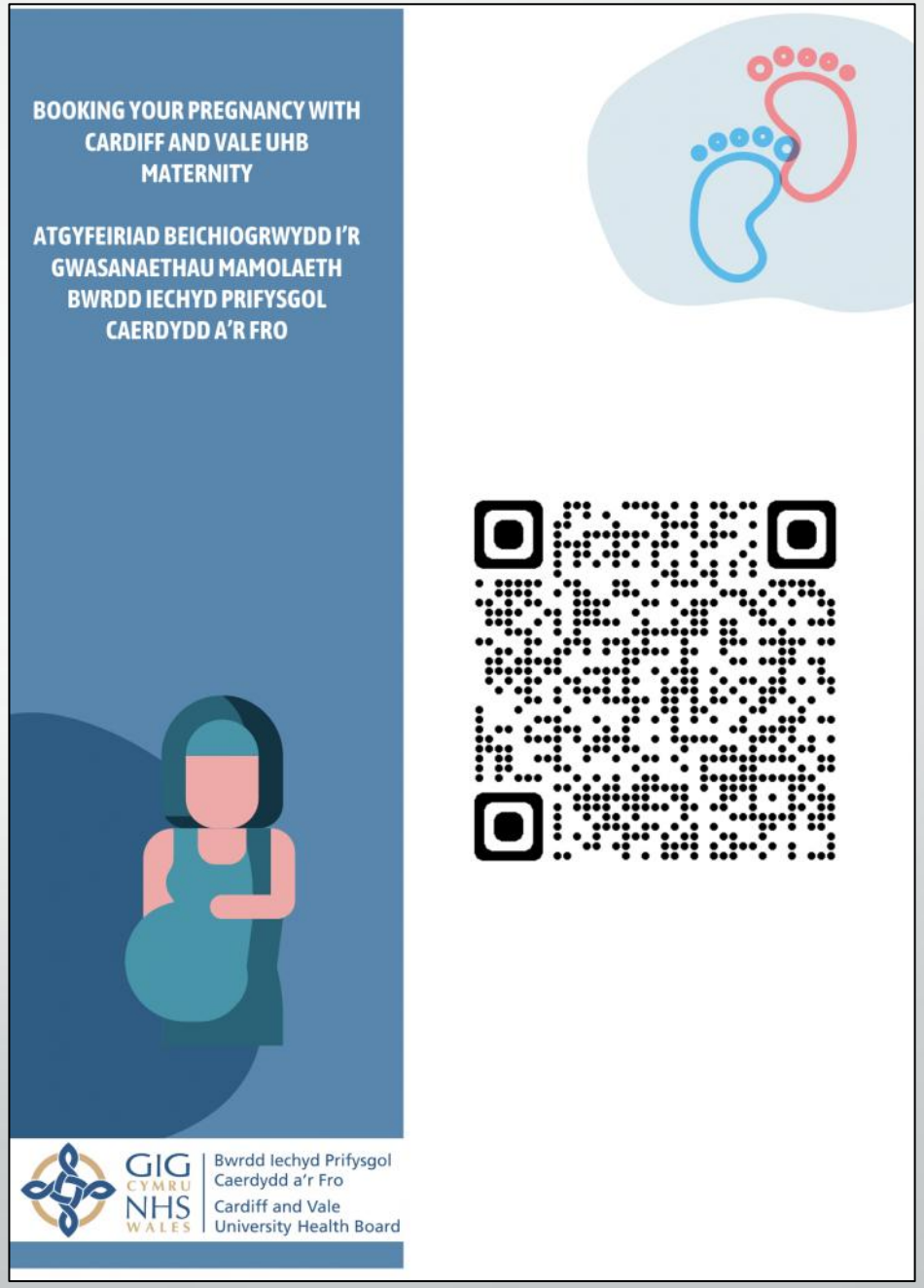
Who, What, Where?

- **Consultants:** Dr Bryan Beattie, Dr Mark Denbow, Dr Armin Vandeperre
- **Midwives:** Judith, Jacqui, Jayne, Denise, Carole-Ann
- UHW, Antenatal clinic, Ground floor Women's Unit
  - Women's unit entrance turn right in front of the lifts

Pathway

- Start
- Referral
- FMU appointment
- First time in FMU
- Ultrasound
- Counselling
- Options

How does it start?



Screening ultrasounds

- **Performed in the local unit:**
  - **Dating 11-14w:**
    - Increased NT
    - cystic hygroma
    - Gastroschisis
    - Acrania
  - **Anomaly 19-22w:**
    - Cardiac anomalies
    - Open spina bifida
    - Talipes
    - Echogenic bowel
    - Renal anomalies
  - **Growth 24-42w:**
    - Hydrops
    - Dilated bowel
    - Ventriculomegaly
    - Polyhydramnios
    - Arrhythmias
    - SGA

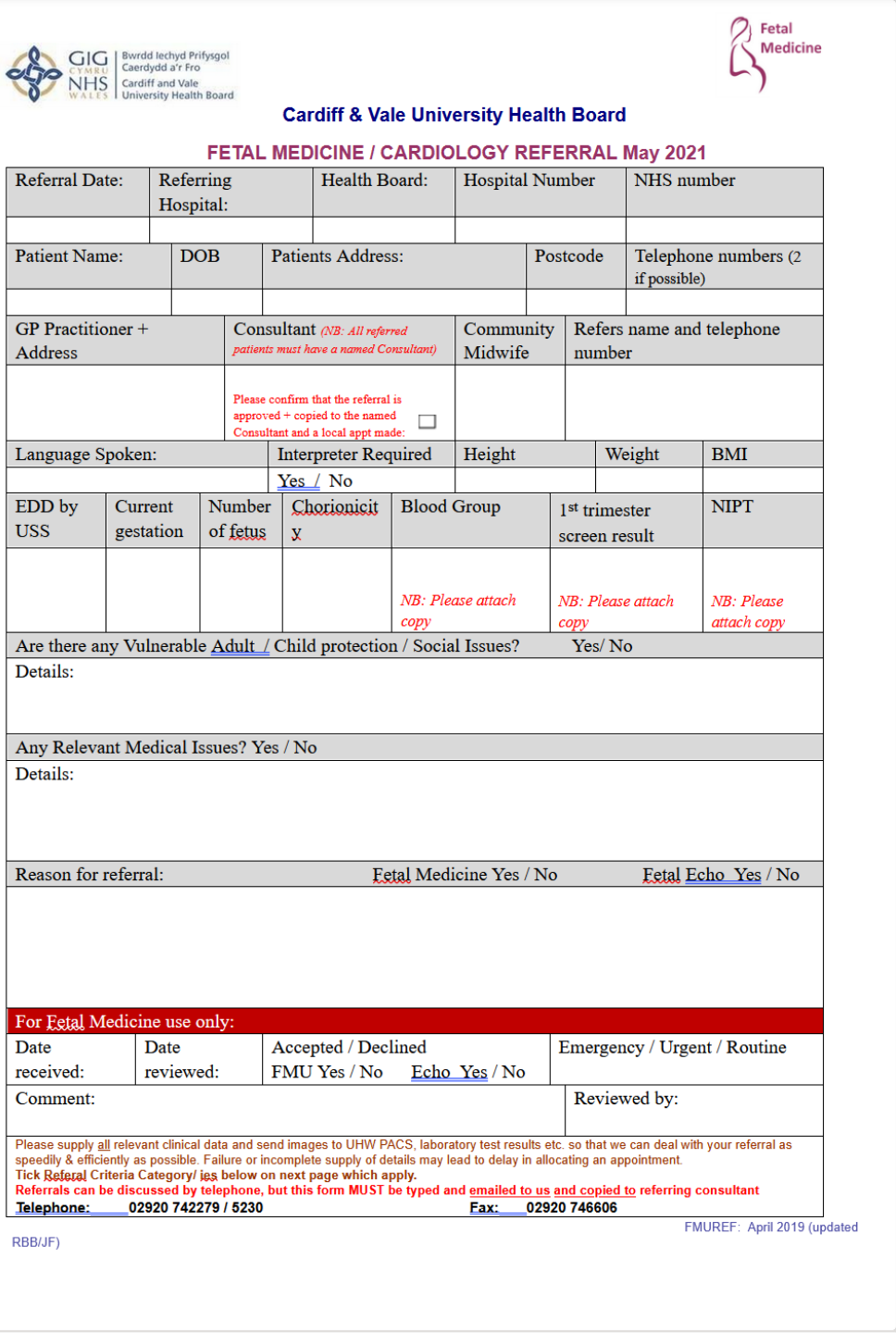
Sgrinio Cyn Geni Cymru
Antenatal Screening Wales

Other reasons for referral

- **Previous anomalies:**
  - Open spina bifida
  - Previous hydrops
- **Infections:**
  - Varicella
  - Syphilis
  - CMV
- **Antibodies:**
  - D
  - Kell
  - C
  - NAIT
- **Genetics:**
  - Balanced translocations
  - Sickle cell
  - Monogenetic syndromes
- **Test:**
  - Combined screening
  - NIPT
- **Procedures:**
  - Selective fetal reduction
  - Amniocentesis
  - CVS

Referral to fetal medicine

- BOOKED
- No written, ONLY TYPED
- Demographics
- Referring consultant
- Bloud group
- First trimester screening result
- Social issues
- Interpreter needed?
- Reason for referral
- Fetal medicine and/or fetal cardiology



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Fetal Medicine

Cardiff & Vale University Health Board

FETAL MEDICINE / CARDIOLOGY REFERRAL May 2021

Referral Date:	Referring Hospital:	Health Board:	Hospital Number	NHS number

Patient Name:	DOB	Patients Address:	Postcode	Telephone numbers (2 if possible)

GP Practitioner + Address	Consultant *(NB: All referred patients must have a named Consultant)*	Community Midwife	Refers name and telephone number
	Please confirm that the referral is approved + copied to the named Consultant and a local appt made: ☐		

Language Spoken:	Interpreter Required	Height	Weight	BMI
	Yes / No			

EDD by USS	Current gestation	Number of fetus	Chorionicity	Blood Group	1st trimester screen result	NIPT
				NB: Please attach copy	*NB: Please attach copy*	*NB: Please attach copy*

Are there any Vulnerable Adult / Child protection / Social Issues? Yes/ No

Details:

Any Relevant Medical Issues? Yes / No

Details:

Reason for referral: Fetal Medicine Yes / No Fetal Echo Yes / No

For Fetal Medicine use only:

Date received:	Date reviewed:	Accepted / Declined FMU Yes / No Echo Yes / No	Emergency / Urgent / Routine

Comment:	Reviewed by:

Please supply all relevant clinical data and send images to UHW PACS, laboratory test results etc. so that we can deal with your referral as speedily & efficiently as possible. Failure or incomplete supply of details may lead to delay in allocating an appointment.
Tick Referal Criteria Category/ ies below on next page which apply.
Referrals can be discussed by telephone, but this form MUST be typed and emailed to us and copied to referring consultant
Telephone: 02920 742279 / 5230 **Fax: 02920 746606**

FMUREF: April 2019 (updated RBB/JF)

Referral form - Page 1

Bwrdd Iechyd Prifysgol Caerdydd a'r Fro
Cardiff and Vale University Health Board

Fetal Medicine

Cardiff & Vale University Health Board

FETAL MEDICINE / CARDIOLOGY REFERRAL May 2021

Referral Date:	Referring Hospital:	Health Board:	Hospital Number	NHS number

Patient Name:	DOB	Patients Address:	Postcode	Telephone numbers (2 if possible)

GP Practitioner + Address	Consultant *(NB: All referred patients must have a named Consultant)*	Community Midwife	Refers name and telephone number
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Language Spoken:	Interpreter Required	Height	Weight	BMI
	Yes / No			

EDD by USS	Current gestation	Number of fetus	Chorionicity	Blood Group	1st trimester screen result	NIPT
				NB: Please attach copy	*NB: Please attach copy*	*NB: Please attach copy*

Referral form - page 1 continued

Are there any Vulnerable Adult / Child protection / Social Issues? Yes/ No
Details:
Any Relevant Medical Issues? Yes / No
Details:
Reason for referral: Fetal Medicine Yes / No Fetal Echo Yes / No

For Fetal Medicine use only:

Date received:	Date reviewed:	Accepted / Declined FMU Yes / No Echo Yes / No	Emergency / Urgent / Routine
Comment:			Reviewed by:

Please supply all relevant clinical data and send images to UHW PACS, laboratory test results etc. so that we can deal with your referral as speedily & efficiently as possible. Failure or incomplete supply of details may lead to delay in allocating an appointment.

Tick Referal Criteria Category/ ies below on next page which apply.

Referrals can be discussed by telephone, but this form MUST be typed and emailed to us and copied to referring consultant

Telephone: 02920 742279 / 5230 **Fax: 02920 746606**

Email: fetal.med@ wales.nhs.uk (Secure NHS Email)

Address: Fetal Medicine Unit, Maternity Unit, University Hospital of Wales, Heath Park, Cardiff, CF14 4XW

Referral form - Page 2

FETAL MEDICINE / CARDIOLOGY REFERRAL CRITERIA Nov 2020

Fetal Medicine Referral Criteria Nov 2020 **Please tick All that apply**

Patients in these groups are eligible for a Fetal Medicine Referral

1) Specialised ultrasound examination and subsequent care of fetuses at risk of or with suspected malformations, dysmorphic or genetic syndromes	
2) Relevant family history of chromosomal or genetic disorders	
3) Relevant chromosomal or genetic disorder - *will be assessed on individual basis*	
4) Previous relevant structural anomaly - *will be assessed on individual basis.*	
5) Ultrasound guided Invasive testing – i.e Chorionic villus sampling, amniocentesis, fetal blood sampling *(excludes amniocentesis for maternal age and increased Downs Risk on Combined Screening)*	
6) Ultrasound guided therapies – i.e Amniotic fluid, drainage, transfusion therapy, feto-amniotic shunting,	
7) Procedures for the selective reduction of high multiple pregnancies *(Triplets or greater)*	
8) Complicated multiple pregnancies - *TTTS and growth discrepancy*	
9) Feticide in pregnancies more than 21+6 weeks	
10) Pregnancies at risk of Iso-immunisation and allo-immune thrombocytopaenia *(NAIT)*	
11) Fetal Infection – *Toxoplasmosis, CMV, Parvovirus, Varicella, Rubella and Syphilis*	
12) Cardiac arrhythmias – *Fetal SVT, Heart block (also see Fetal Echo Criteria below)*	
13) Exposure to Teratogens	
14) IUGR – *severe early onset in current pregnancy (growth below 3rd centile, prior to 32/40 and AREDV (see RCOG: "The management of the Small for Dates Fetus GTG 31")*	

Exclusion: *Preterm Premature Rupture of the Membranes (PPROM) is an obstetric complication to be managed locally*

Referral form - page 2 cont.: Fetal cardiology

Fetal Echo Referral Welsh Criteria Nov 2020:

Patients in these groups are eligible for a Fetal Echo

Please tick All that apply

1) Suspicion of fetal cardiac abnormality during an obstetric scan i. Most cases of fetal congenital heart disease will occur in this group ii. Pericardial effusion > 3mm.	
2) Fetal arrhythmias i. Sustained bradycardia - *heart rate <110 beats per minute* ii. Tachycardia – *heart rate >180 beats per minute*	
4) Paternal congenital heart disease *(risk 2-6%)*	
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8) Chromosomal anomalies, gene disorders or syndromes associated with congenital heart disease or cardiomyopathy *(risk will depend on individual disorder)*	
9) Nuchal translucency under 14 weeks gestation measuring =/>3.5mm	

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2) Maternal collagen disease with anti Ro/SSA and/or anti La/SSB *(risk 2-3%)*	
3) Maternal medication with Non Steroidal Anti-Inflammatory (NSAID) drugs	

Fetal Medicine Department C+V:

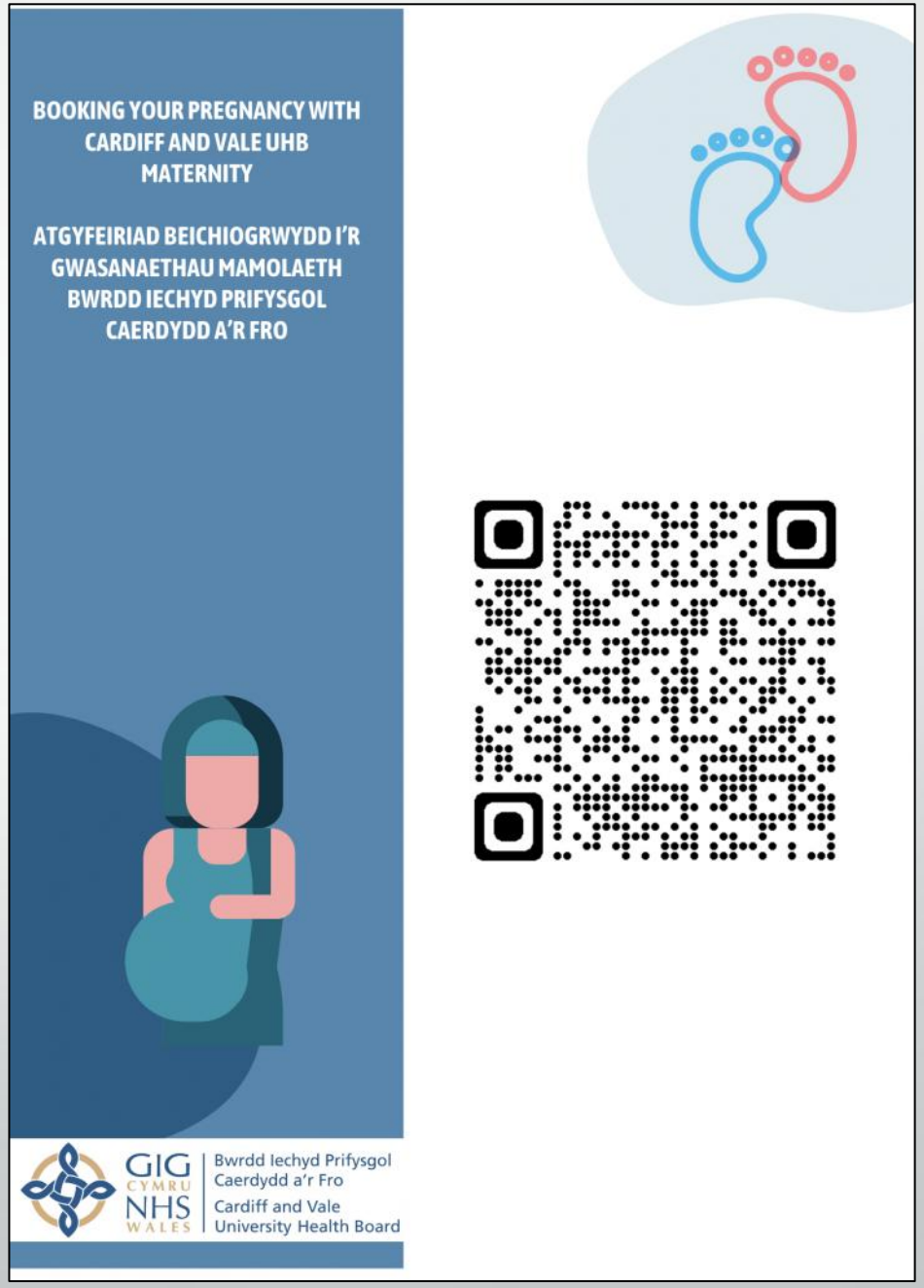
Who, What, Where?

- **Consultants:** Dr Bryan Beattie, Dr Mark Denbow, Dr Armin Vandeperre
- **Midwives:** Judith, Jacqui, Jayne, Denise, Carole-Ann
- UHW, Antenatal clinic, Ground floor Women's Unit
  - Women's unit entrance turn right in front of the lifts

Pathway

- Start
- Referral
- FMU appointment
- First time in FMU
- Ultrasound
- Counselling
- Options

How does it start?



Screening ultrasounds

- **Performed in the local unit:**
  - **Dating 11-14w:**
    - Increased NT
    - cystic hygroma
    - Gastroschisis
    - Acrania
  - **Anomaly 19-22w:**
    - Cardiac anomalies
    - Open spina bifida
    - Talipes
    - Echogenic bowel
    - Renal anomalies
  - **Growth 24-42w:**
    - Hydrops
    - Dilated bowel
    - Ventriculomegaly
    - Polyhydramnios
    - Arrhythmias
    - SGA

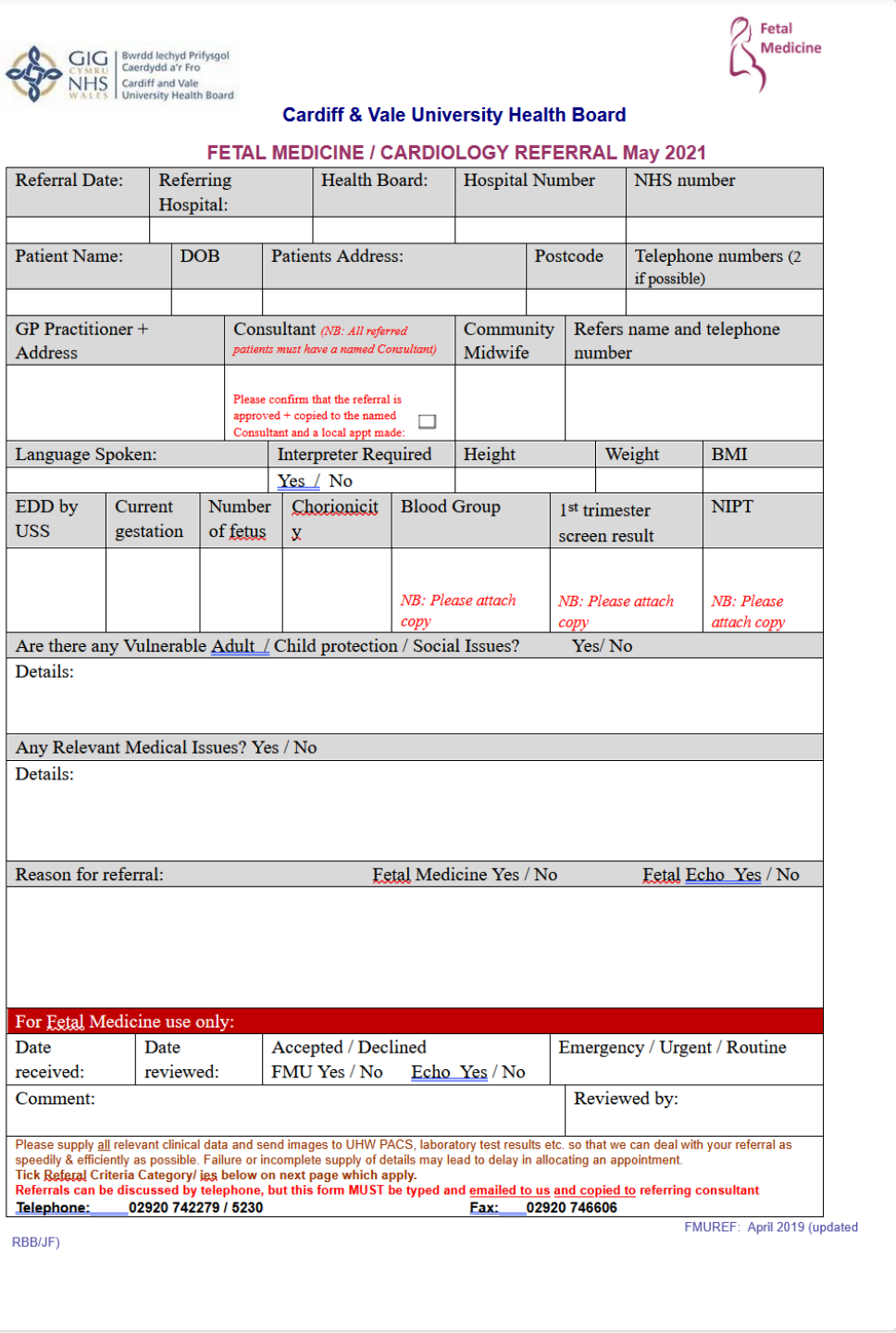
Sgrinio Cyn Geni Cymru
Antenatal Screening Wales

Other reasons for referral

- **Previous anomalies:**
  - Open spina bifida
  - Previous hydrops
- **Infections:**
  - Varicella
  - Syphilis
  - CMV
- **Antibodies:**
  - D
  - Kell
  - C
  - NAIT
- **Genetics:**
  - Balanced translocations
  - Sickle cell
  - Monogenetic syndromes
- **Test:**
  - Combined screening
  - NIPT
- **Procedures:**
  - Selective fetal reduction
  - Amniocentesis
  - CVS

Referral to fetal medicine

- BOOKED
- No written, ONLY TYPED
- Demographics
- Referring consultant
- Bloud group
- First trimester screening result
- Social issues
- Interpreter needed?
- Reason for referral
- Fetal medicine and/or fetal cardiology



GIG CYMRU NHS WALES — Bwrdd Iechyd Prifysgol Caerdydd a'r Fro / Cardiff and Vale University Health Board

Fetal Medicine

Cardiff & Vale University Health Board

FETAL MEDICINE / CARDIOLOGY REFERRAL May 2021

Referral Date:	Referring Hospital:	Health Board:	Hospital Number	NHS number

Patient Name:	DOB	Patients Address:	Postcode	Telephone numbers (2 if possible)

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Details:

Any Relevant Medical Issues? Yes / No

Details:

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For Fetal Medicine use only:

Date received:	Date reviewed:	Accepted / Declined FMU Yes / No Echo Yes / No	Emergency / Urgent / Routine

Comment:	Reviewed by:

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FMUREF: April 2019 (updated RBB/JF)

Referral form - Page 1

Bwrdd Iechyd Prifysgol Caerdydd a'r Fro
Cardiff and Vale University Health Board

Fetal Medicine

Cardiff & Vale University Health Board

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3) Maternal medication with Non Steroidal Anti-Inflammatory (NSAID) drugs	

Appointment timing in FMU

- Emergency: 24h (only after consultant-to-consultant referral)
- Urgent: within 5 working days
- Routine: when appropriate

First time in FMU

- **Personal History**
  - Medical
  - Obstetric
    - EDD
    - Previous screening
  - Drugs
- **Results**
  - FTS/NIPT
  - Serology
- **Family History**
  - Congenital anomalies
  - Genetic conditions
  - Consanguinity

- **Fetal Medicine Sieve**
  - Normal
  - Aneuploidy
  - Syndromes
  - Isolated structural
  - Infection
  - Haematological tests
  - Neoplasia
  - Toxic
  - Maternal/placental

FMU ultrasound

- Detailed anomaly or dating scan
- Situational extra images:
  - Facial profile
  - Eyes
  - Neck
  - Long bones
- Situational measurements:
  - Long bones
  - Chest circumference
  - Uterine arteries
  - Ductus Venosus
- 3/4D:
  - Face: nose, lip, …
  - Skeleton

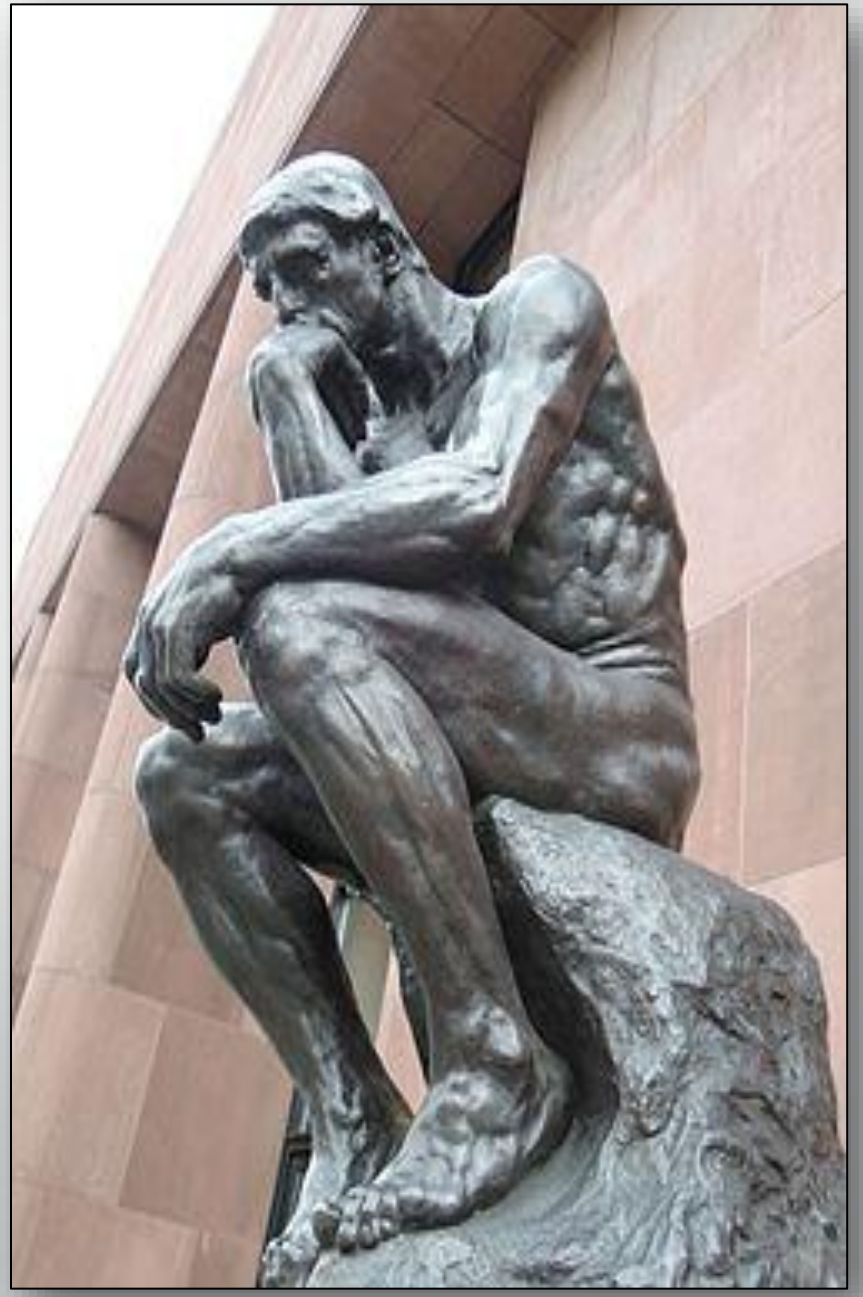
Counselling about the anomaly

- What is the problem?
- How mild or severe?
- Isolated or multiple anomalies?
- Risk of underlying reason:
  - Normal variant
  - Placenta
  - Infectious
  - syndromes or genetic conditions
- Prognosis in pregnancy and after birth?
- Morbidity?

- **Fetal Medicine Sieve**
  - Normal
  - Aneuploidy
  - Syndromes
  - Isolated structural
  - Infection
  - Haematological
  - Neoplasia
  - Toxic
  - Maternal/placental

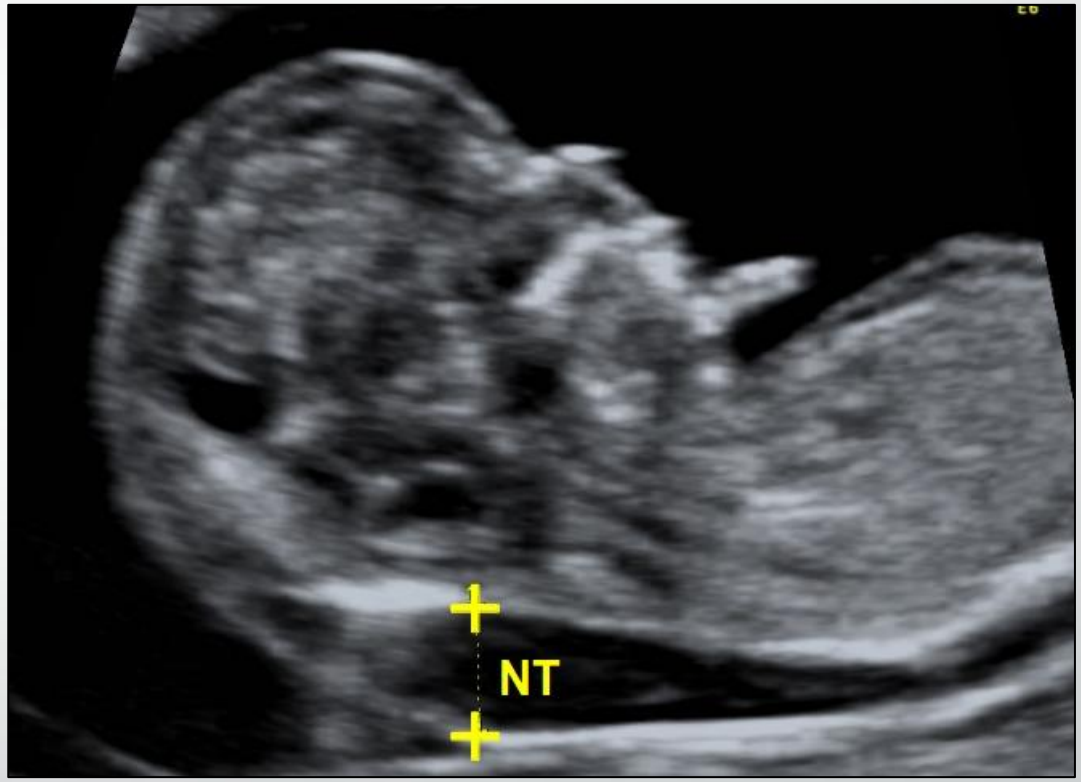
Options

- Conservative (Do nothing)
- Investigate
  - Invasive testing:
    - CVS
    - Amniocentesis
    - Thoracocentesis
  - NIPT (if high chance FTS)
  - Fetal Cardiac echo
  - Fetal brain MRI
- Monitor
  - Serial ultrasound
  - Growth
  - Dopplers
  - Structures
- Deliver/TOP
  - <24w
  - >24w
- In utero therapy
  - Open spina bifida
  - IUT
  - CDH
  - Laser



Example: Increased NT

- Referred after dating scan:
  - Include blood group
  - First trimester screening result
- Should be seen within 5 working days
  - Sometimes delayed if patient wants to wait for FTS or NIPT result



First

- **Personal History**
  - Medical
  - Obstetric
    - EDD
    - Previous screening
  - Drugs
- **Results**
  - FTS/NIPT
- **Family History**
  - Congenital anomalies
  - Genetic conditions
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- **Fetal Medicine Sieve**
  - Normal
  - Aneuploidy
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FMU ultrasound

- **Detailed dating scan:**
  - Midline in the brain
  - Normal shape of the skull
  - Car crash sign?
  - 2 arms/2 legs
  - Abdomen closed?
  - NT?
  - Heart
  - 2 kidneys
  - Bladder and stomach filled
  - Hydrops?

- **Fetal Medicine Sieve**
  - Normal
  - Aneuploidy
  - Syndromes
  - Isolated structural
  - Infection
  - Haematological
  - Neoplasia
  - Toxic
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Increased NT counselling

Nuchal translucency	Chromosomal defects	Fetal death	Major fetal abnormalities	Alive and well
<95th centile	0.2%	1.3%	1.6%	97%
95th-99th centiles	3.7%	1.3%	2.5%	93%
3.5-4.4 mm	21.1%	2.7%	10.0%	70%
4.5-5.4 mm	33.3%	3.4%	18.5%	50%
5.5-6.4 mm	50.5%	10.1%	24.2%	30%
>6.5 mm	64.5%	19.0%	46.2%	15%

Souka et al., 2005

Options Monitor + Investigate

- Now: CVS
- 16w: amniocentesis

- **20 week:**
  - Detailed anomaly scan
  - Fetal Cardiac echo

- **Serial ultrasound:**
  - 28, 32, 36, 39
  - More if further concerns
  - Local vs FMU

Options in detail: invasive

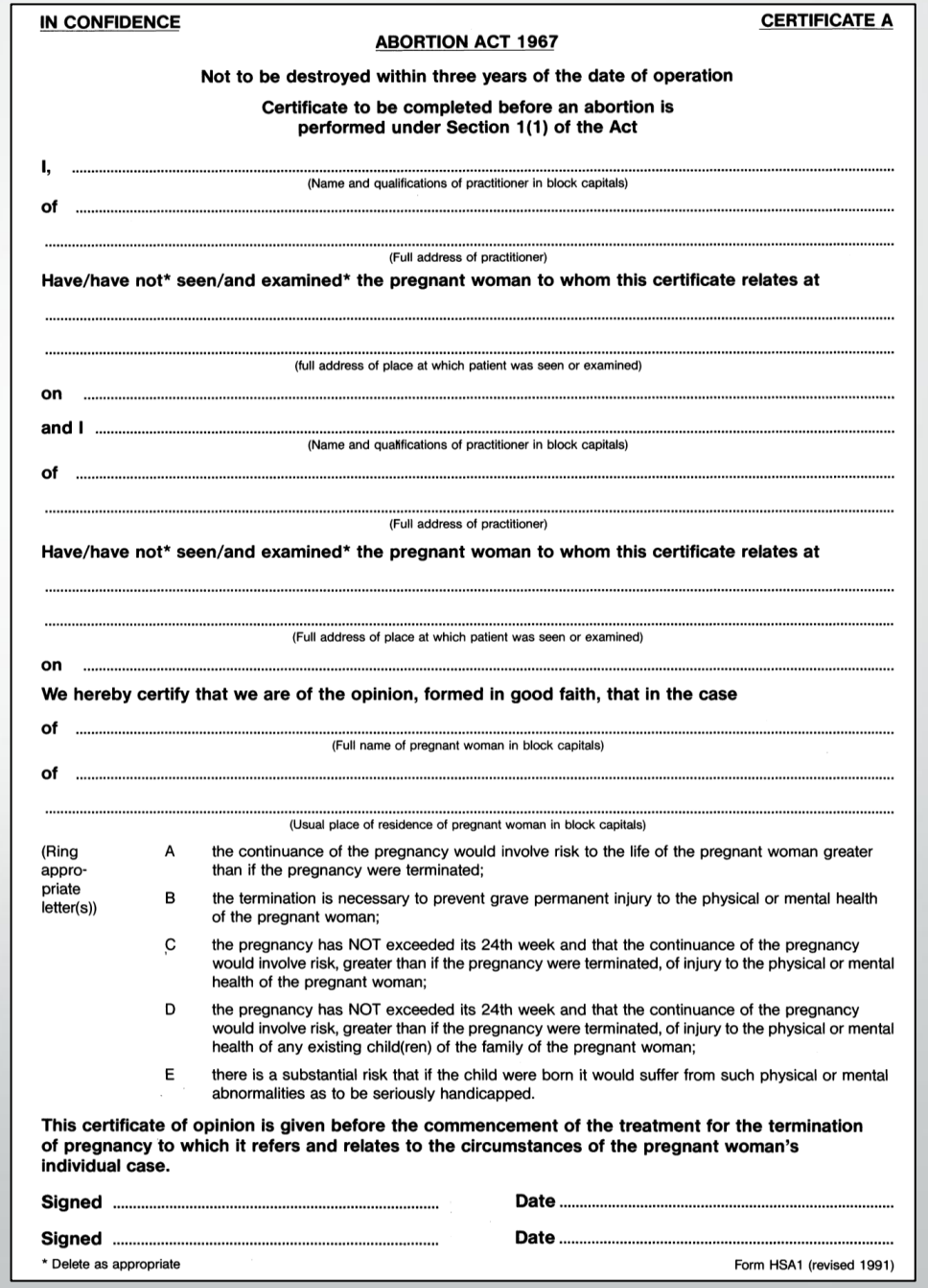
CVS	Amniocentesis
11-14 weeks	From 15+3 weeks (? to avoid 24-32w?)
0.5% risk miscarriage	- 0.1-0.5% risk miscarriage - 1-3% risk of preterm birth if late
~1% risk of confined placental mosaicism	Fetal DNA
3-5 days PCR ~14 days SNP array 14-21 days FAGP Specific testing (BW, sickle cell, …)	3-5 days PCR ~14 days SNP array 14-21 days FAGP Specific testing (BW, sickle cell, …)
Local to the skin offered	No local anesthetic offered
Not possible or advisable: - Overlying bowel - Placenta low posterior - Placenta posterior and retroverted uterus	Not possible or advisable: - Overlying bowel - Chorioamniotic membrane separation - Anhydramnios

Options in detail: TOP

<24 weeks	>24 weeks
Indication: - Social (clause D) - Maternal (clause A, B,C) - Fetal (clause E)	Indication: - Fetal (clause E)
1st line: medical termination of pregnancy If patient wants surgical can go to BPAS, ?surgical available in South Wales	1st line: medical termination of pregnancy Discourage surgical but in rare cases possible: hysterotomy
Risk: bleeding, infection, incomplete procedure, evacuation of the uterus	Risk: bleeding, infection, incomplete procedure, evacuation of the uterus
up to 21+6: feticide can be offered from 22 week: feticide recommended	Feticide recommended
If no genetic test done yet: - If feticide is performed: Offer FBS for genetic testing - Offer postnatal genetic test (higher failure rate)	If no genetic test done yet: - Offer FBS for genetic testing - Offer postnatal genetic test (higher failure rate)

Options in detail: MTOP

- MTOP medication protocol in C+V:
  - No previous C-section:
    - Day 1: Mifepristone 200mcg
    - Day 3: Misoprostol dosing depends on gestation
  - Previous C-section:
    - Day 1: Mifepristone 600mcg
    - Day 2: Mifepristone 600mcg
    - Day 3: Misoprostol reduced dosing depends on gestation



IN CONFIDENCE — CERTIFICATE A

ABORTION ACT 1967

Not to be destroyed within three years of the date of operation

Certificate to be completed before an abortion is performed under Section 1(1) of the Act

I, ... (Name and qualifications of practitioner in block capitals)

of .....

..... (Full address of practitioner)

Have/have not* seen/and examined* the pregnant woman to whom this certificate relates at

.....

..... (full address of place at which patient was seen or examined)

on .....

and I ..... (Name and qualifications of practitioner in block capitals)

of .....

..... (Full address of practitioner)

Have/have not* seen/and examined* the pregnant woman to whom this certificate relates at

.....

..... (Full address of place at which patient was seen or examined)

on .....

We hereby certify that we are of the opinion, formed in good faith, that in the case

of ..... (Full name of pregnant woman in block capitals)

of .....

..... (Usual place of residence of pregnant woman in block capitals)

(Ring appropriate letter(s))

A the continuance of the pregnancy would involve risk to the life of the pregnant woman greater than if the pregnancy were terminated;

B the termination is necessary to prevent grave permanent injury to the physical or mental health of the pregnant woman;

C the pregnancy has NOT exceeded its 24th week and that the continuance of the pregnancy would involve risk, greater than if the pregnancy were terminated, of injury to the physical or mental health of the pregnant woman;

D the pregnancy has NOT exceeded its 24th week and that the continuance of the pregnancy would involve risk, greater than if the pregnancy were terminated, of injury to the physical or mental health of any existing child(ren) of the family of the pregnant woman;

E there is a substantial risk that if the child were born it would suffer from such physical or mental abnormalities as to be seriously handicapped.

This certificate of opinion is given before the commencement of the treatment for the termination of pregnancy to which it refers and relates to the circumstances of the pregnant woman's individual case.

Signed ..... **Date** .....

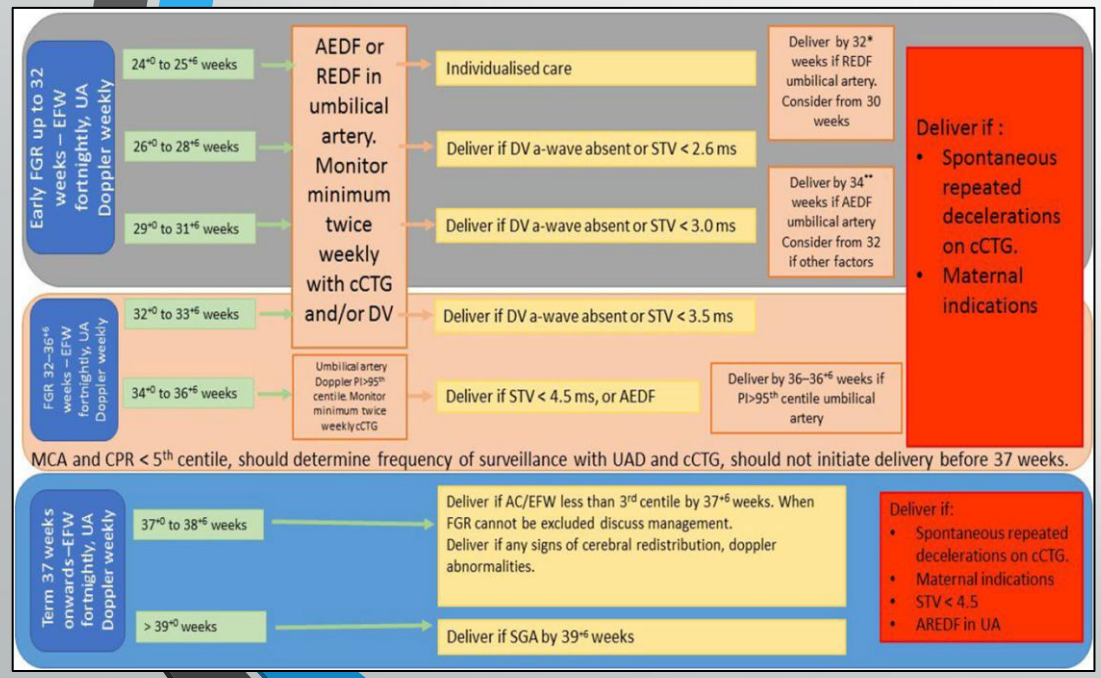
Signed ..... **Date** .....

* Delete as appropriate

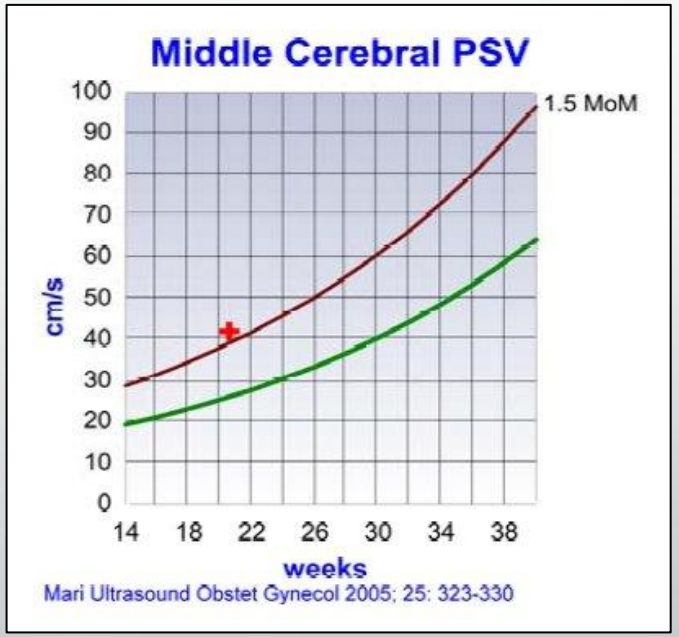
Form HSA1 (revised 1991)

Options in detail: deliver

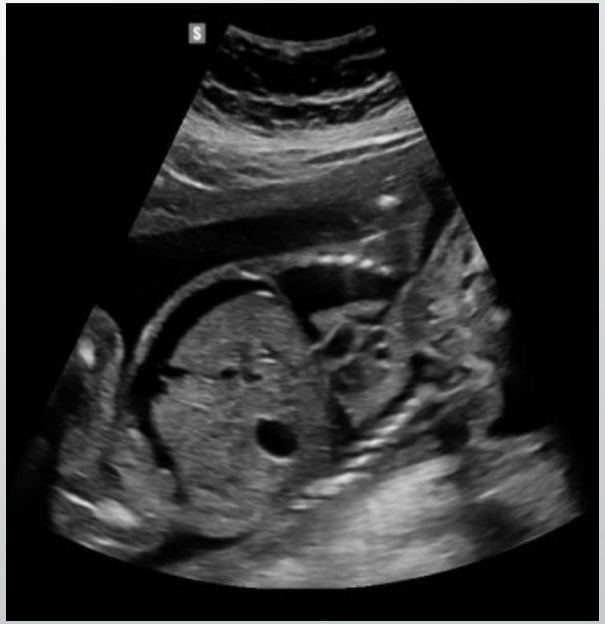
- If all test normal at Term: good prognosis: 90-95%
- Earlier depending on gestation and pathology:
  - **SGA**



Fetal anaemia



Hydrops



Sources:

- https://fetalmedicine.org/nuchal-translucency-scan
- Souka AP, Von Kaisenberg CS, Hyett JA, Sonek JD, Nicolaides KH. Increased nuchal translucency with normal karyotype. Am J Obstet Gynecol. 2005 Apr;192(4):1005-21. Doi: 10.1016/j.ajog.2004.12.093.
- Morris RK, Johnstone E, Lees C, Morton V, Smith G; the Royal College of Obstetricians and Gynaecologists. Investigation and Care of a Small-for-Gestational-Age Fetus and a Growth Restricted Fetus (Green-top Guideline No. 31). *BJOG*. 2024; 131(9): e31–e80.
- Mari G, Deter RL, Carpenter RL, Rahman F, Zimmerman R, Moise KJ Jr, Dorman KF, Ludomirsky A, Gonzalez R, Gomez R, Oz U, Detti L, Copel JA, Bahado-Singh R, Berry S, Martinez-Poyer J, Blackwell SC. Non-invasive diagnosis by Doppler ultrasonography of fetal anaemia due to maternal red-cell alloimmunization. Collaborative Group for Doppler Assessment of the Blood Velocity in Anaemic Fetuses. N Engl J Med. 2000 Jan 6;342(1):9-14.

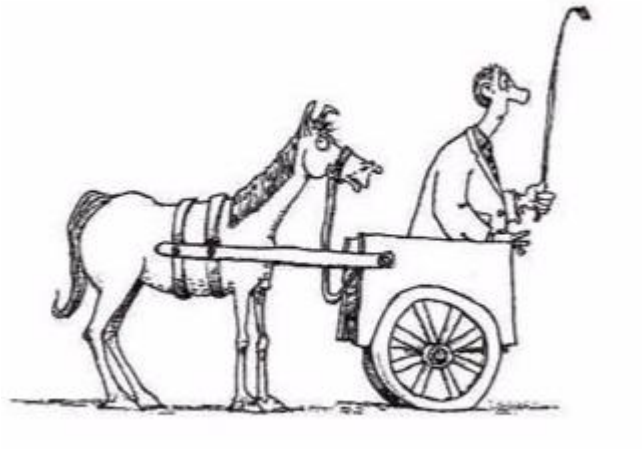
HYPOPLASTIC LEFT HEART - PRENATAL DIAGNOSIS AND POSTNATAL MANAGEMENT PATHWAY

DR ALAN PATEMAN – Consultant Paediatric Cardiologist, UHW.

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

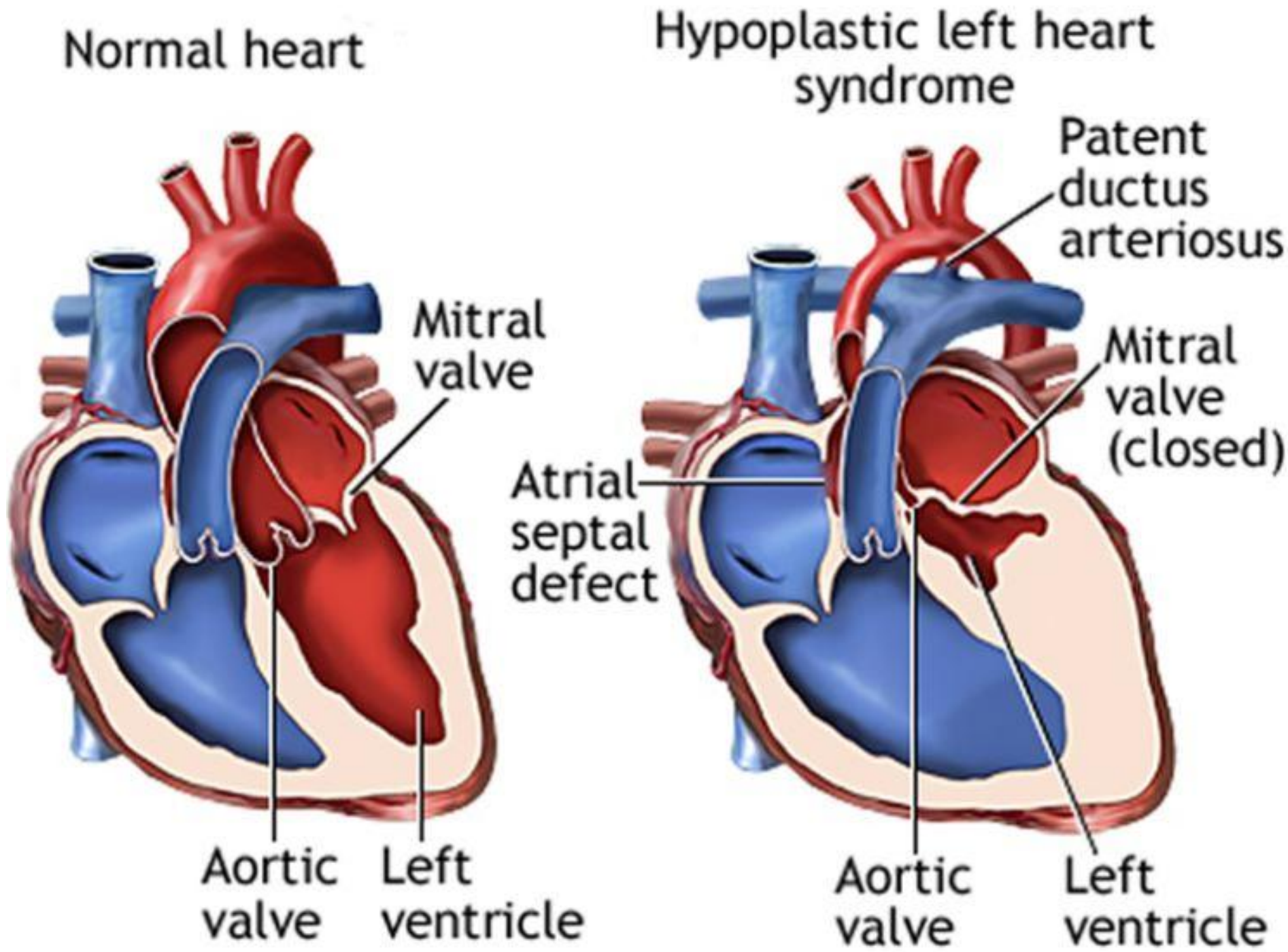
CONTENTS

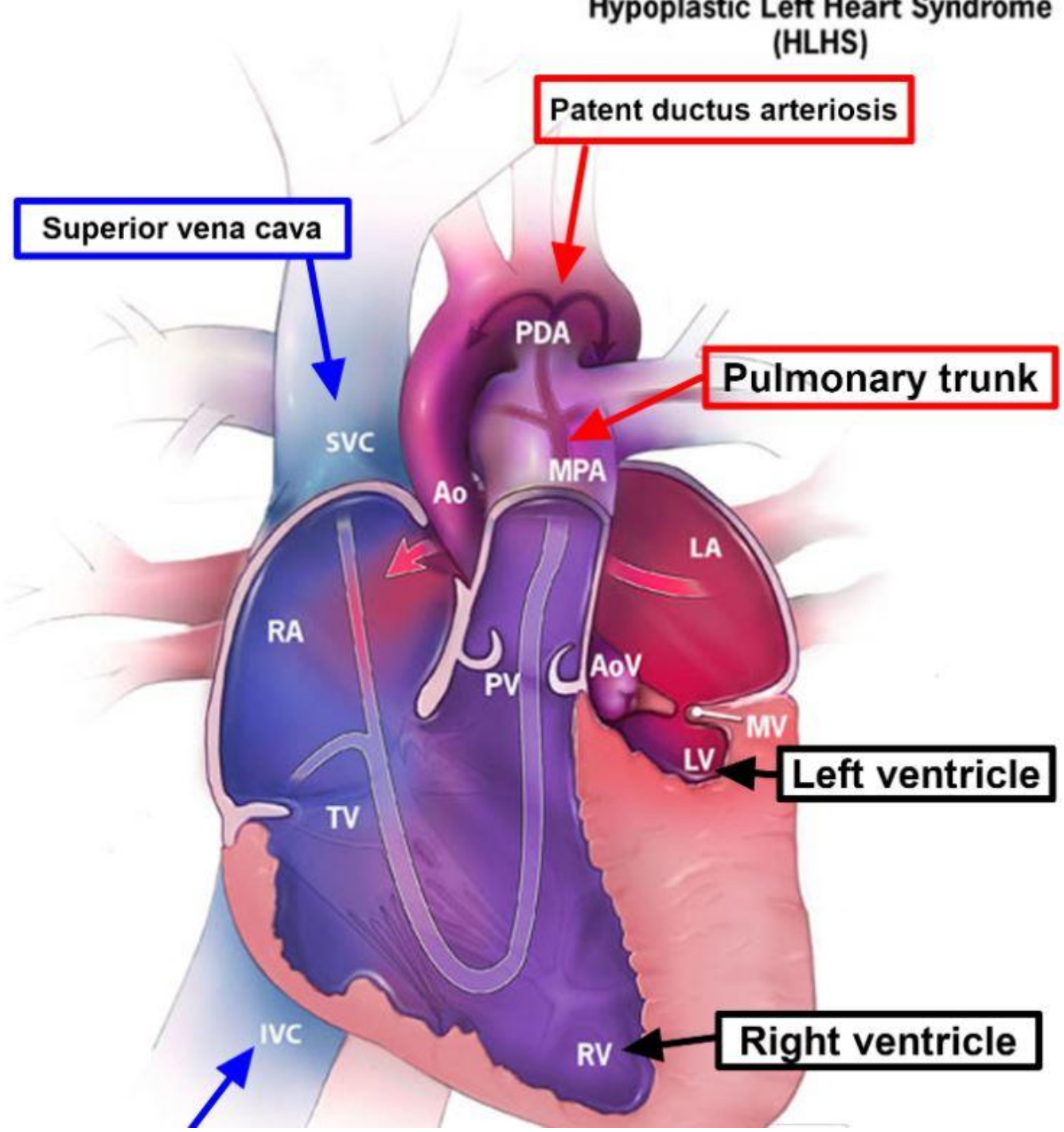
- What is Hypoplastic Left Heart Syndrome?
- Postnatal management – a chronologic walk through



- Fetal Diagnosis

Hypoplastic Left Heart


Normal heart
Mitral valve
Aortic valve
Left ventricle
Hypoplastic left heart syndrome
Patent ductus arteriosus
Mitral valve (closed)
Atrial septal defect
Aortic valve
Left ventricle


Hypoplastic Left Heart Syndrome
(HLHS)
Patent ductus arteriosis
Superior vena cava
PDA
Pulmonary trunk
SVC
MPA
Ao
LA
RA
PV
AoV
MV
LV
Left ventricle
TV
IVC
RV
Right ventricle

Immediate management after birth

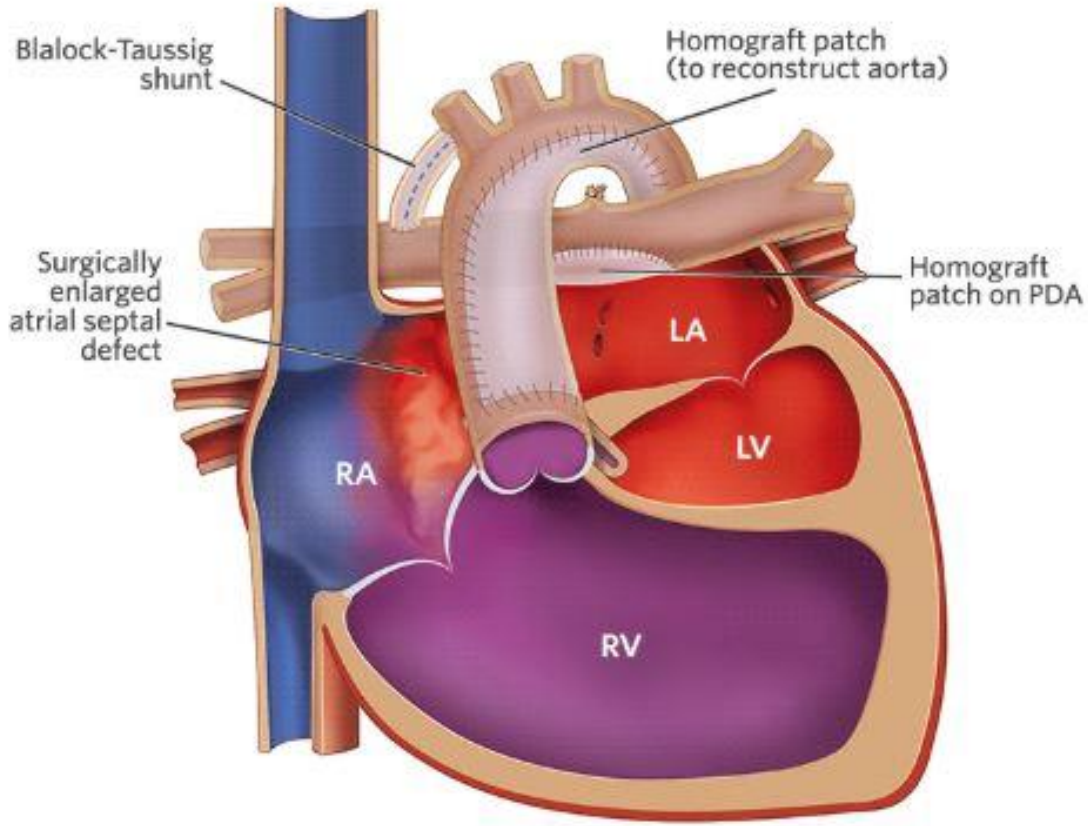
- Delivery in surgical centre
- Prostin after birth
- ICU management

- Possibly:
- Balloon Atrial Septostomy – if restrictive atrial septum
- Hybrid procedure if small and unwell – PDA stent plus bilateral PA bands

Norwood procedure

First week of life

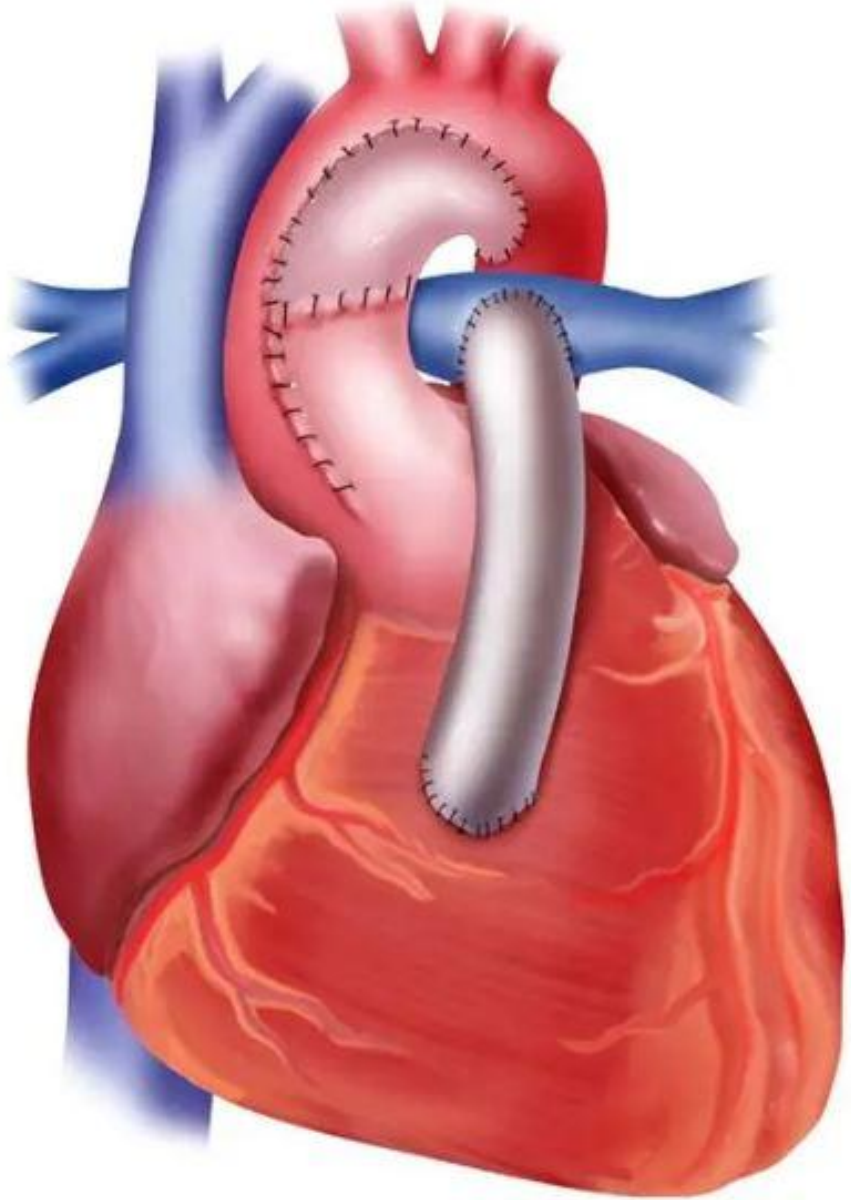
Hypoplastic Left Heart Syndrome (HLHS)
Stage 1 – Norwood



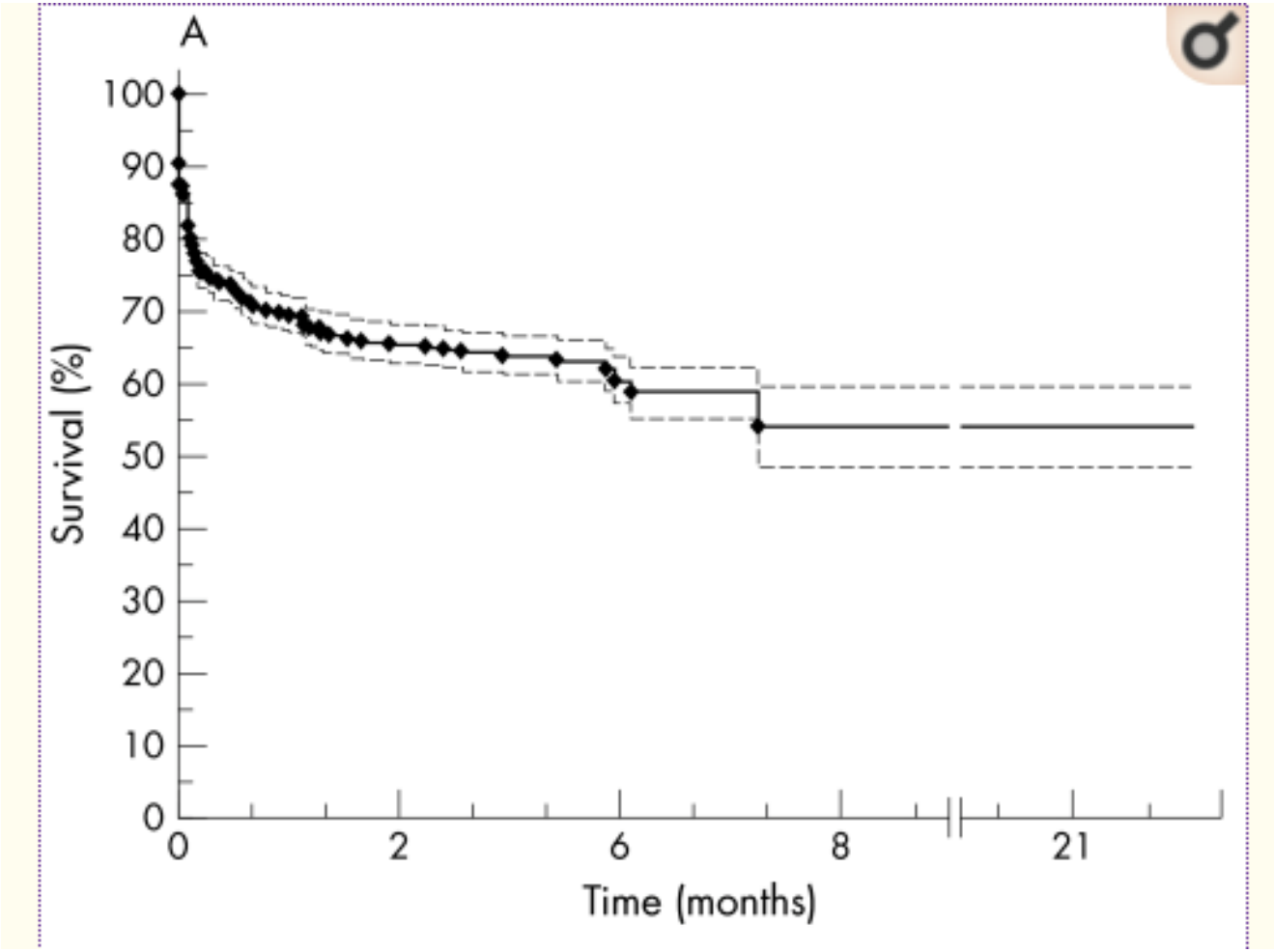
- Oxygen-rich blood
- Oxygen-poor blood
- Mixed blood
- Mixed blood

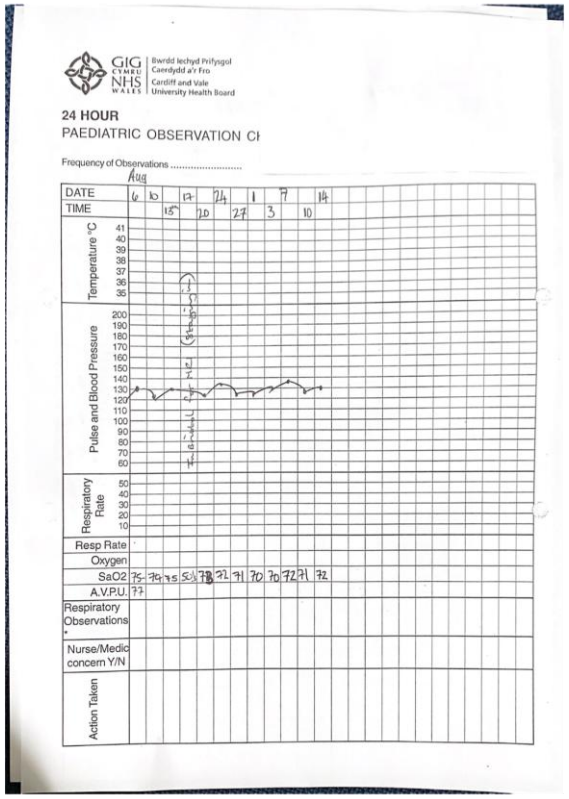
AO: Aorta
LA: Left atrium
RA: Right atrium

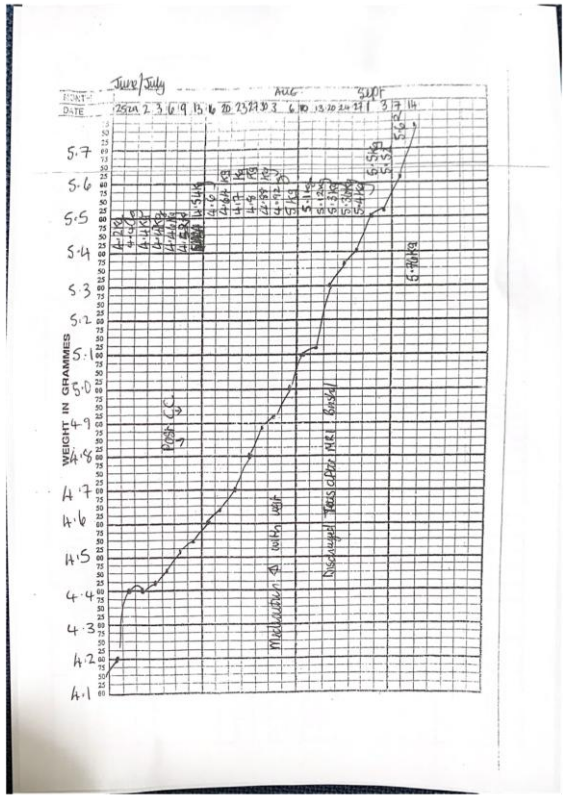
PA: Pulmonary artery
LV: Left ventricle
RV: Right ventricle

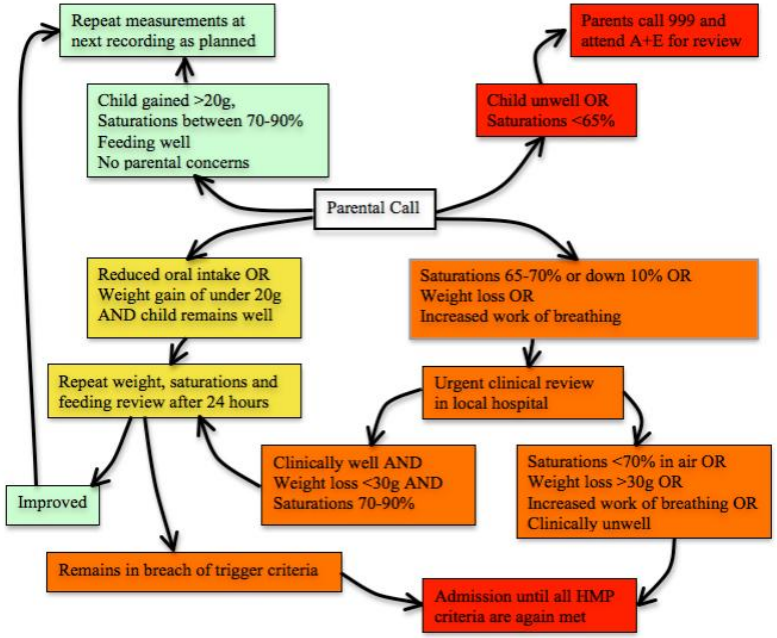


Norwood actuarial survival







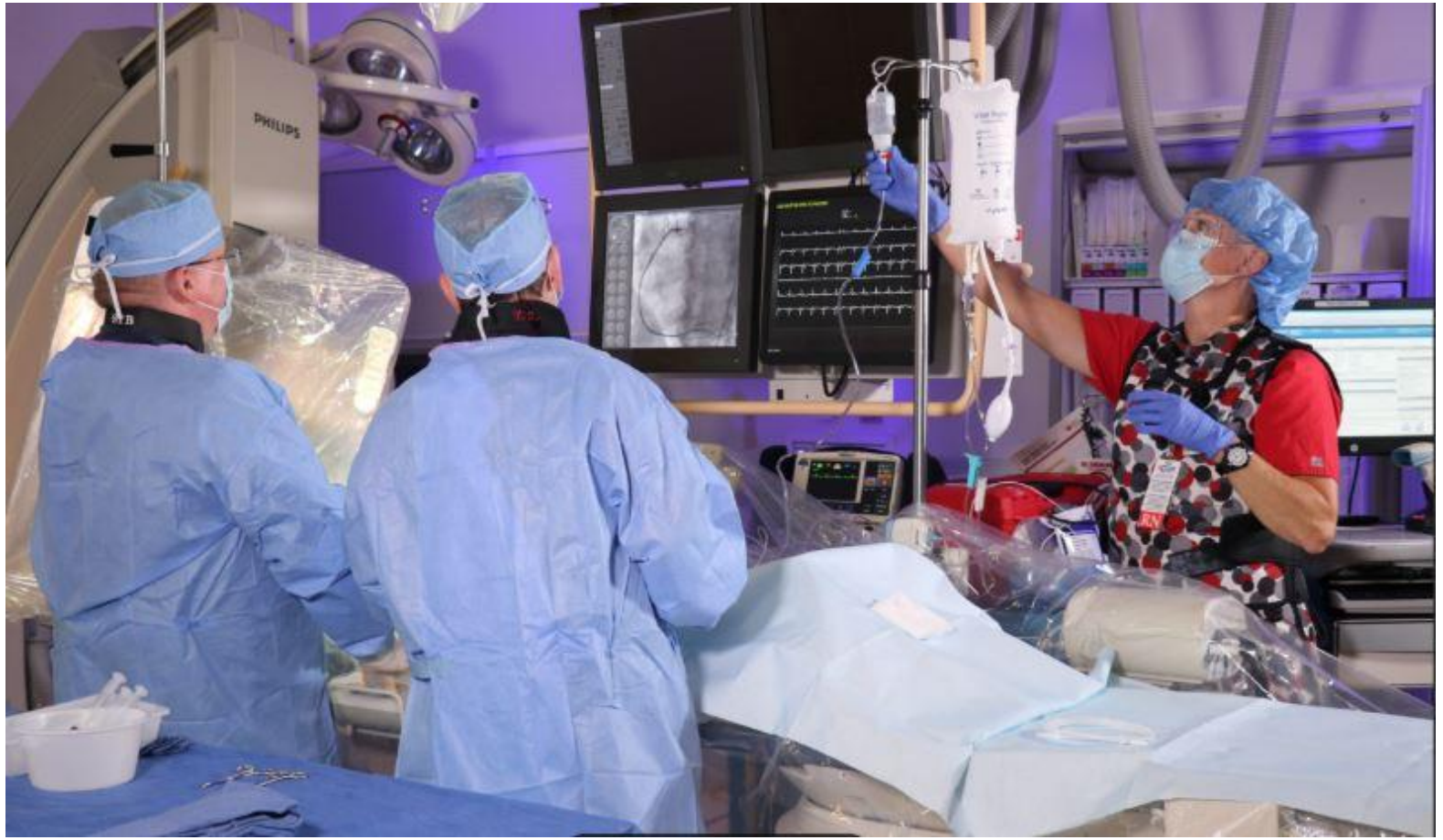


Home Monitoring Programme

designed to detect problems early

- Regular OPD with cardiologist
- Often have additional dietary support requirements eg NG tube
- Saturations ideally 85% often lower
- Medications:
  - Aspirin (and clopidogrel if BT shunt) + PPI
  - Diuretics
  - Possibly ACEi
  - Anti GORD

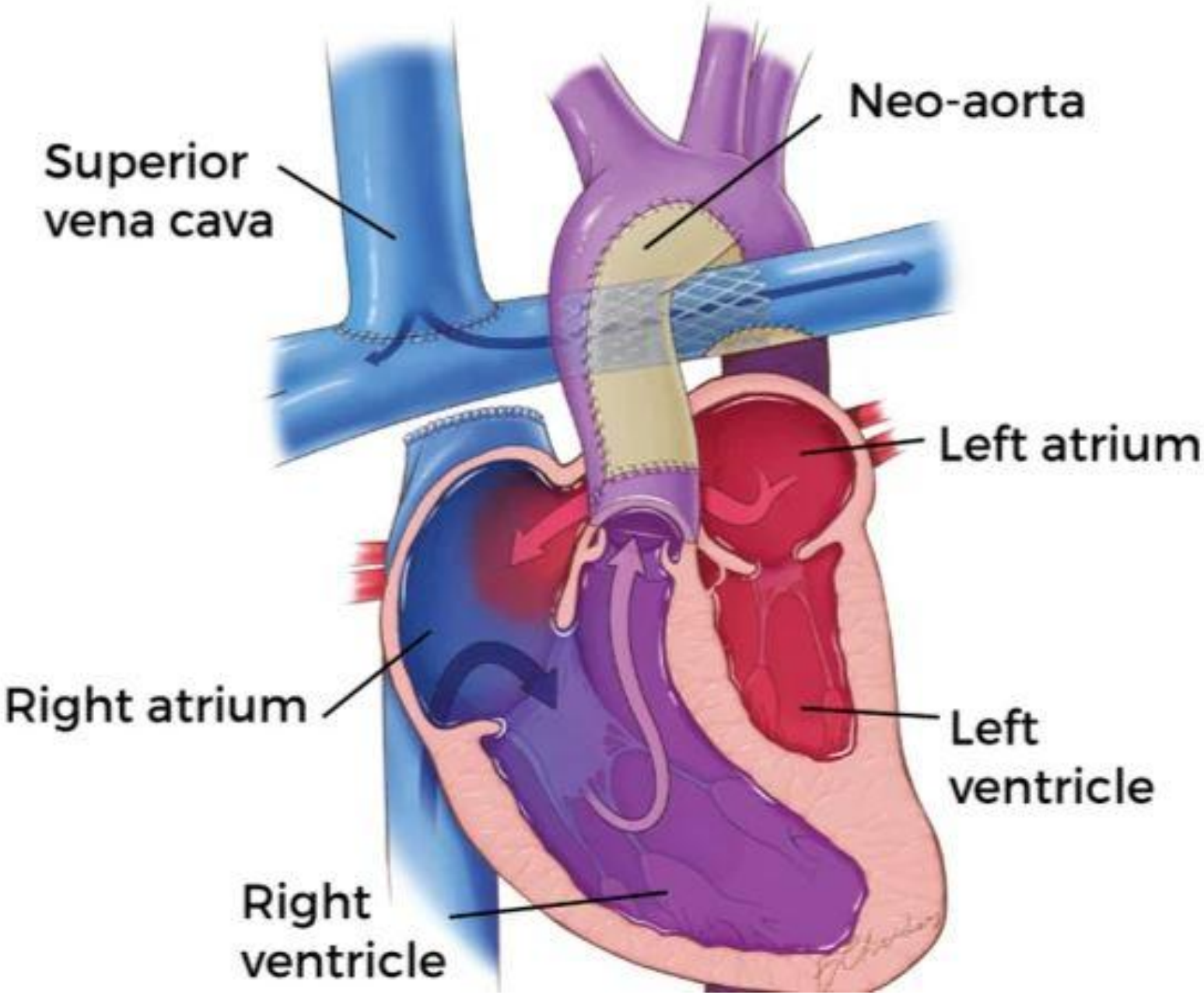
Cardiac catheter 2-3 months old



Glenn Procedure

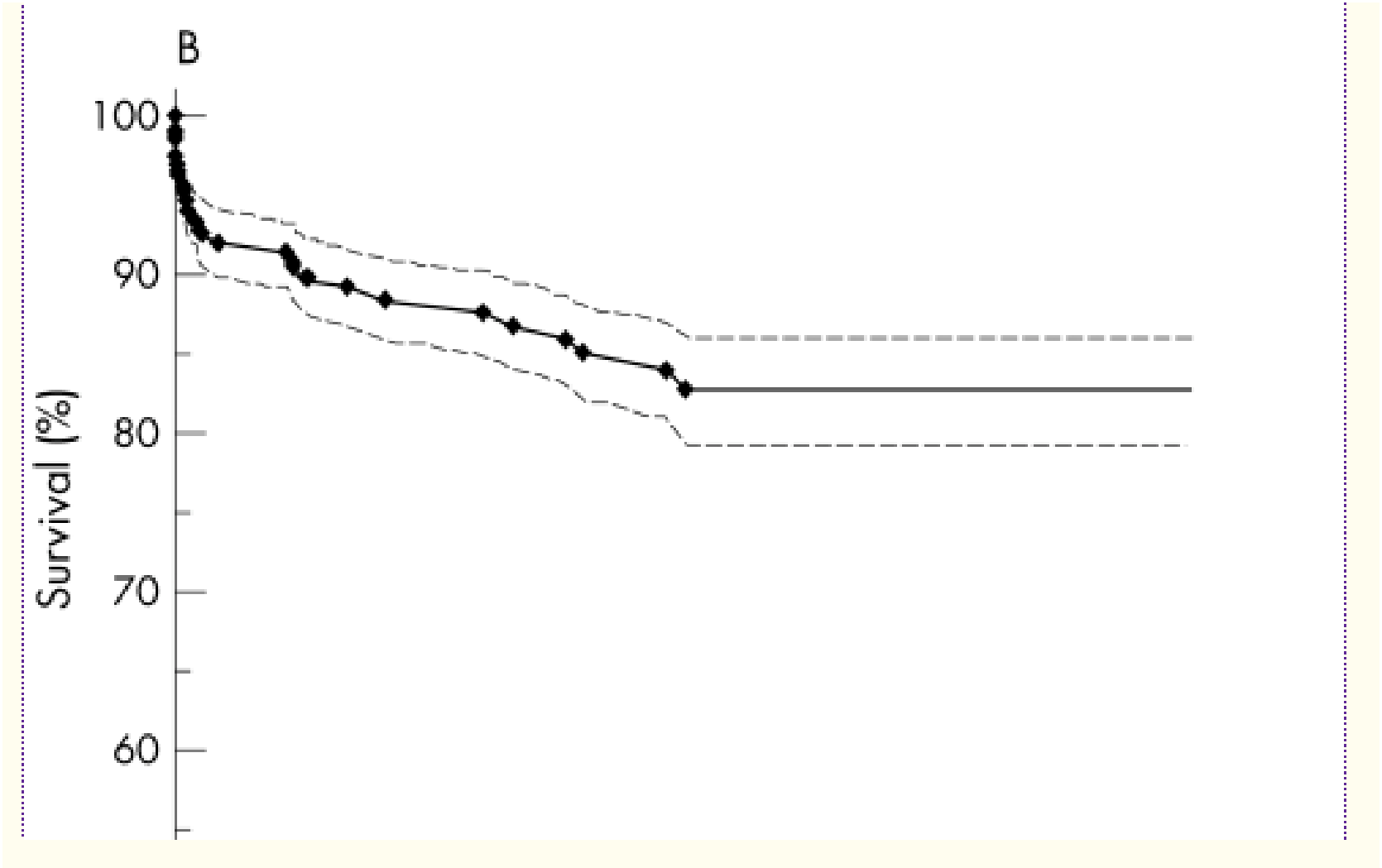
Superior cavopulmonary connection (SCVC)

3-4 months age

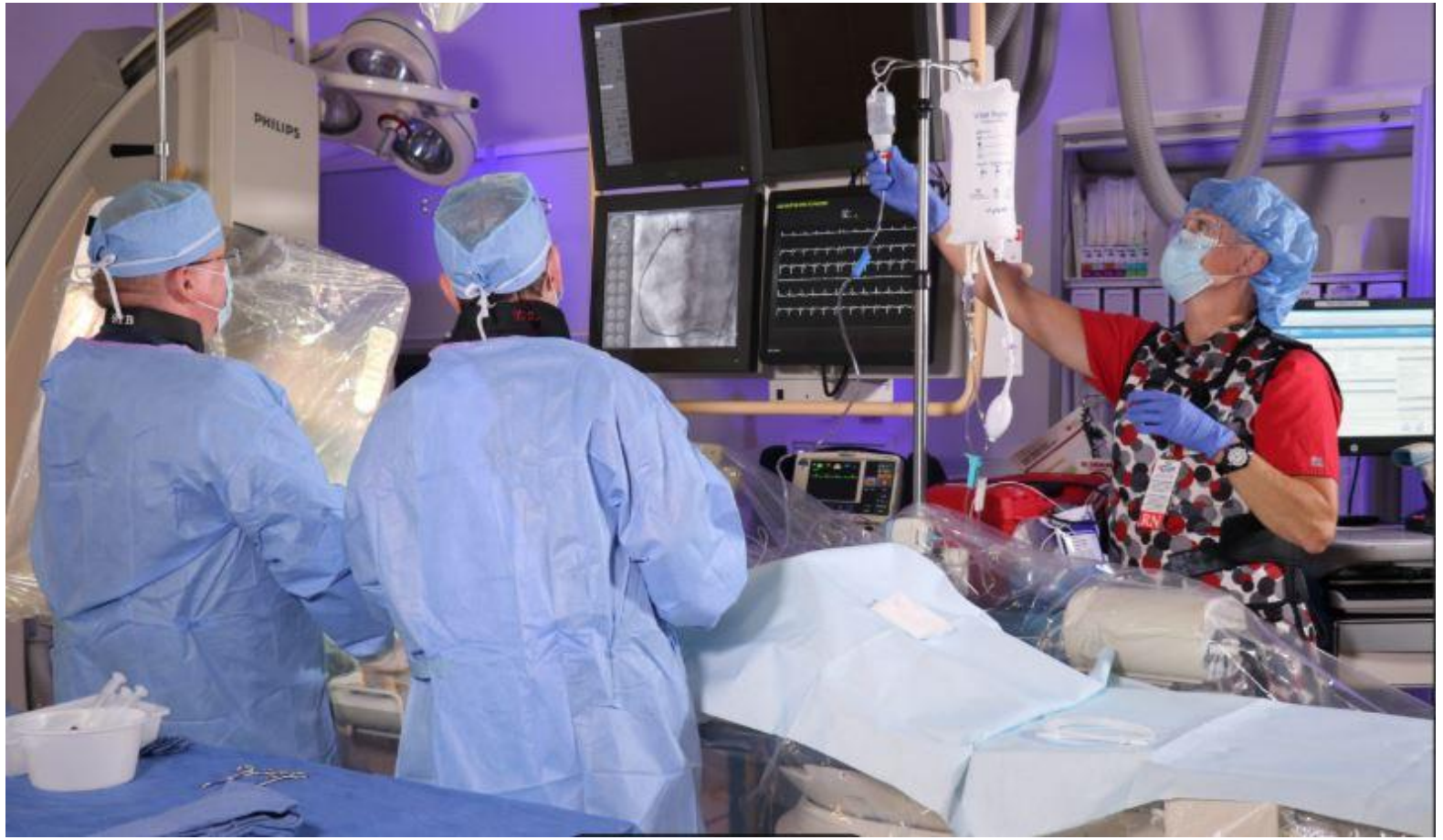

Superior
vena cava
Neo-aorta
Left atrium
Right atrium
Left
ventricle
Right
ventricle

- More stable so home monitoring programme is stopped
- OPD follow up
- Saturations – ideally 85% (often lower)
- Medications:
  - Diuretics
  - ACEi – possibly
  - Aspirin Plus PPI
  - Anti GORD

Glenn actuarial survival

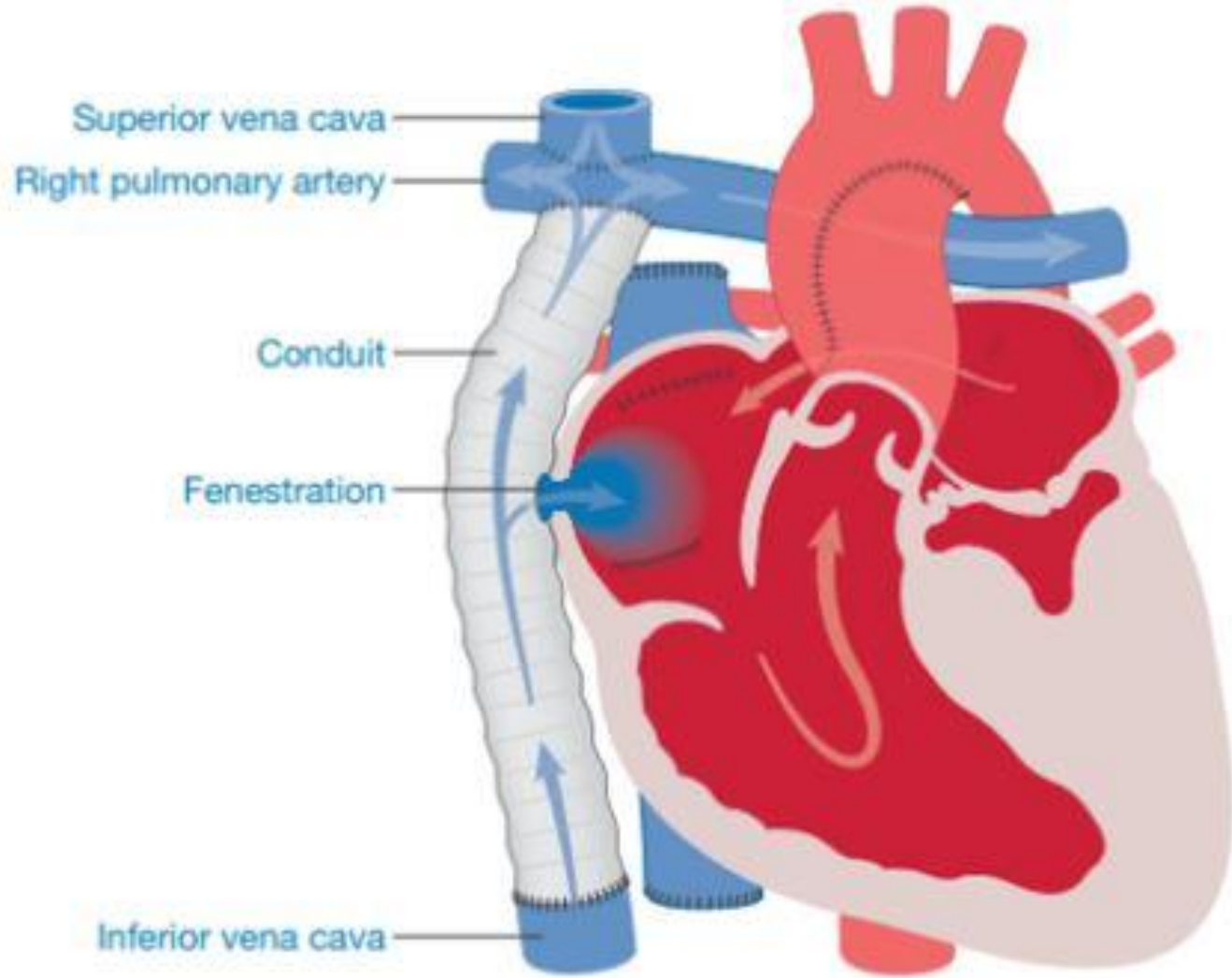


Cardiac catheter – 15kg 4-5 years approx.

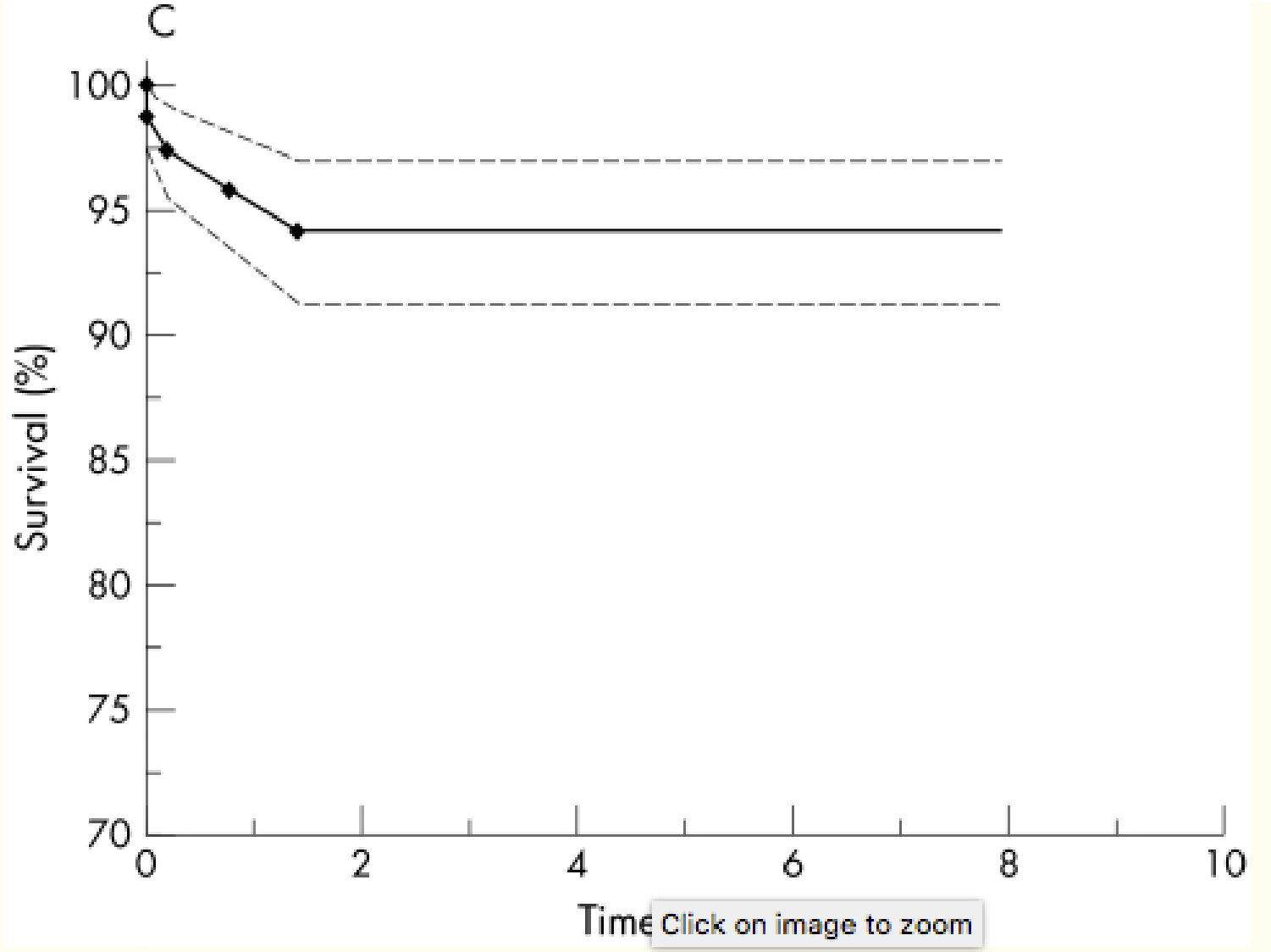


Total Cavopulmonary connection (TCPC) - Fontan

4-5 years age or approx. 15kg


Superior vena cava
Right pulmonary artery
Conduit
Fenestration
Inferior vena cava

Fontan actuarial survival



- Annual follow up with Echo, bloods, Holter
- Saturations over 90% (may still be lower)
- ‘MOT’ every five years – liver USS, 6MWT, possibly MRI in addition to annual investigations
- Medications:
  - Diuretics – possibly
  - Anticoagulation (centre dependant) Warfarin Rivaroxaban
  - ACEi – possibly

After surgery – the long term?

Staged surgical management of hypoplastic left heart syndrome: a single institution 12 year experience

S P McGuirk, M Griselli, O F Stumper, E M Rumball, P Miller, R Dhillon, J V de Giovanni, J G Wright, D J Barron, and W J Brawn

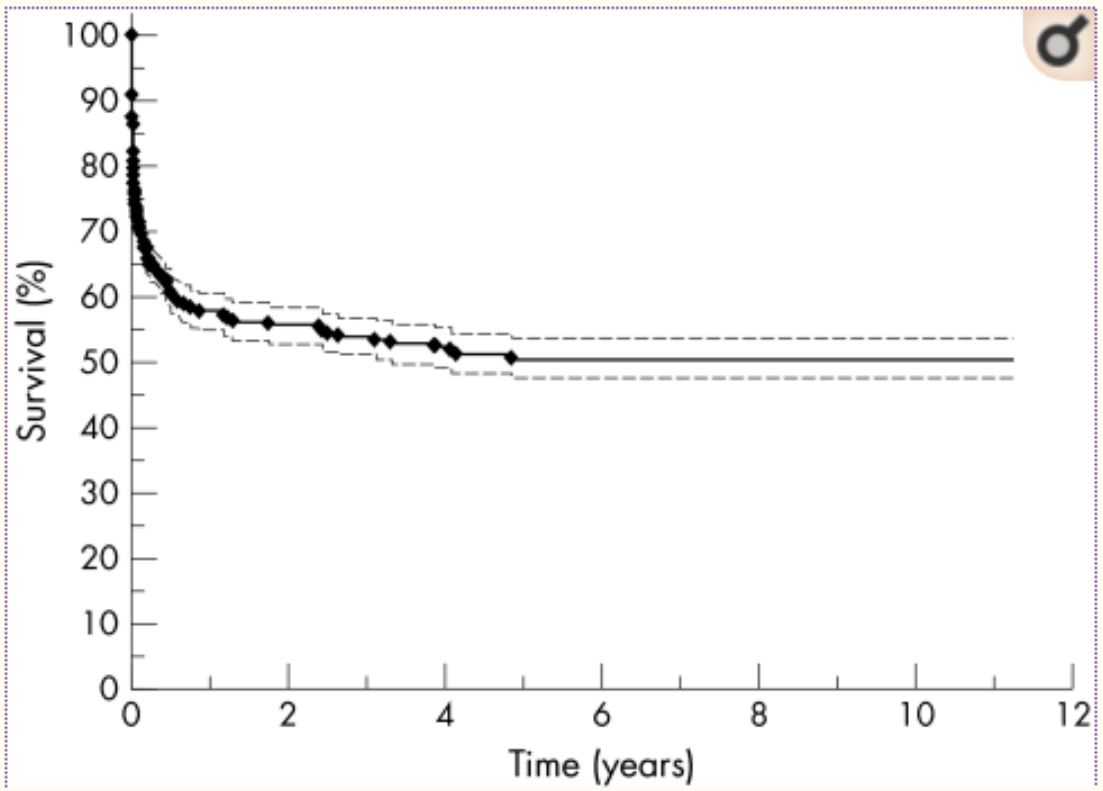
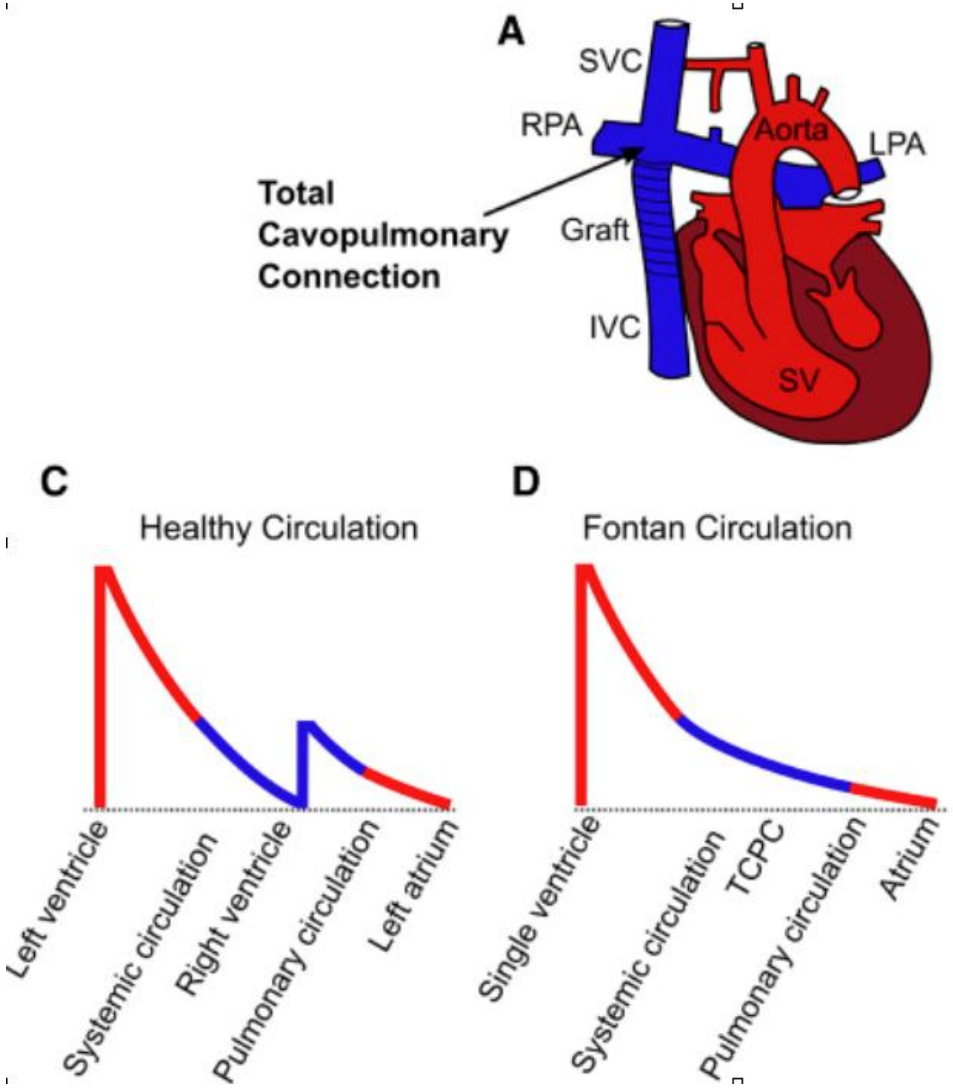


Figure 1 Actuarial survival after staged surgical management of the hypoplastic left heart syndrome. Data are mean (SD).

- 50% in mainstream education
- 50% adults need further intervention minor tweaks up to transplant (if candidate)
- Arrhythmias, Ventricular dysfunction
- Fontan Failure, PLE, Plastic bronchitis

Univentricular Physiology is different!

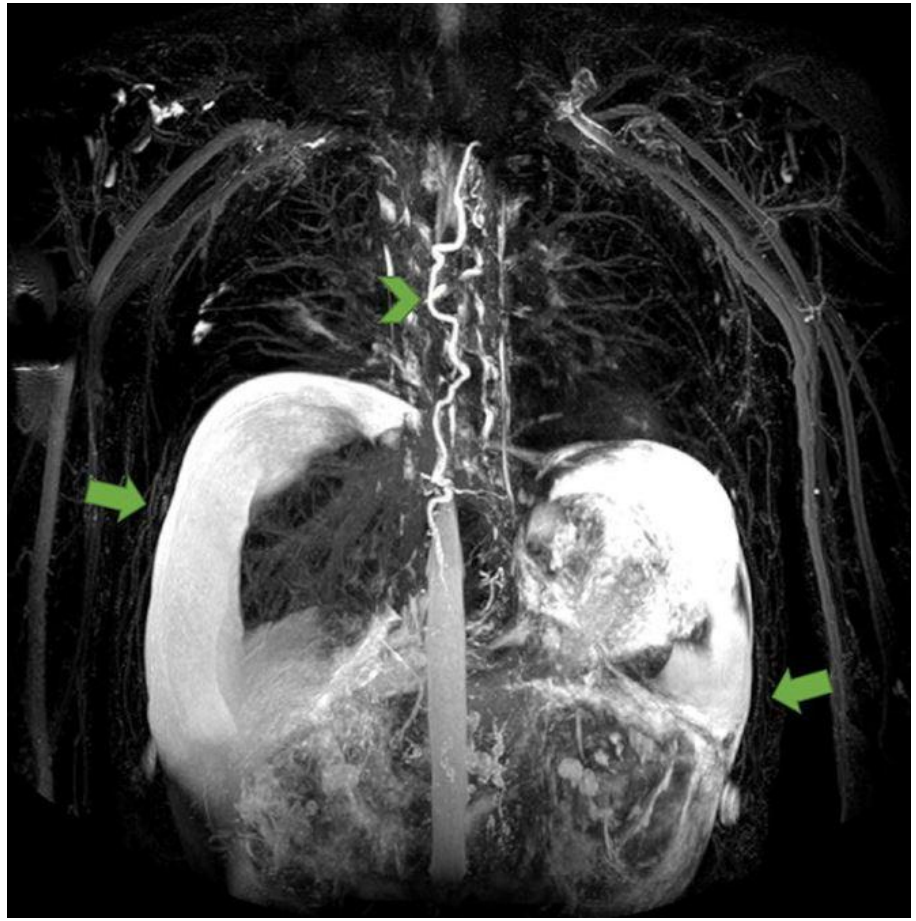


- Mobility is an important contributor to Fontan circulation

- Limited tolerance compared to peers
- Pushchair - Wheelchair

Plastic Bronchitis

PLE



Fetal Diagnosis

Case examples

Appendix 1 Checklist for cardiac screening

Upper abdomen			
	Stomach on left	☐ Yes	☐ No
Four-chamber view			
General	Heart on left, axis –45°	☐ Yes	☐ No
	Heart area ≤ ⅓ chest area	☐ Yes	☐ No
	Regular rhythm, rate 120–160 bpm	☐ Yes	☐ No
Atria	Approximately equal in size	☐ Yes	☐ No
	Patent foramen ovale; foramen ovale flap valve in left atrium	☐ Yes	☐ No
	At least one pulmonary vein entering left atrium	☐ Yes	☐ No
Atrioventricular junction	Two separate valves that open and close freely	☐ Yes	☐ No
	Tricuspid valve more apical than mitral valve (normal valve offset)	☐ Yes	☐ No
Ventricles	Approximately equal in size	☐ Yes	☐ No
	Moderator band at apex of right ventricle	☐ Yes	☐ No
	Septum appears intact	☐ Yes	☐ No
Left ventricular outflow-tract view			
	Vessel in continuity with ventricular septum and does not bifurcate	☐ Yes	☐ No
	Aortic valve leaflets not thickened, open and close freely	☐ Yes	☐ No
Right ventricular outflow-tract view/three-vessel view			
	Vessel arising from right ventricle is anterior to aorta and bifurcates	☐ Yes	☐ No
	Great arteries crossover	☐ Yes	☐ No
	Pulmonary valve leaflets are not thickened, open and close freely	☐ Yes	☐ No
Three-vessel-and-trachea view			
	V-sign (ductal and aortic arches to left of trachea)	☐ Yes	☐ No
	Ductal and aortic arches similar in size	☐ Yes	☐ No

- **ISUOG Practice Guidelines fetal cardiac screening 2023**

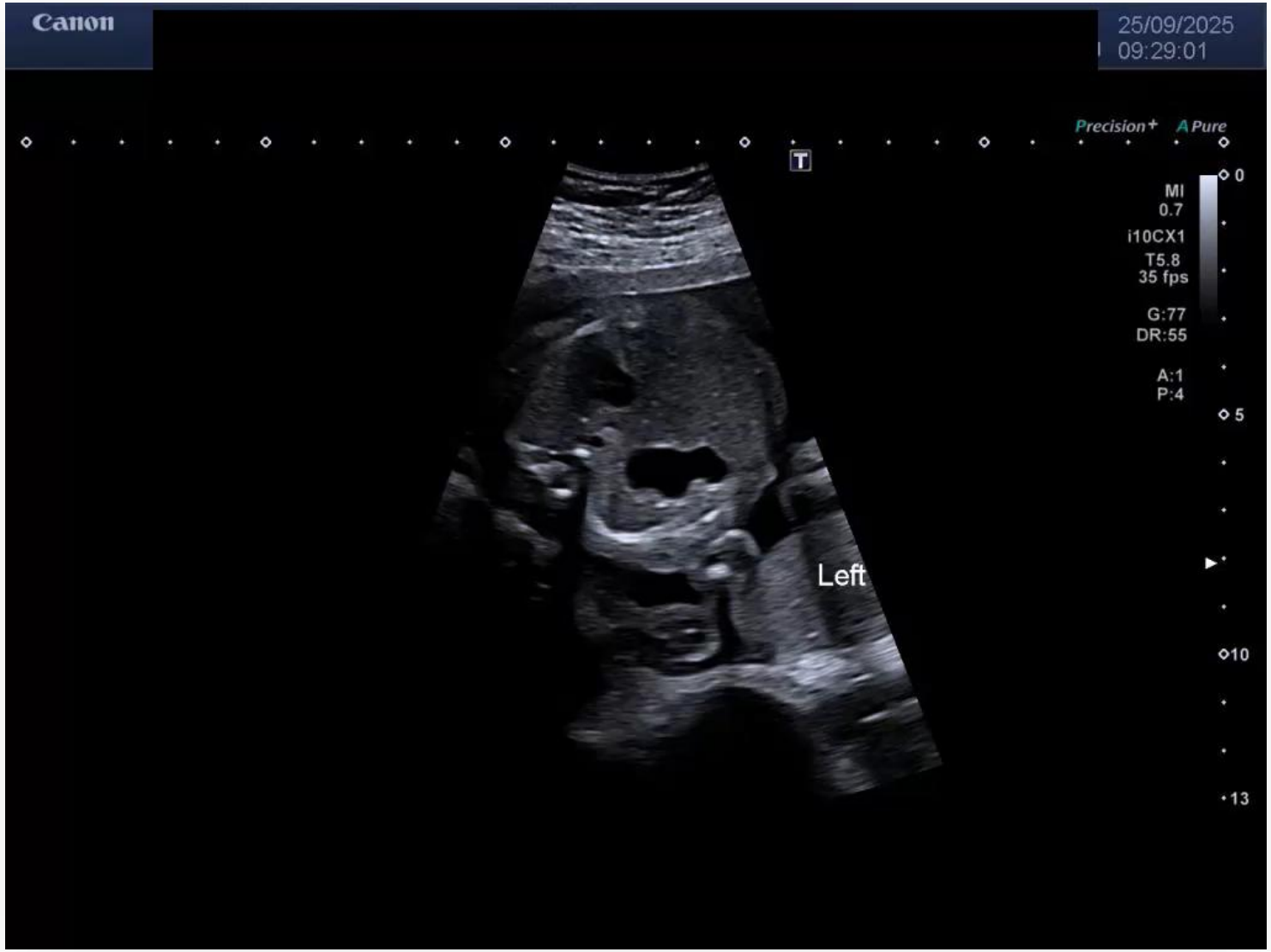
Anatomy	Present/absent: comments. (HLHS Example)
Stomach position	Left abdomen
Heart position and Size and Axis	Left thorax apex left, normal size and axis
Rate and Rhythm	SR 140bpm
Heart Function	Visually good contraction
IVC (ductus venosus)	Connects to RA, systemic venous Doppler trace
SVC	Rt side to RA systemic venous Doppler trace
RA	Usual position and size, connects via TV to RV
TV	Normal size no stenosis , may have regurgitation
RV	Good size and function, connects via PV to MPA
PV	No stenosis or regurgitation, Normal size
MPA	Good sized with confluent BPA
BPA	Laminar flow LPA and RPA good size
Pulmonary veins	Connect to LA, Doppler pattern (normal or reversed flow pattern)
LA	Small connects to LV via MV
MV	Small, stenotic or atretic, ? regurgitation if patent
LV	Small or absent, non-apex forming may show EFE and poor contraction
AoV	Small, atretic or stenotic, AAo can be difficult to see
Aorta	Small, antegrade or retrograde flow? Left arch – hypoplastic.
PFO	Left to right, restrictive?
Interventricular septum	Intact

Case 1

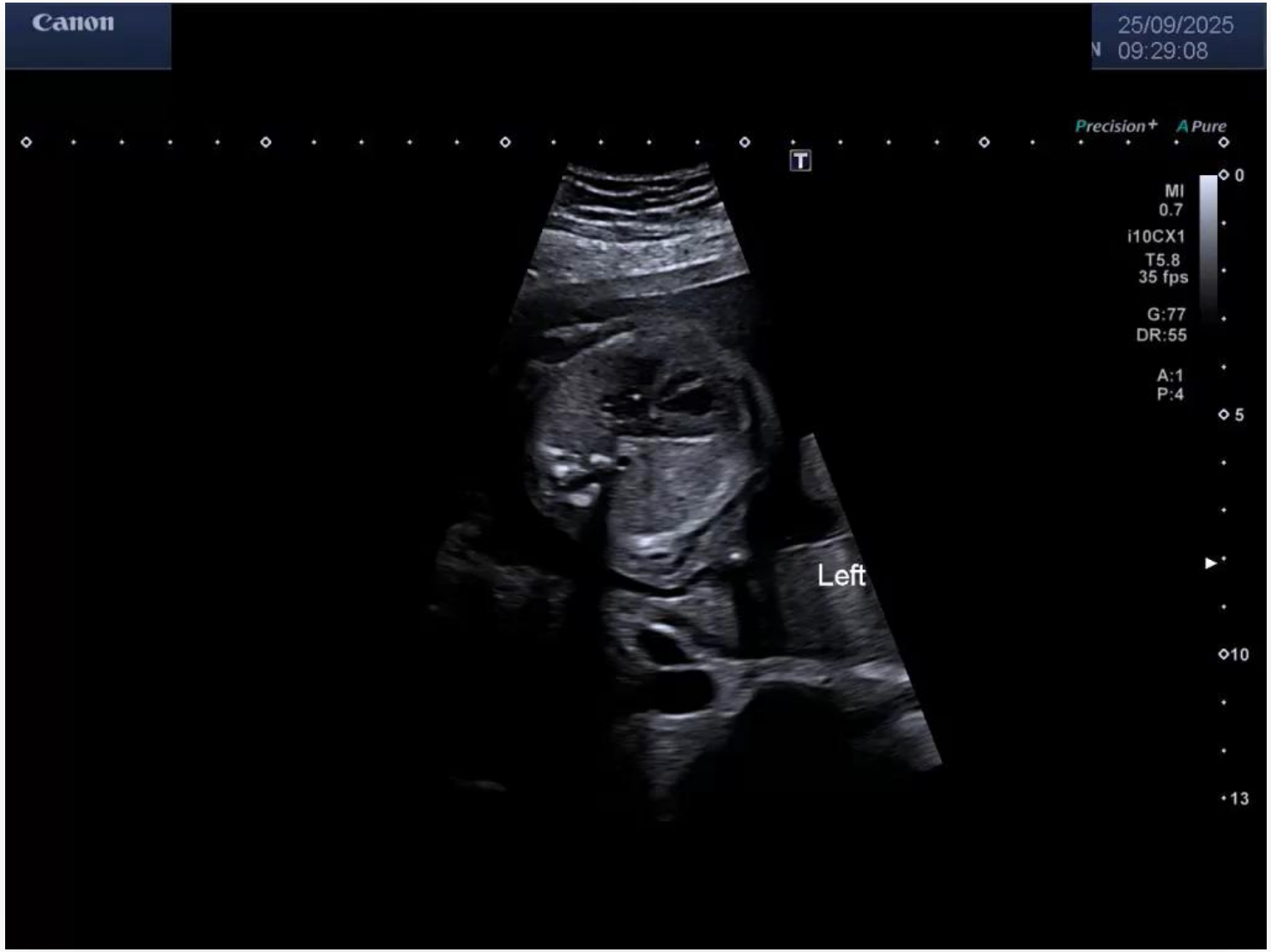
‘full’ study

Initial sweep

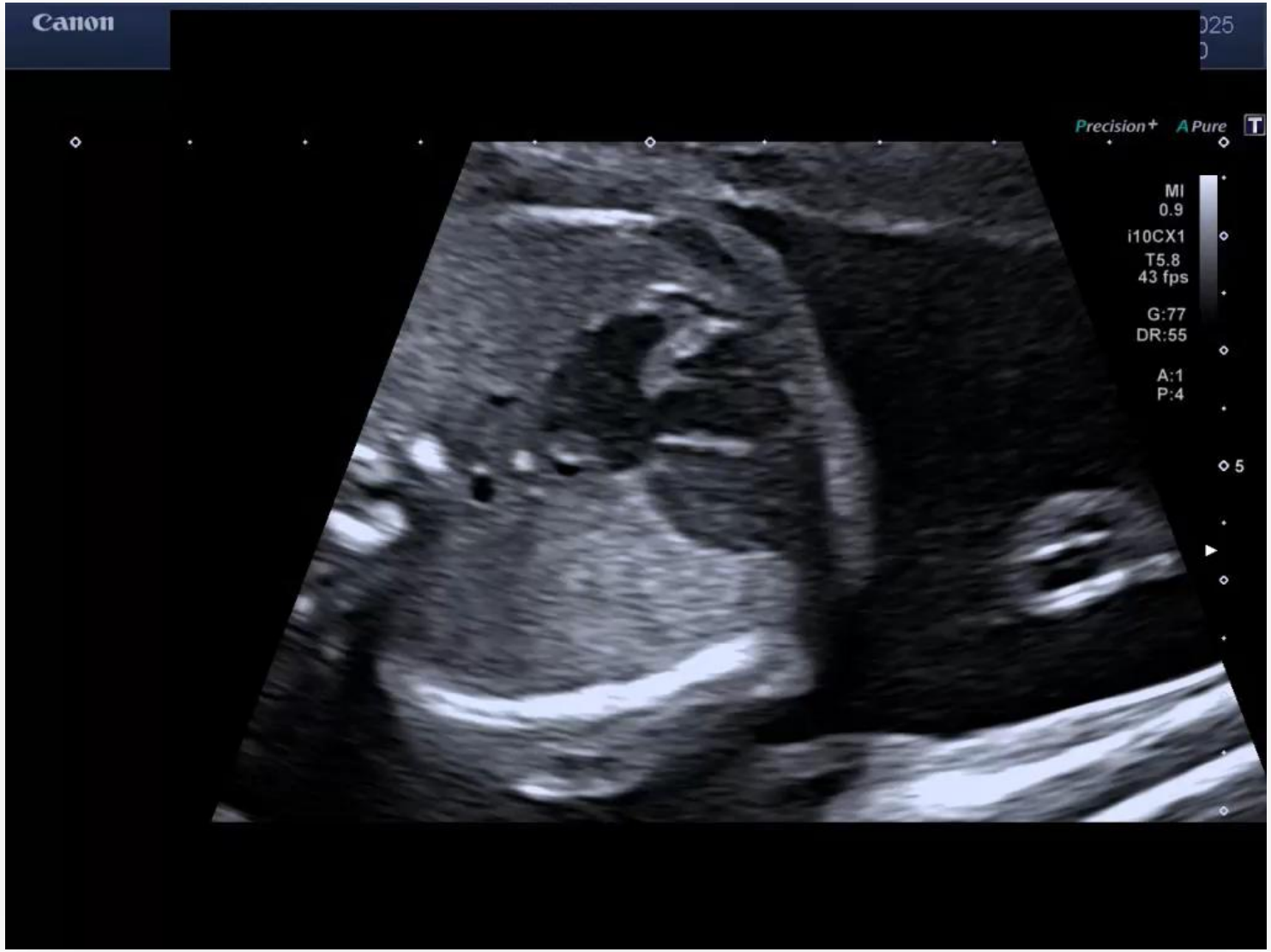
- Initial Sweep
- Establish situs
- Pass through 4CV
- Up to 3VT



- Initial sweep
- 3VT better imaged

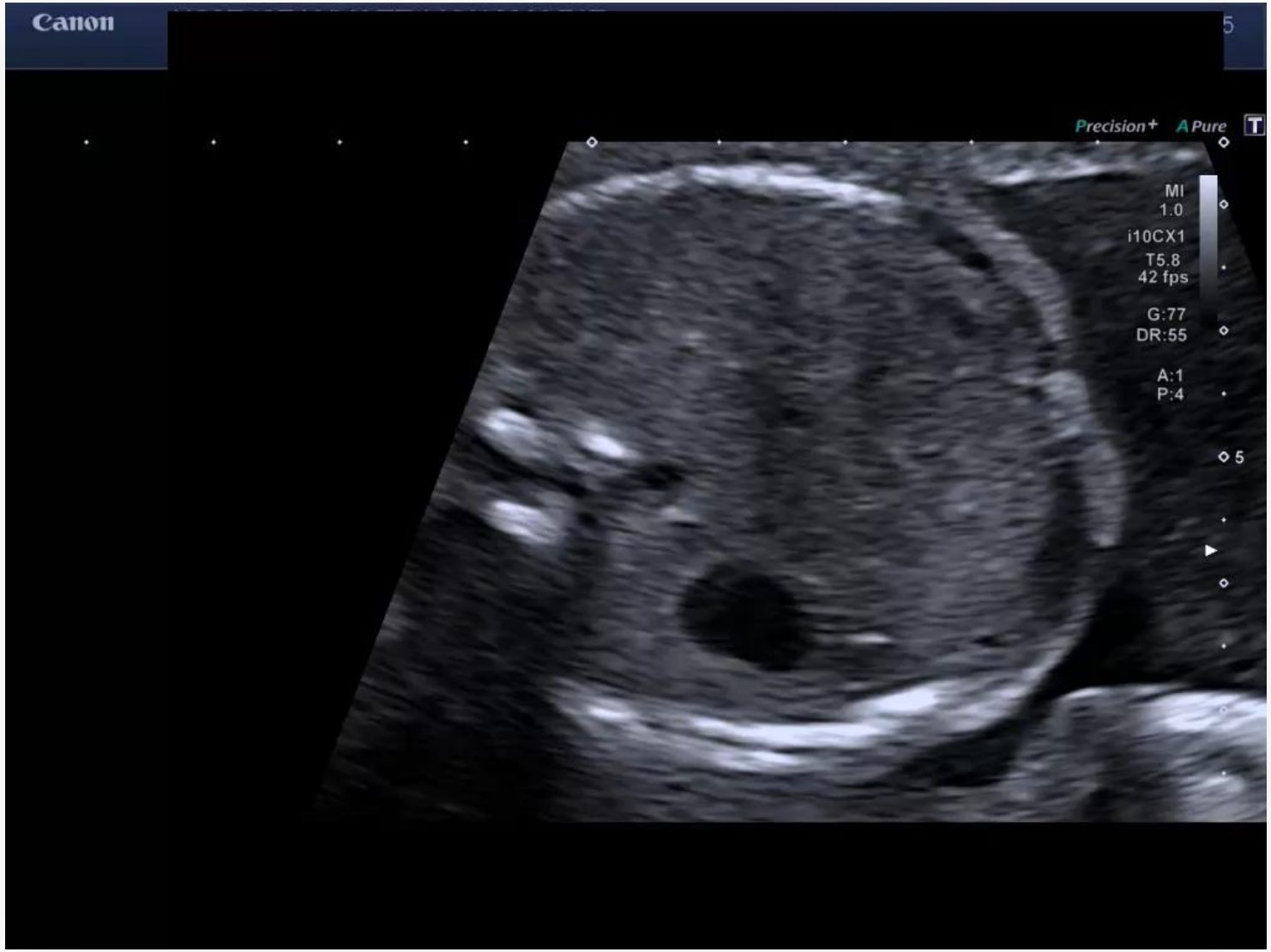


- Zoomed and optimized
- 4CV
- No obvious LV

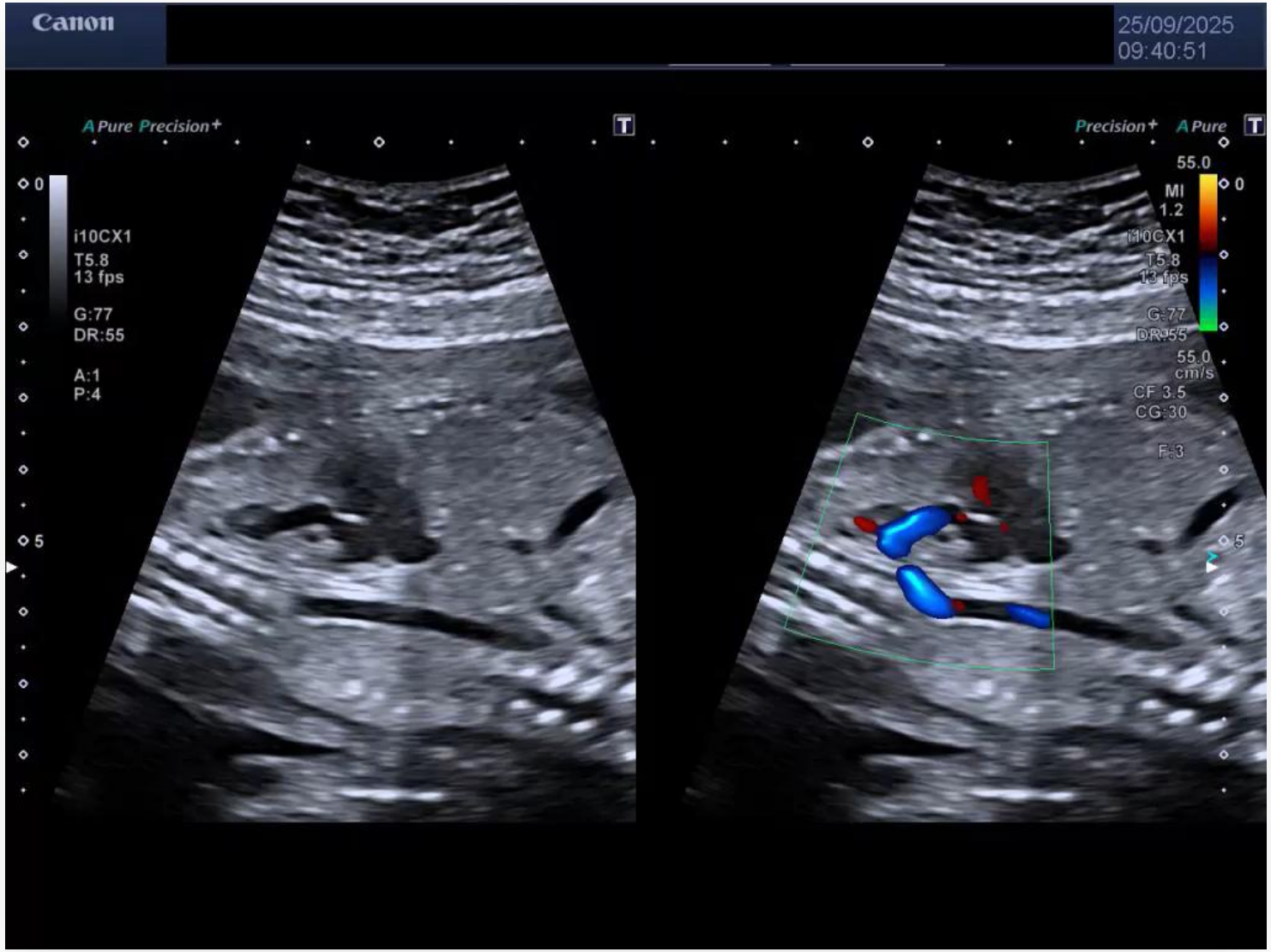


- Sweep to try and find LV
- AAo and LVOT but no obvious LV cavity

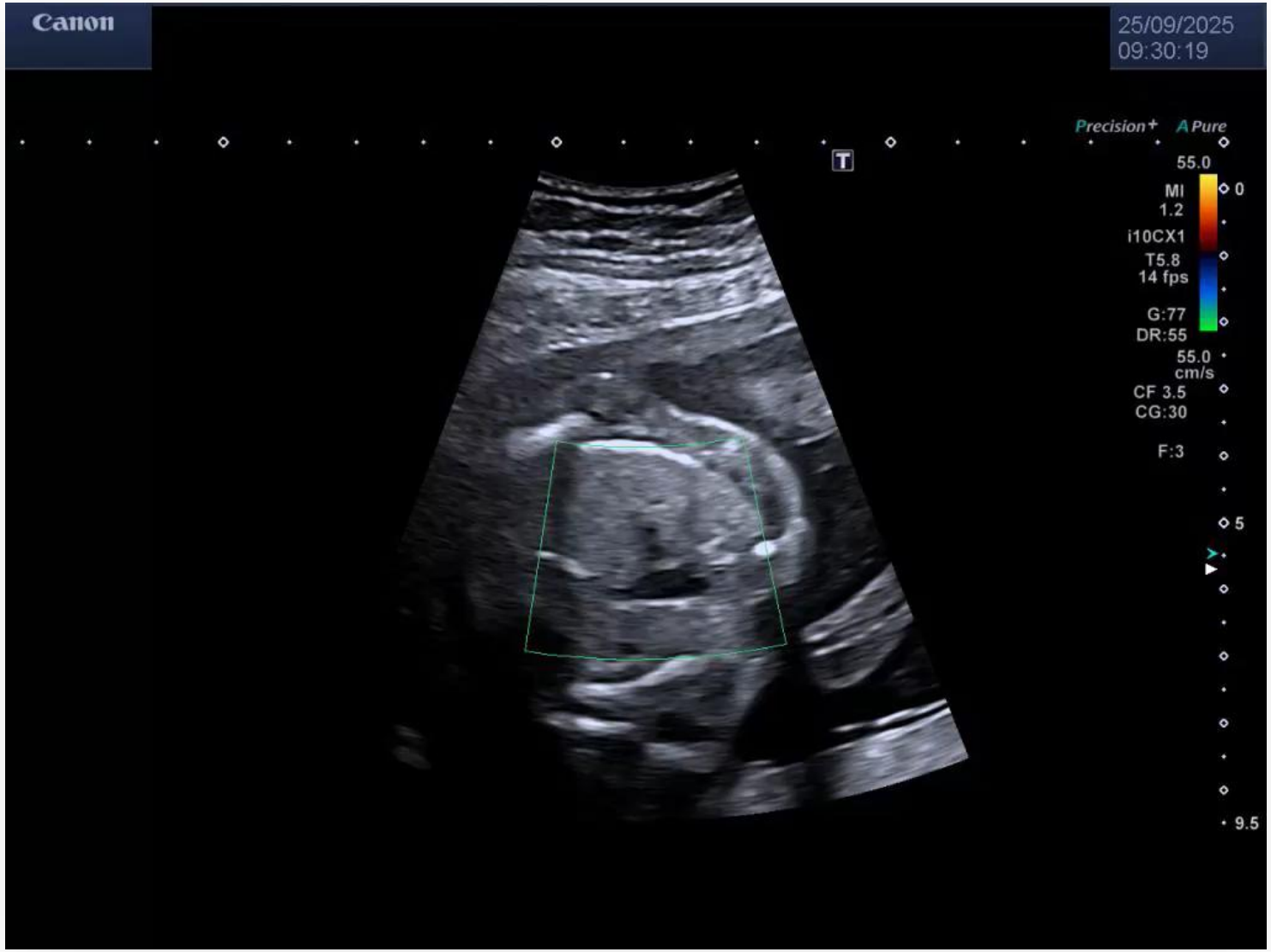
- GA relationship



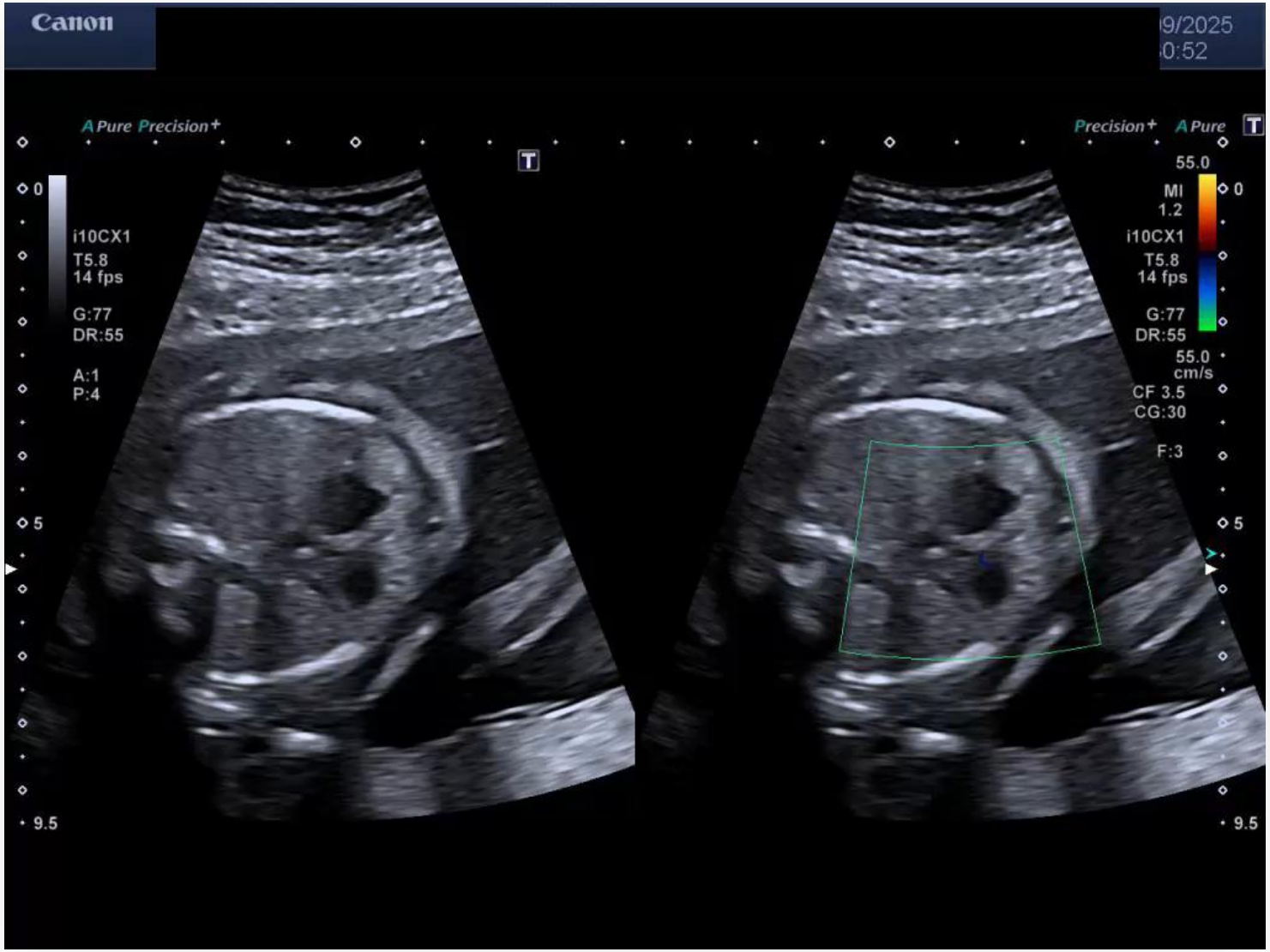
- Sagittal plane true arch antegrade flow
- Sweep to ductal arch



- 3VT
- Small Ao vs PA
- Left arch
- Antegrade flow

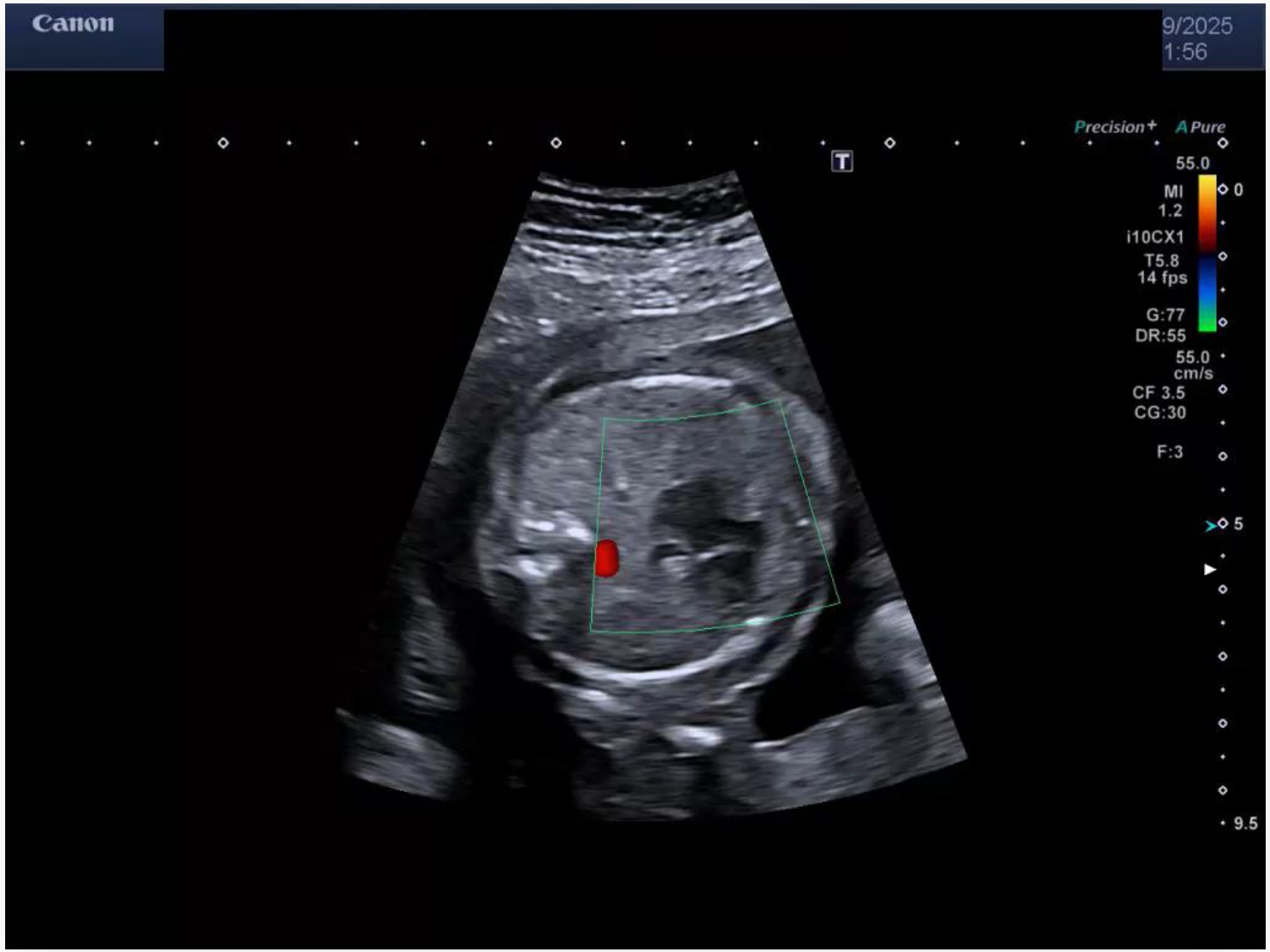


- BPA
- Ductal arch
- Sweep to true arch and AAo

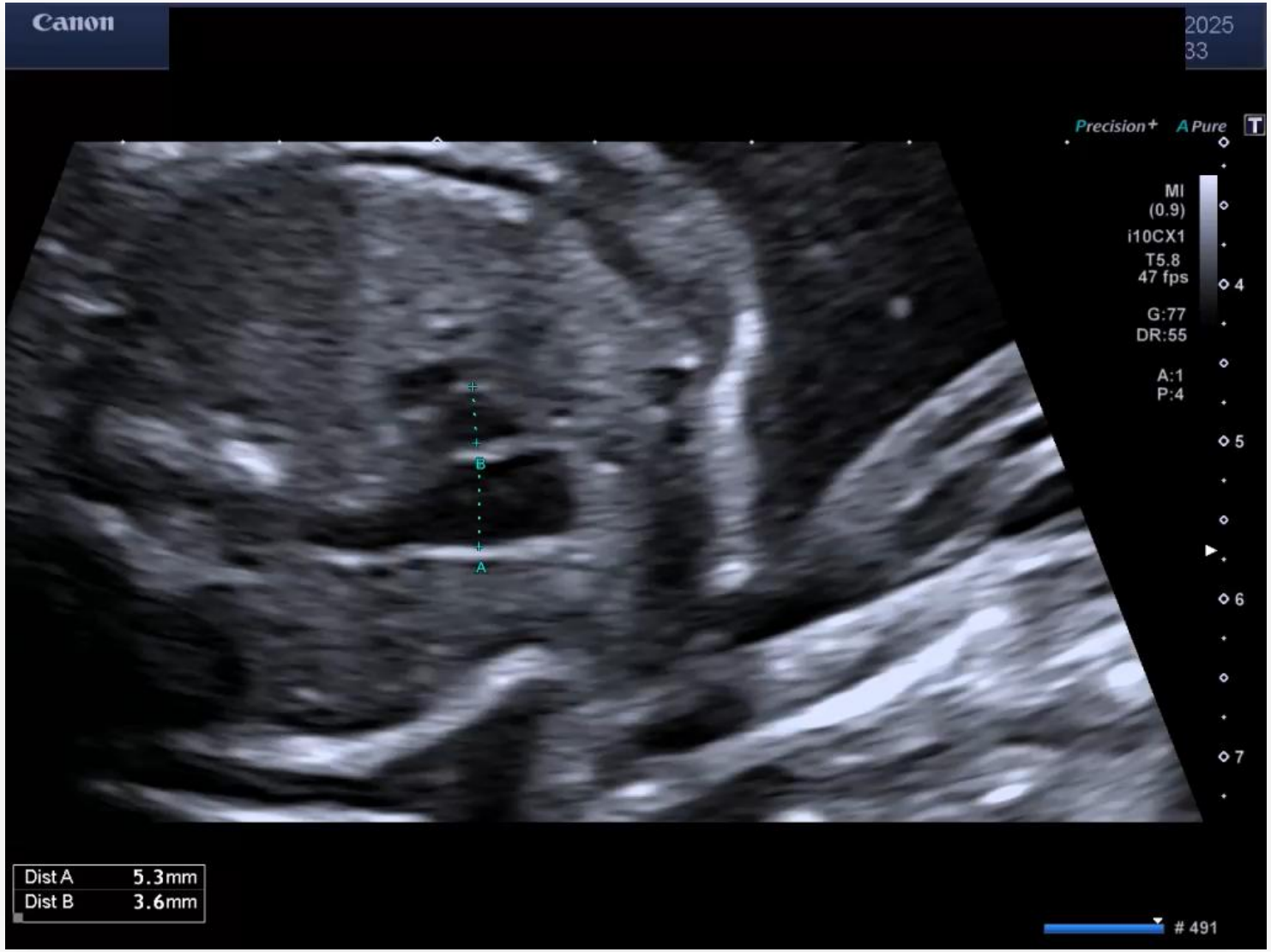


- TV inflow
- No significant TR

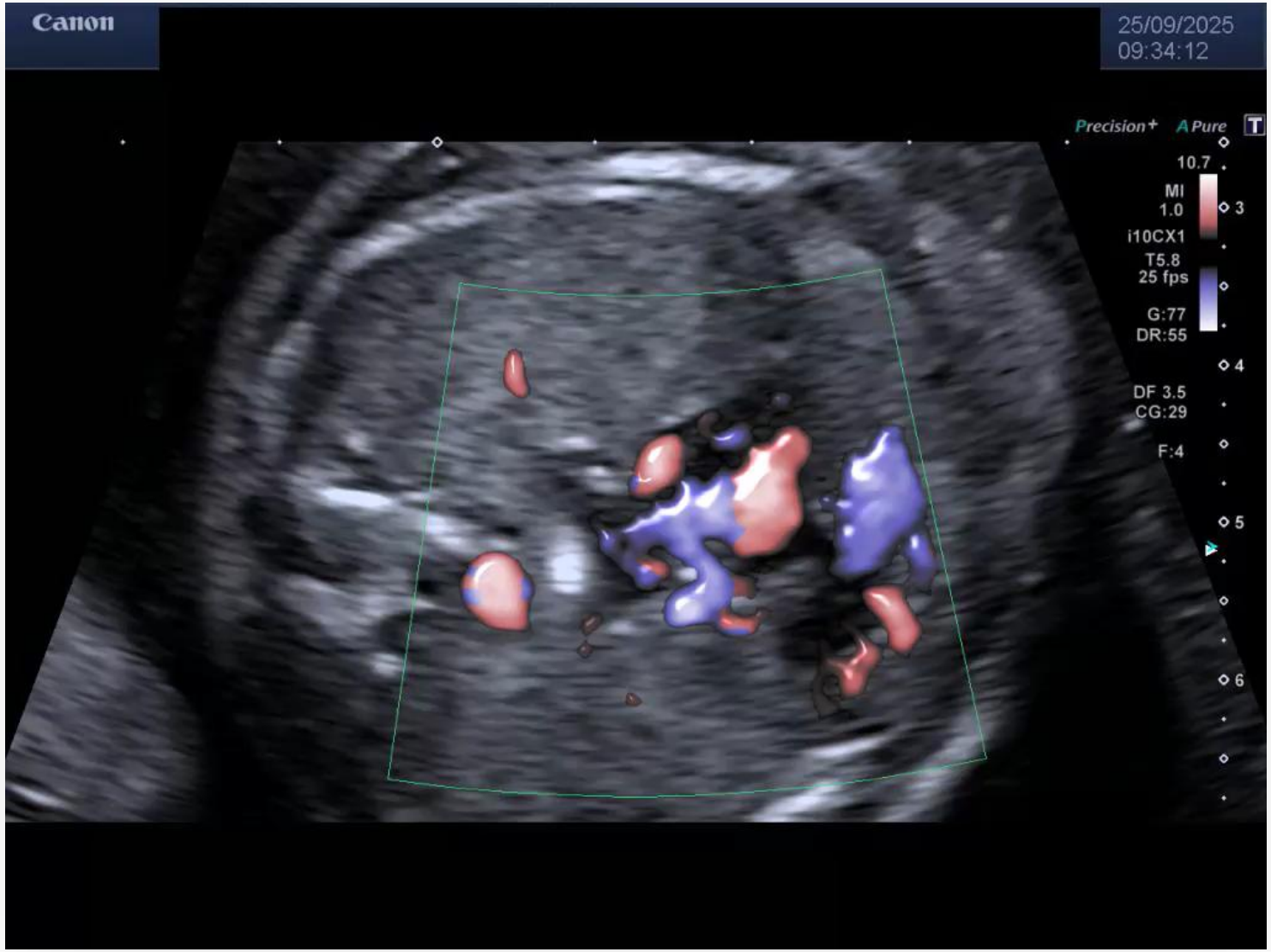
- Red flow of Aao but no obvious LV



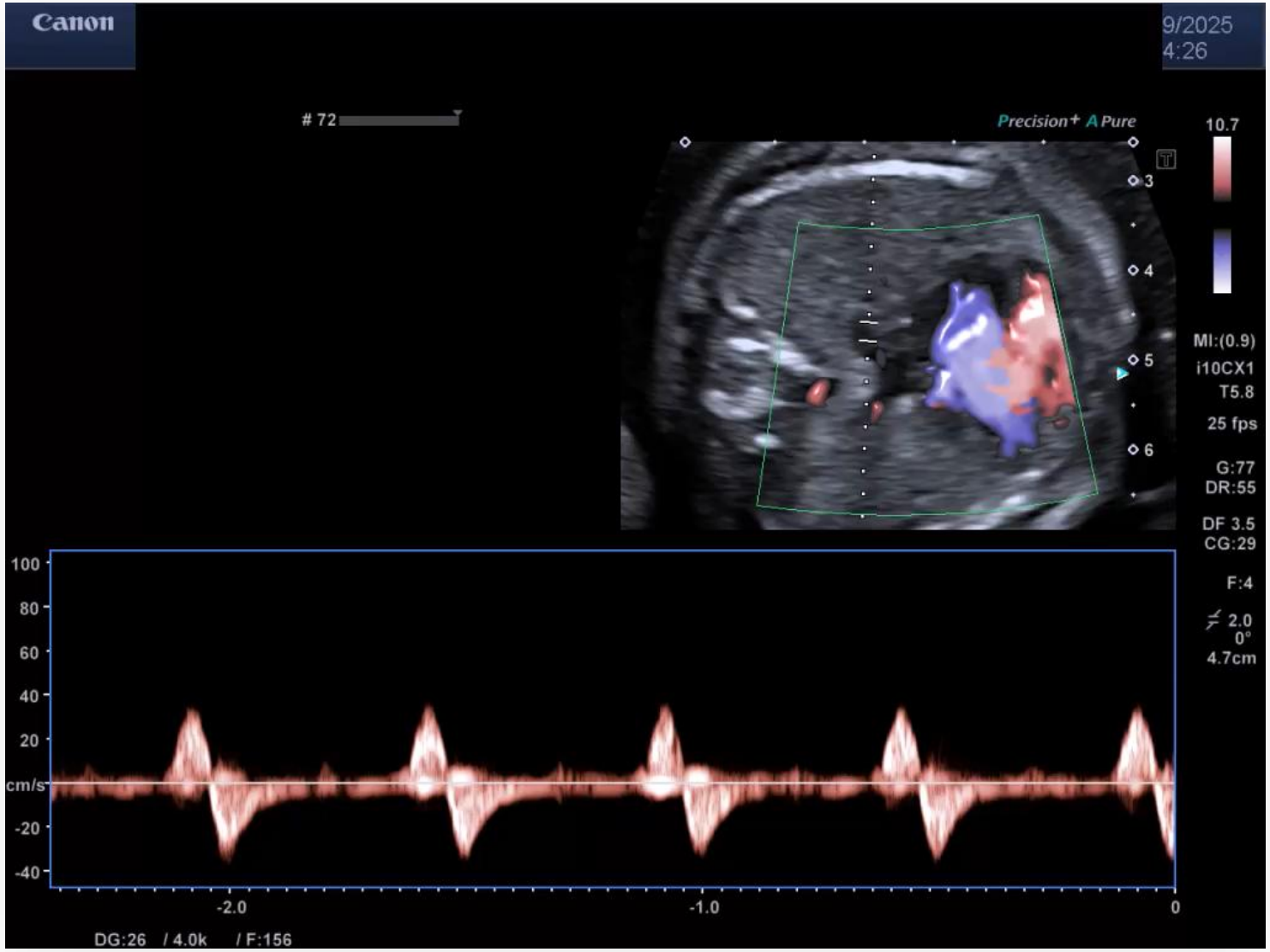
- Size discrepancy of 3VT



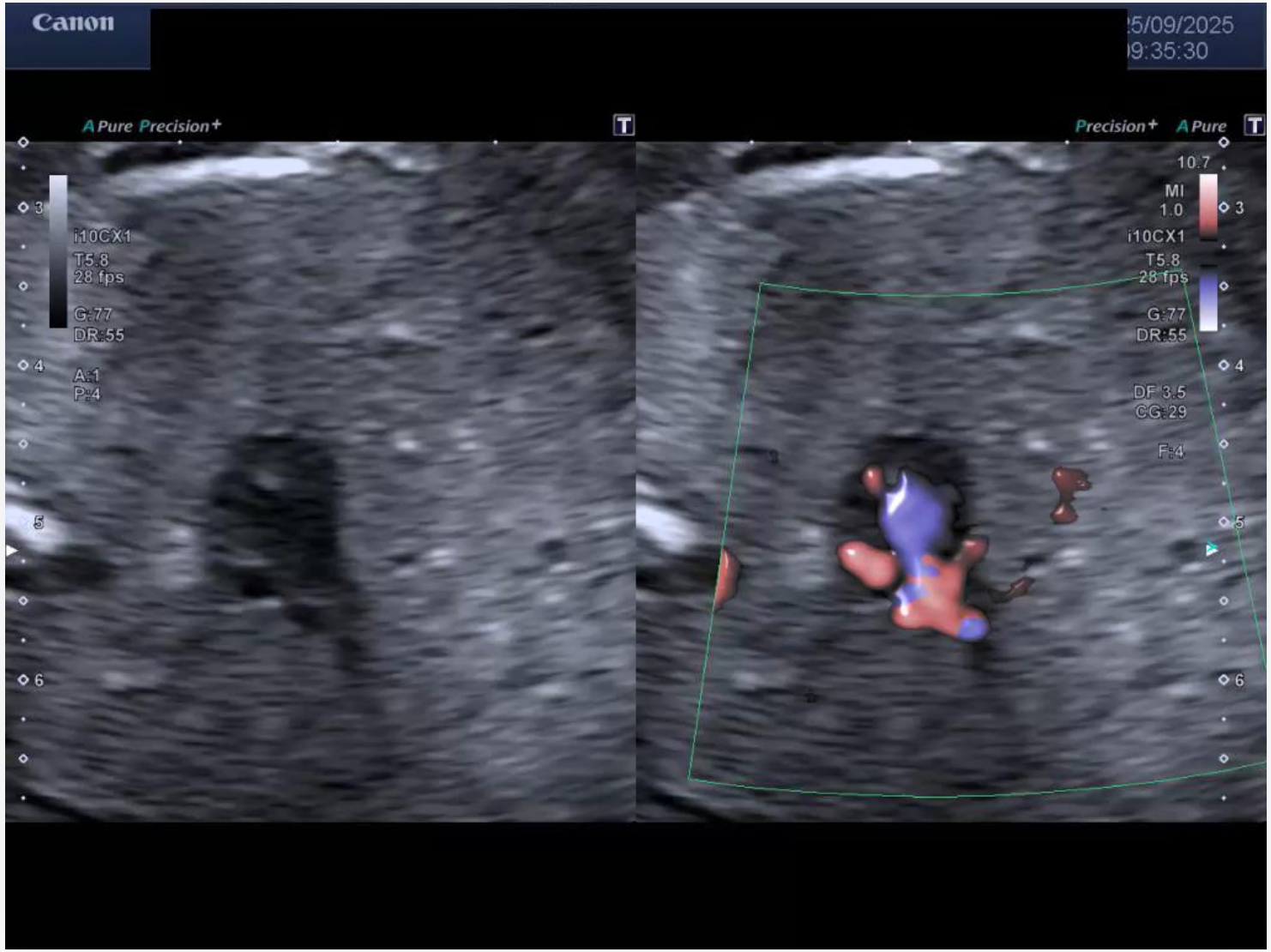
- Pulmonary venous return to LA
- Reversed flow



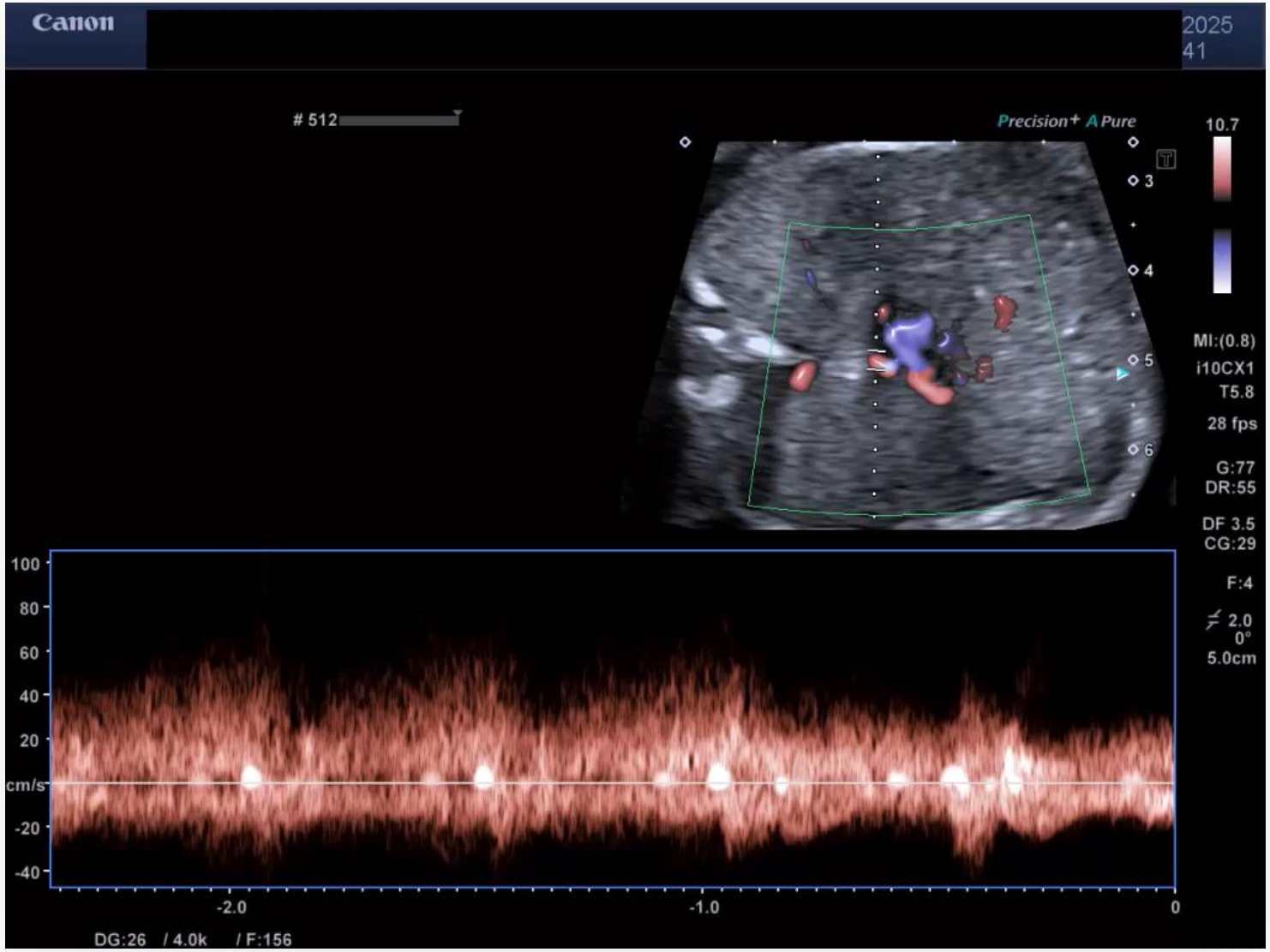
- Doppler of reversed flow
- Restrictive Atrial septum – poor prognostic feature



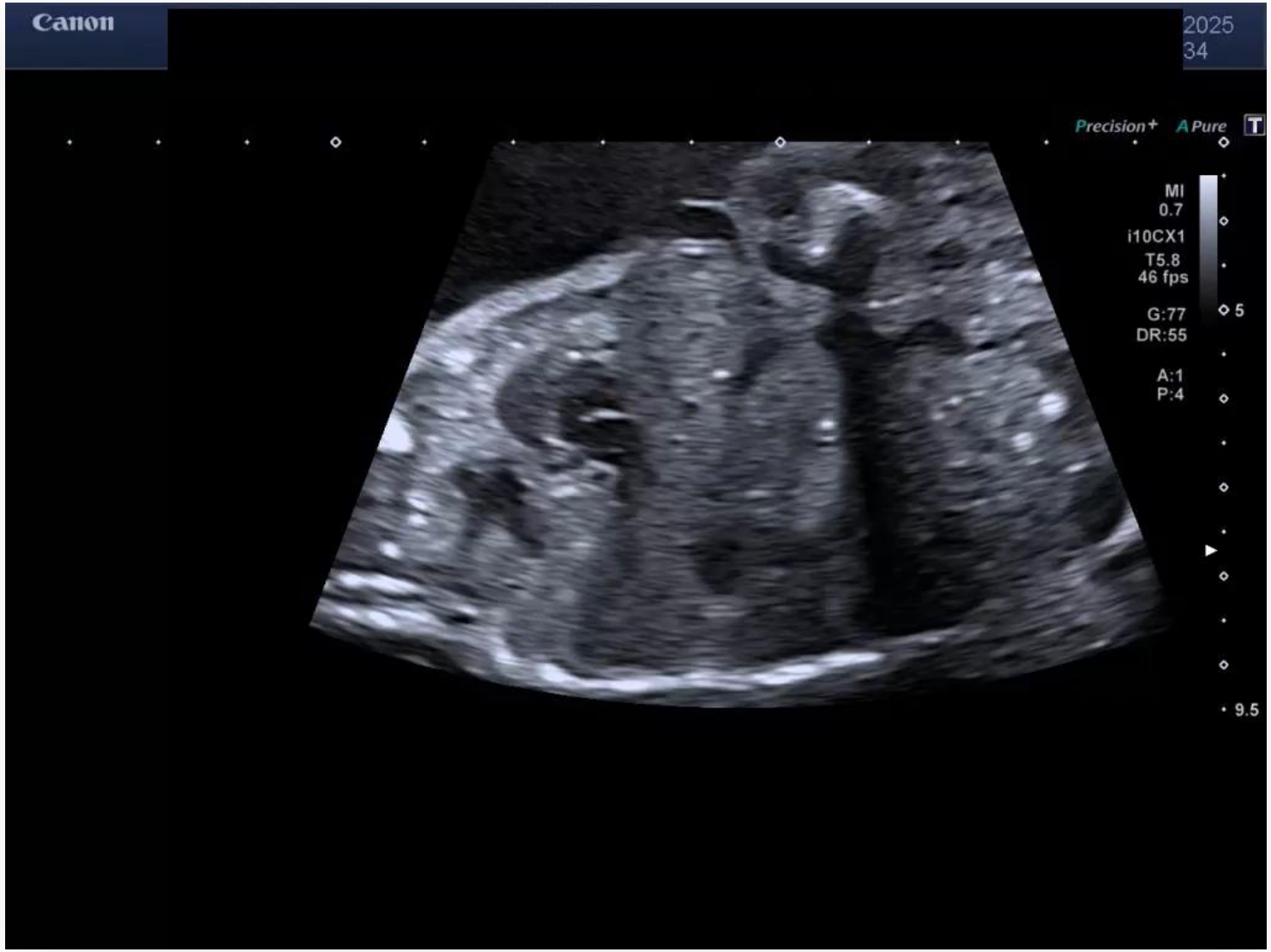
- Small atrial communication



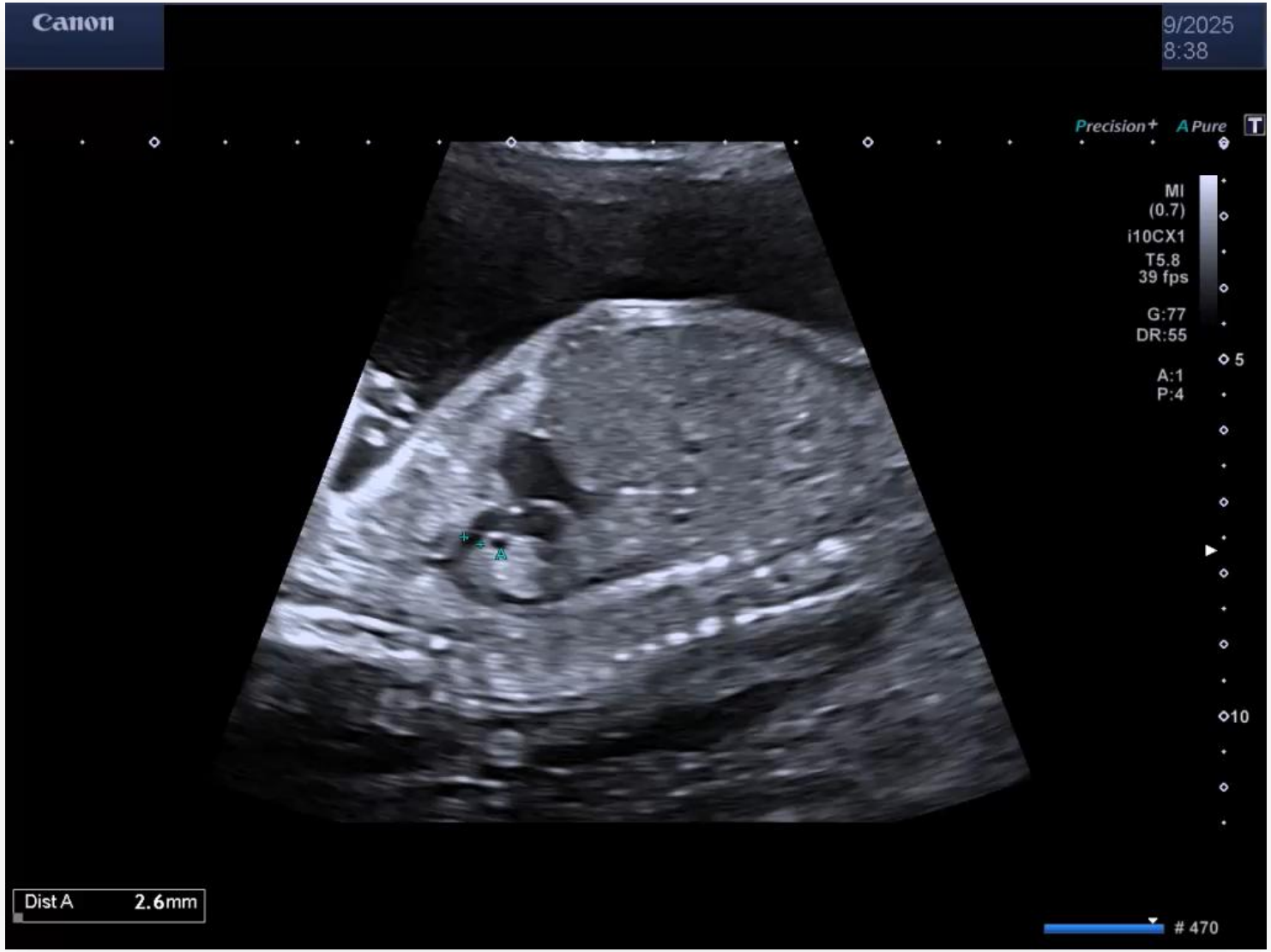
- Continuous trace across IAS



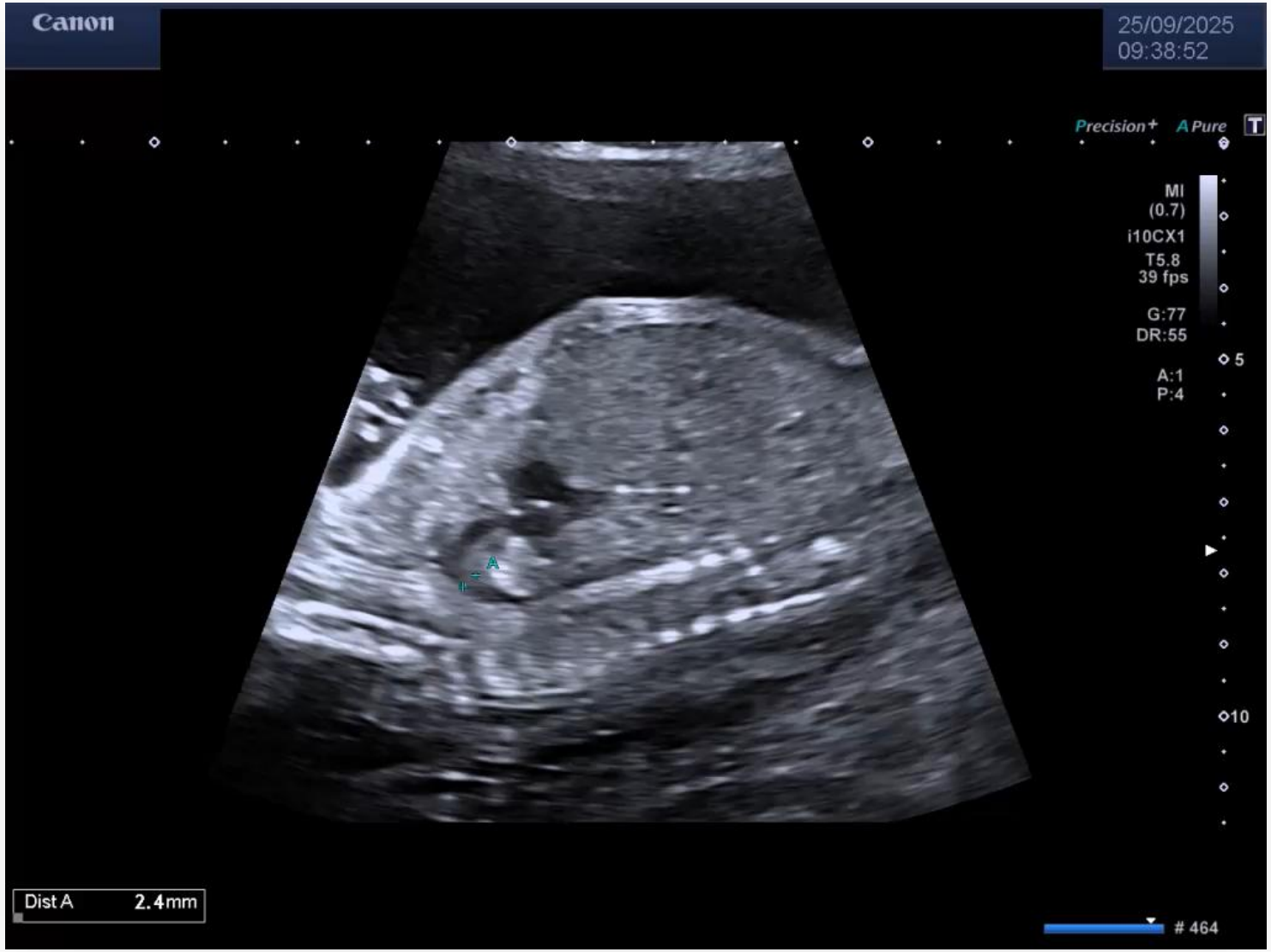
- Sagittal sweep ductal arch

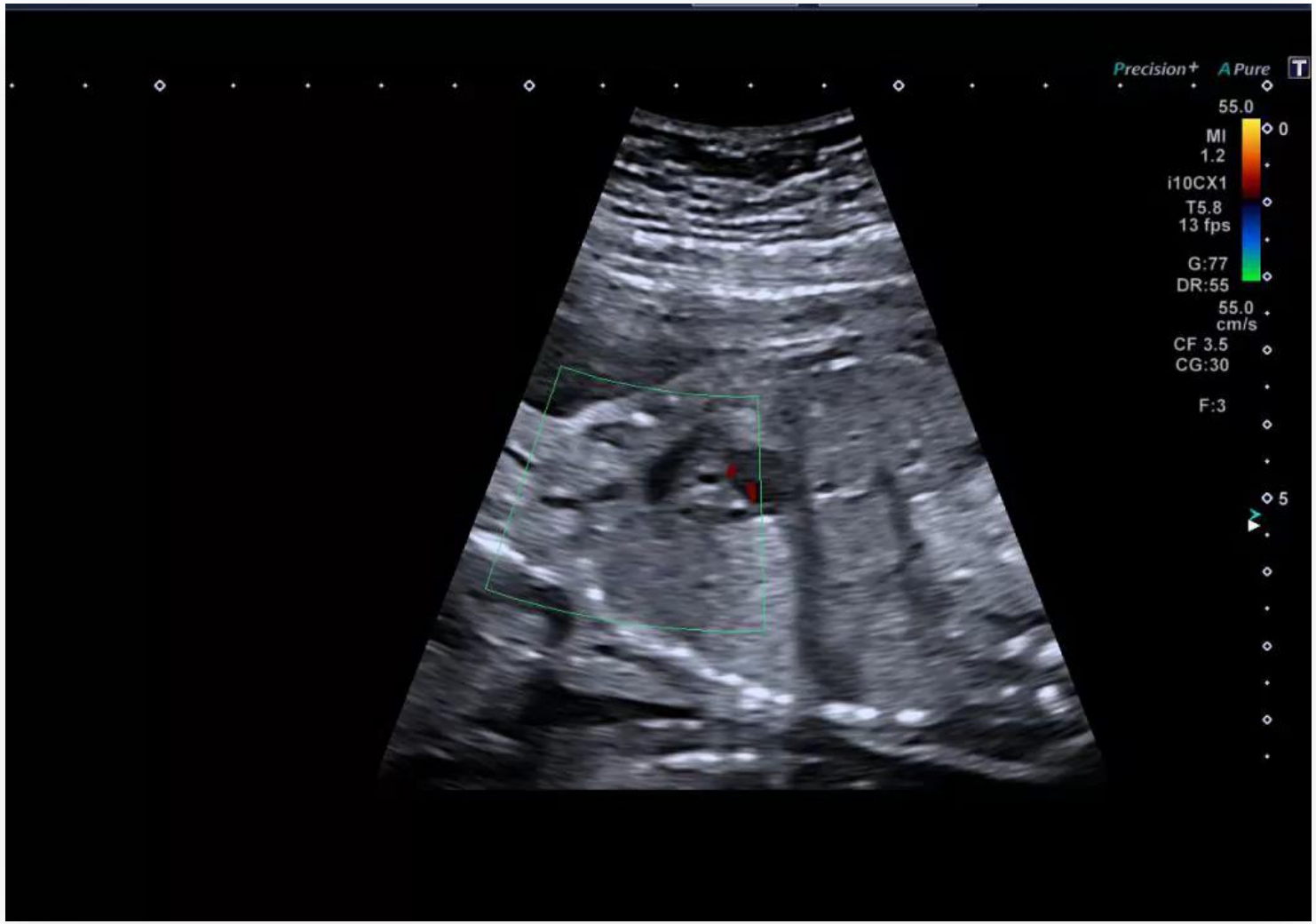


- Measurements on true arch



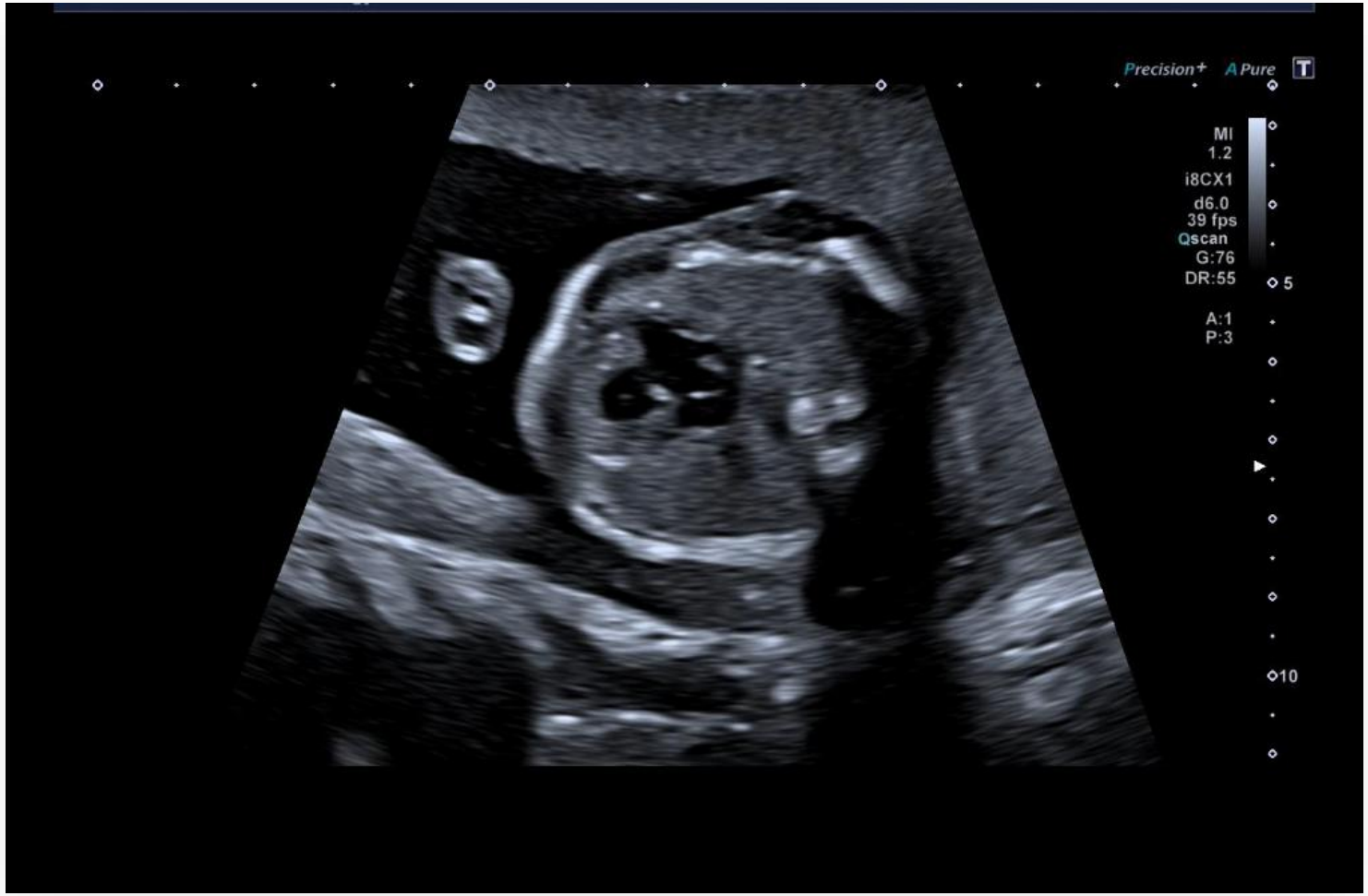
- Measurements extended distally to give picture of hypoplastic arch



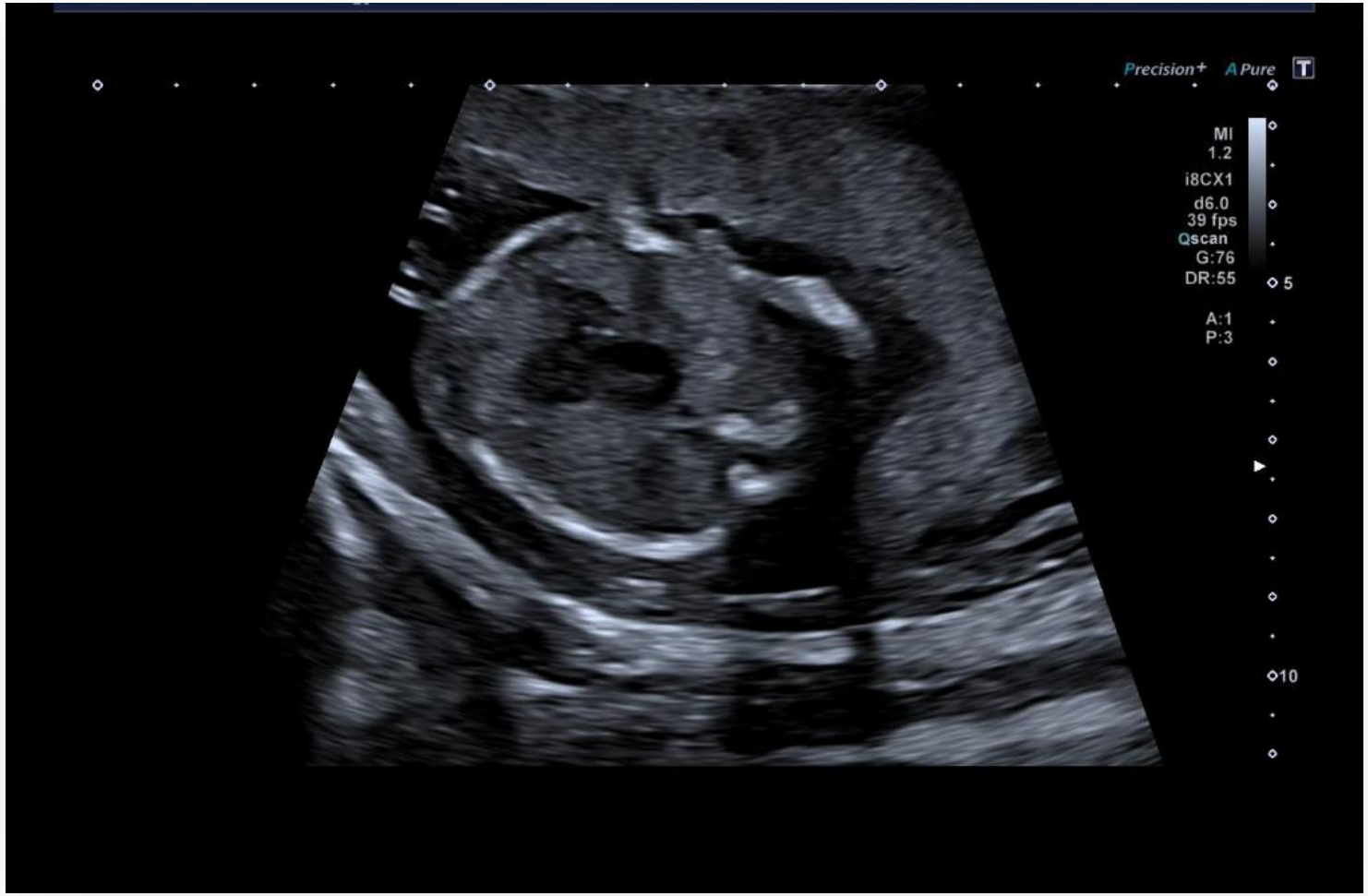

Precision+
A Pure
T
55.0
MI
1.2
i10CX1
T5.8
13 fps
G:77
DR:55
55.0
cm/s
CF 3.5
CG:30
F:3
0
5

Case 2

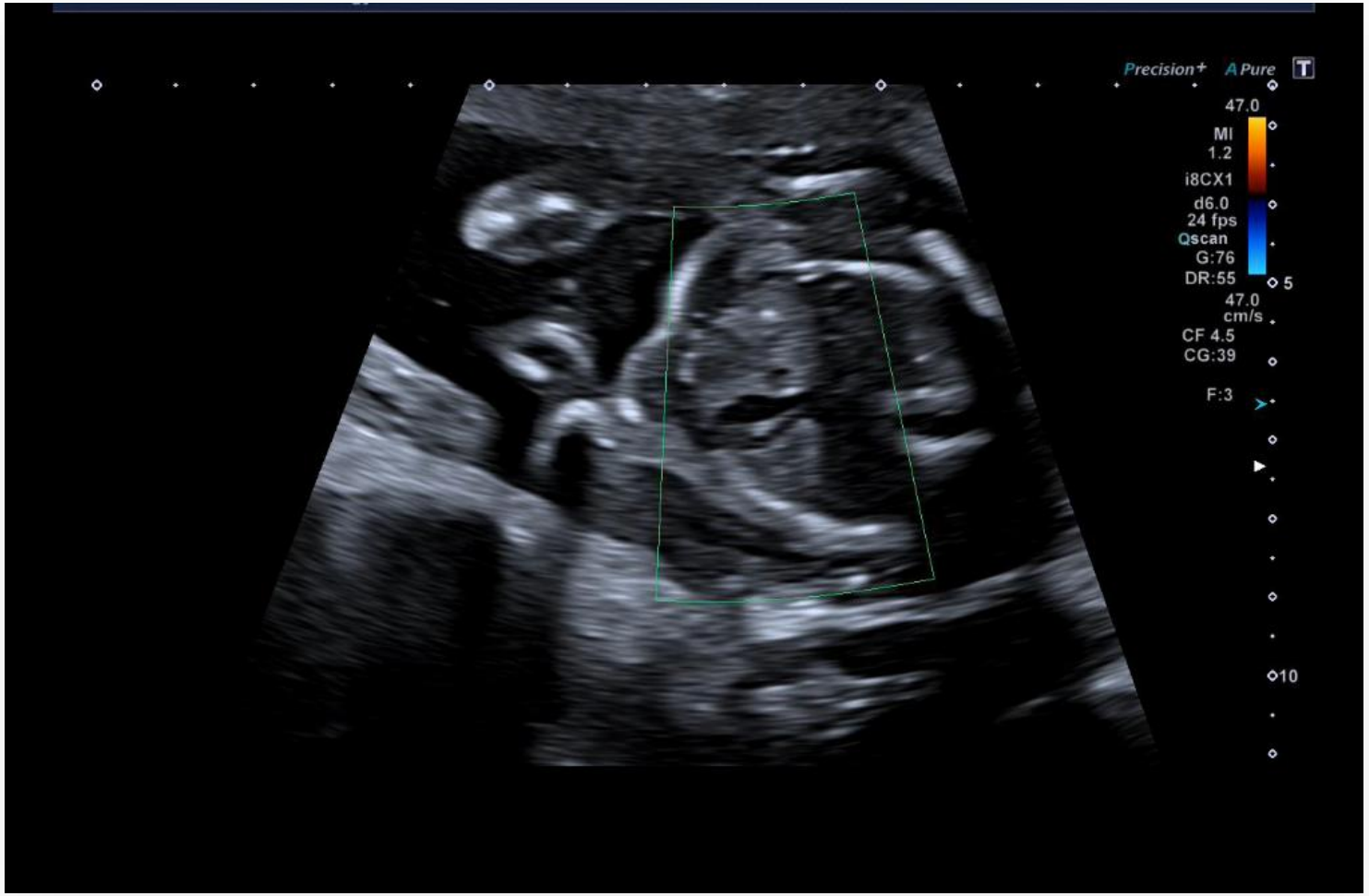
- 4CV
- Small LV cavity seen



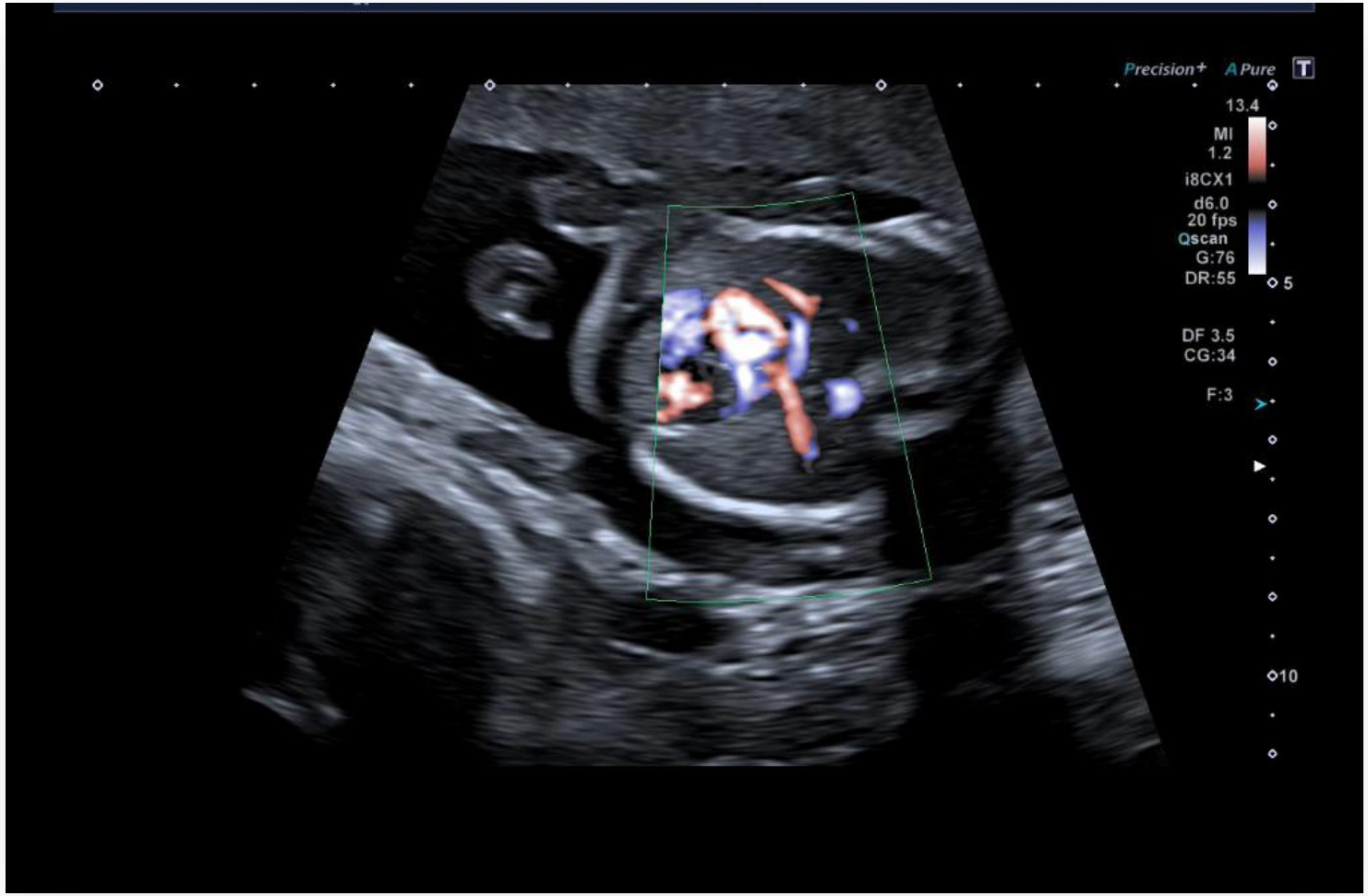
- Sweep to GA
- PA seen but Ao/LVOT not identified



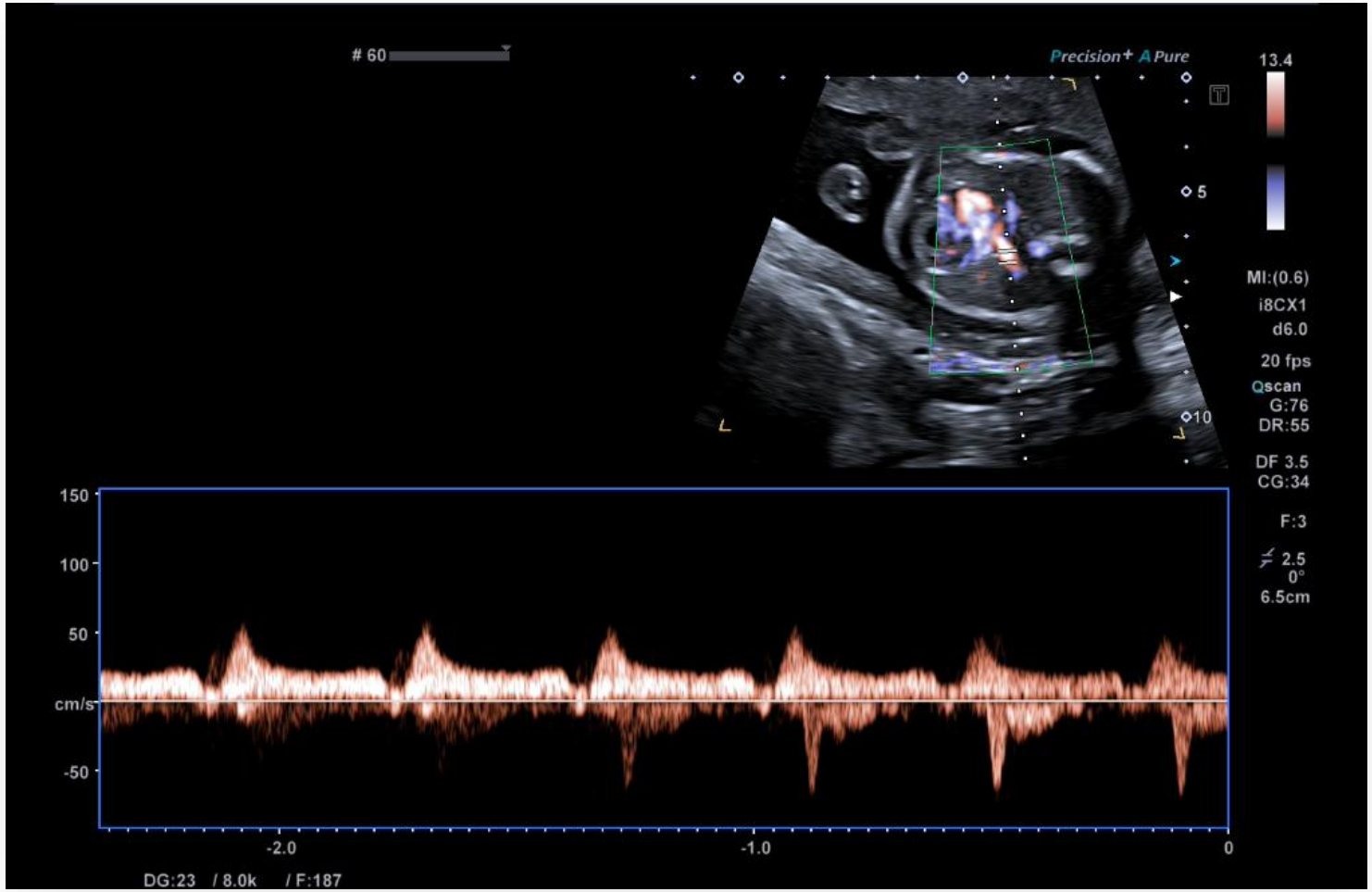
- 3VT CFM
- Retrograde flow in true arch



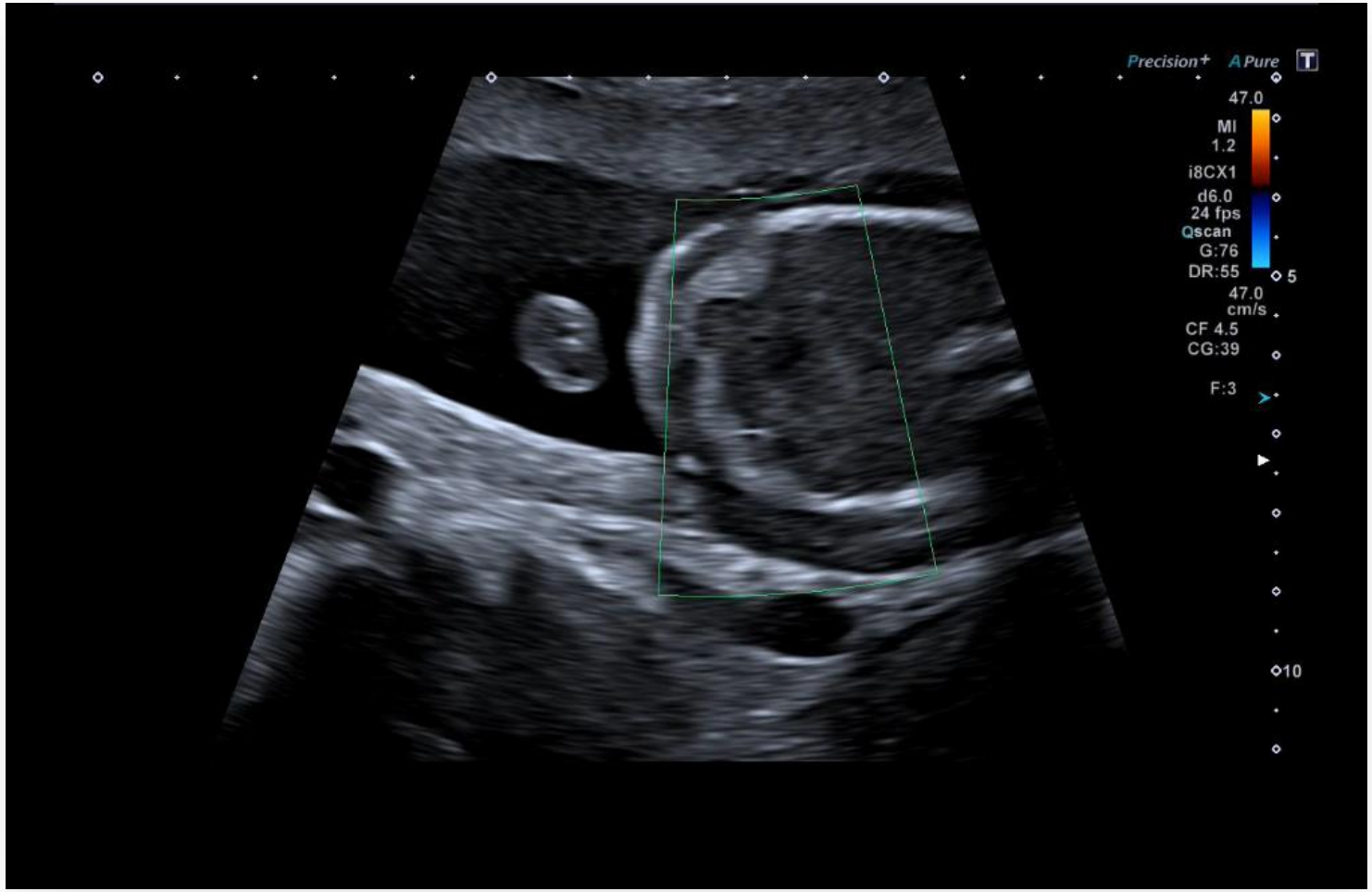
- Pulmonary veins
- Continuous flow to LA



- Normal Pulmonary venous pattern - no reversal

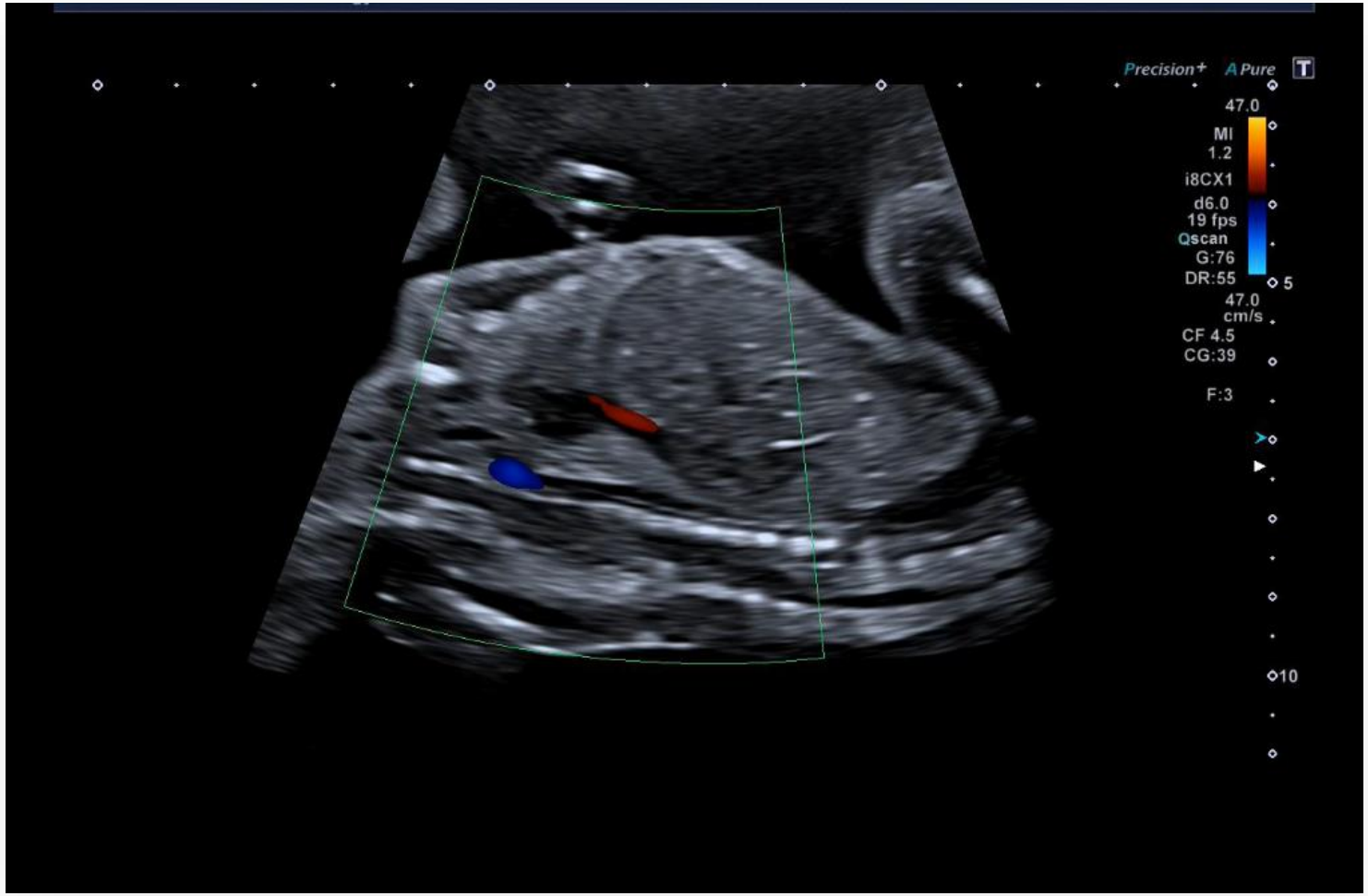


- No TR
- IAS widely patent



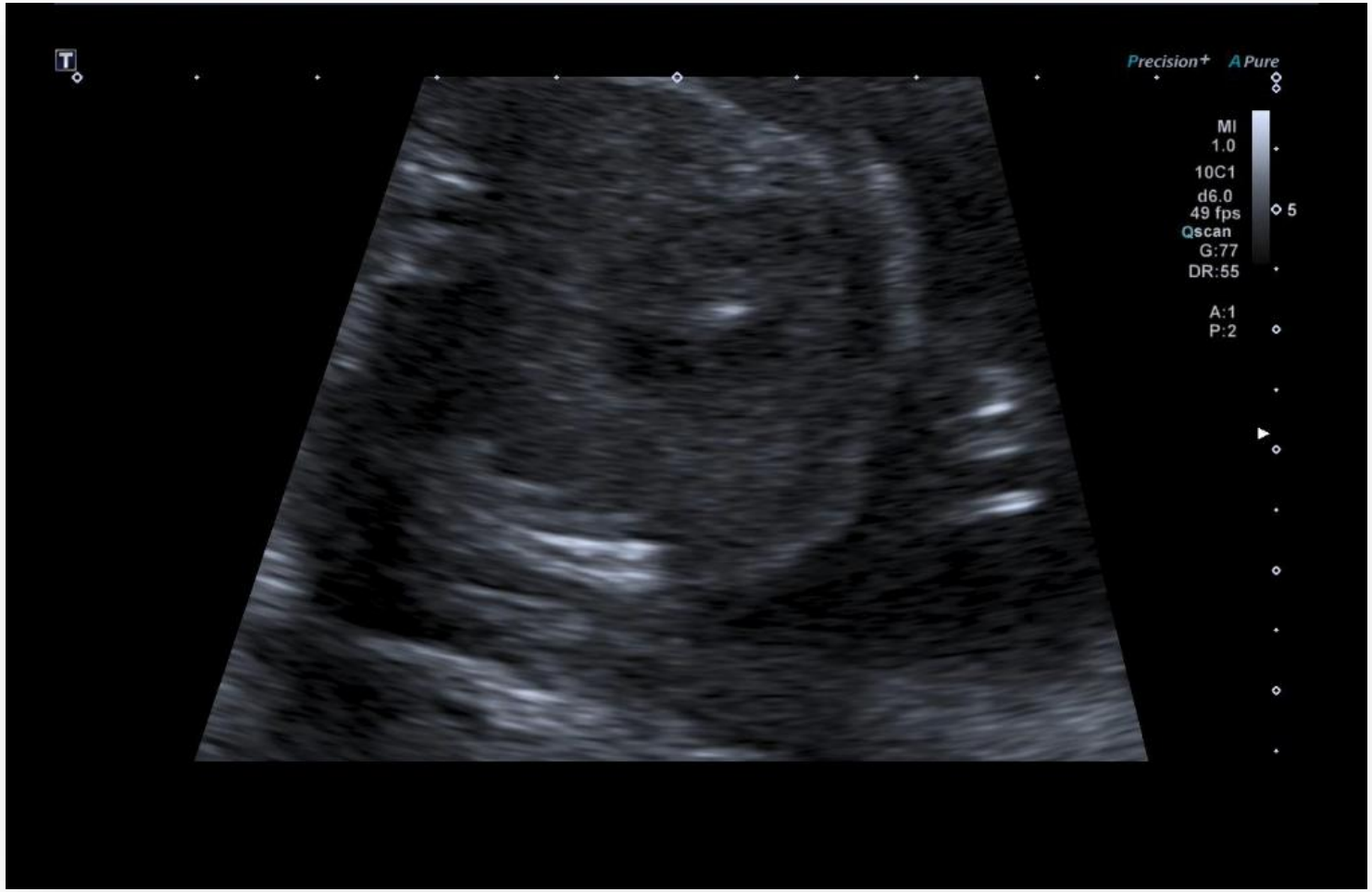
- Sagital sweep
- IVC SVC to RA

- Antegrade Ductal arch
- Retrograde true arch

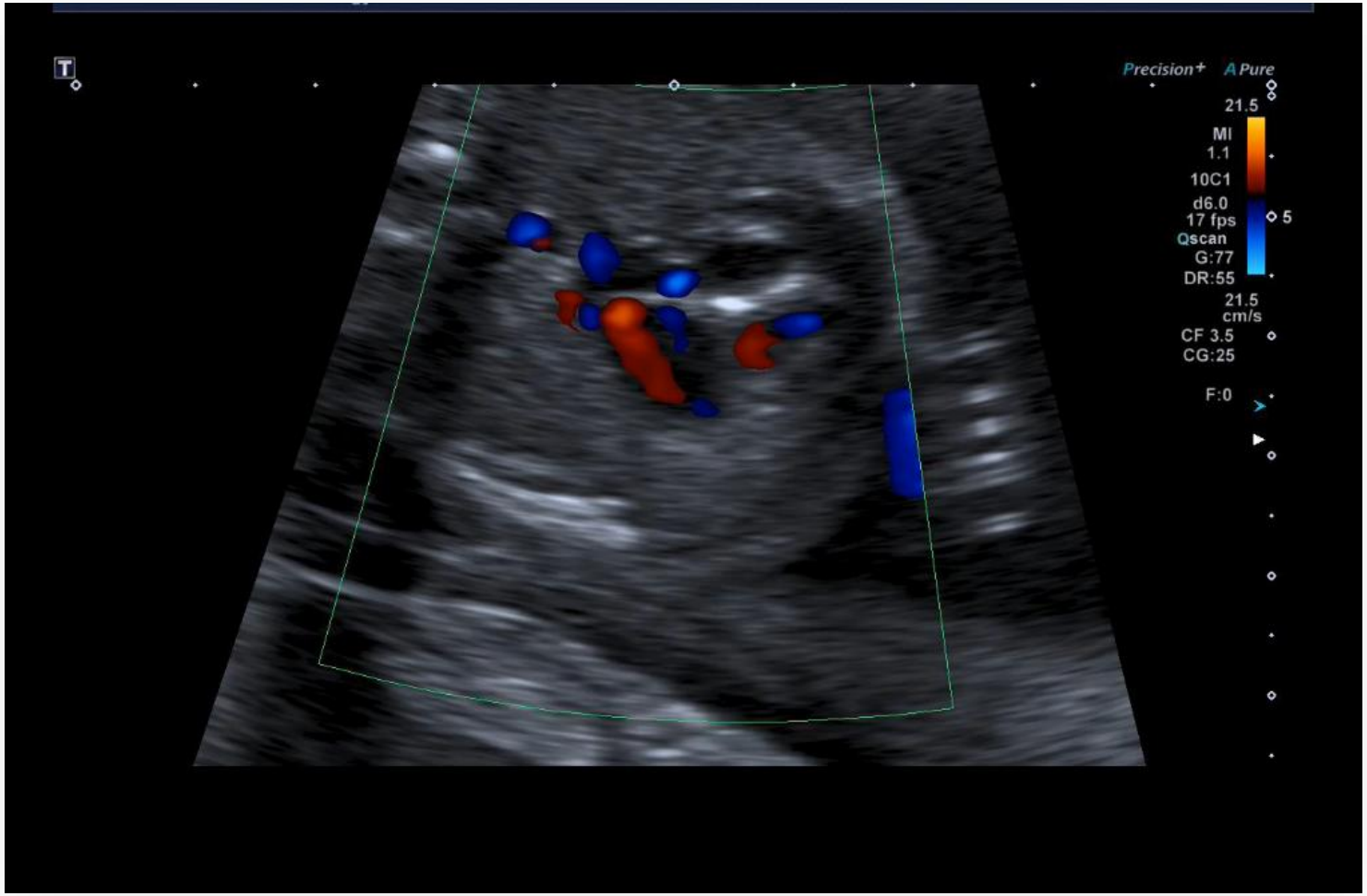


Case 3

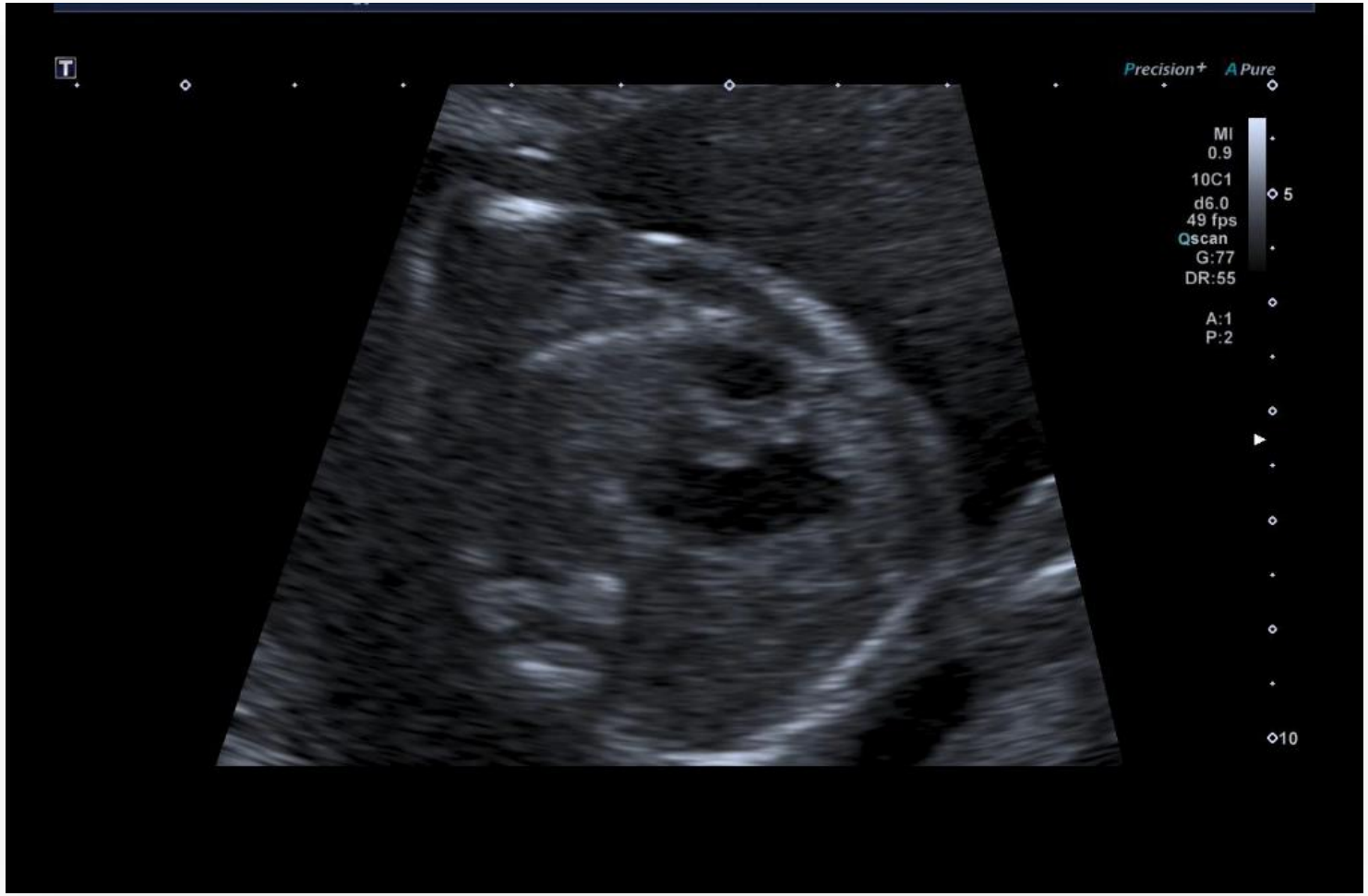
- Endocardial fibroelastosis EFE



- No CFM ‘fill’ in LV cavity
- Possible MS MR



- RV forms apex
- LV does not



INCREASED NUCHAL THICKNESS .v. CYSTIC HYGROMA

DR. MARK DENBOW – Consultant in Obstetrics and Fetal Medicine, UHW.

GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

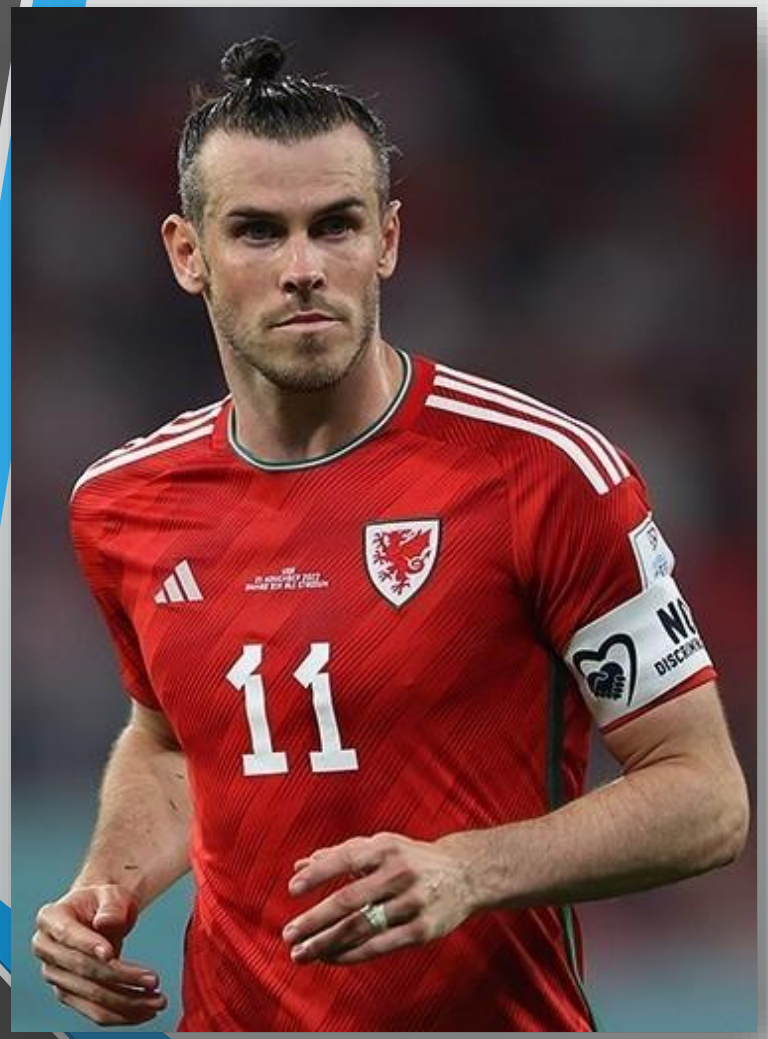
Are they the same?

VS

Are they the same?

VS

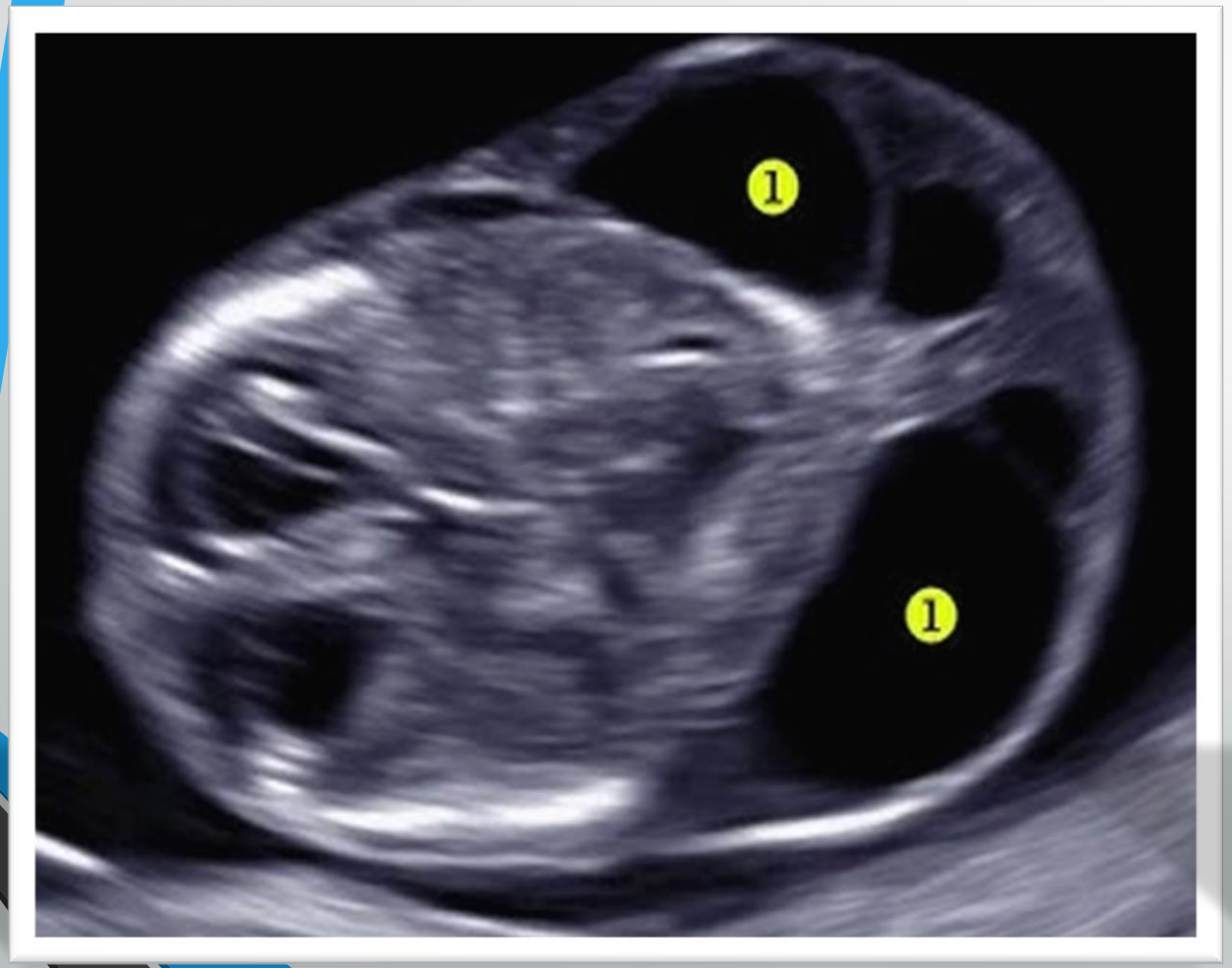
Are they the same?



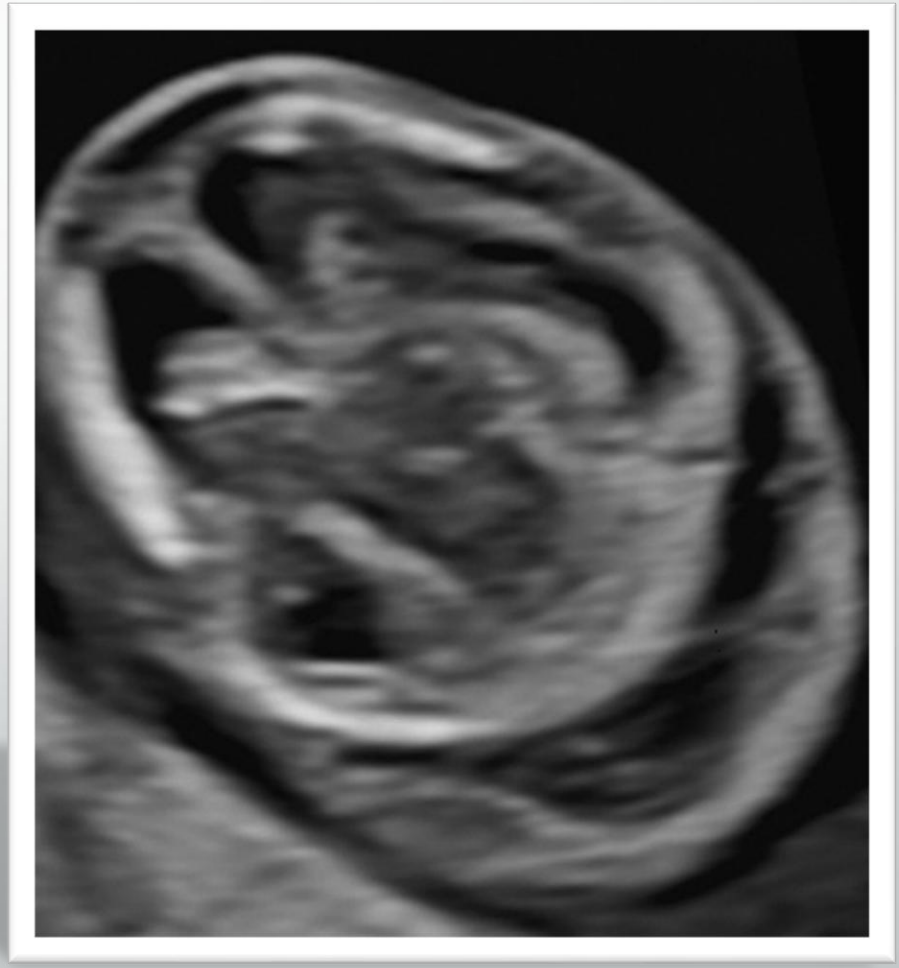
VS

They are not the same

Cystic Hygroma

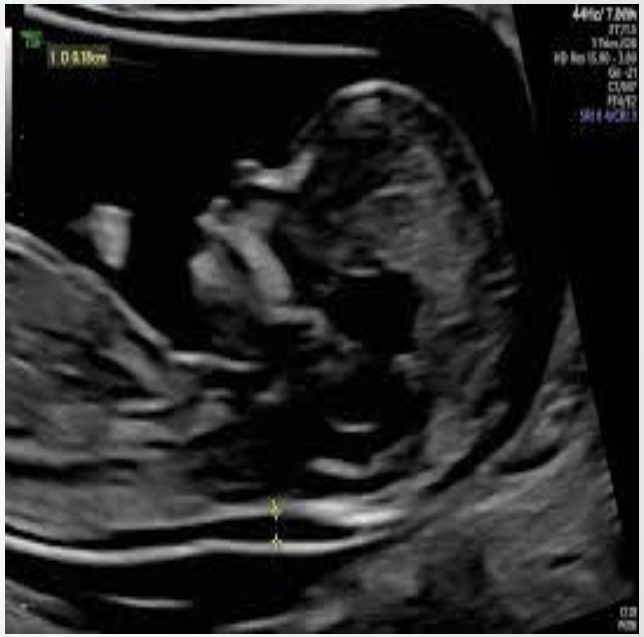


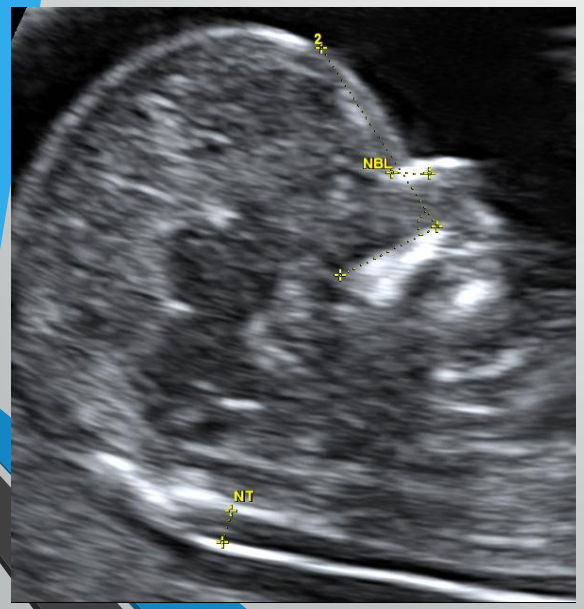
Increased NT

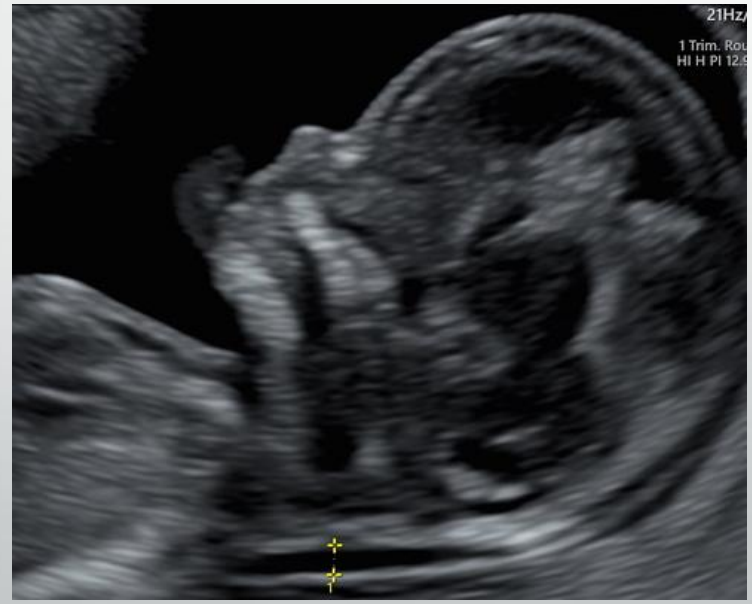


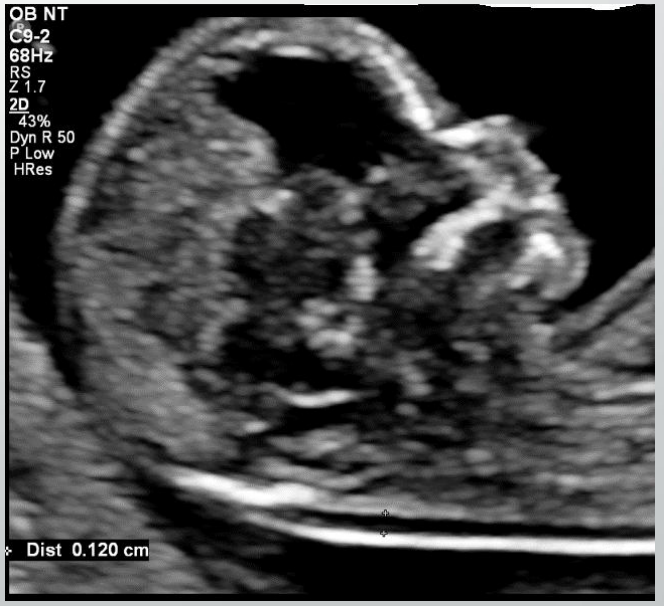
Nuchal Translucency

- Mesenchymal oedema caused by
  - In T21: abnormal collagen in dermis
  - Turner Syndrome: lymphatic dysplasia
  - Congenital heart disease: abnormal endothelium



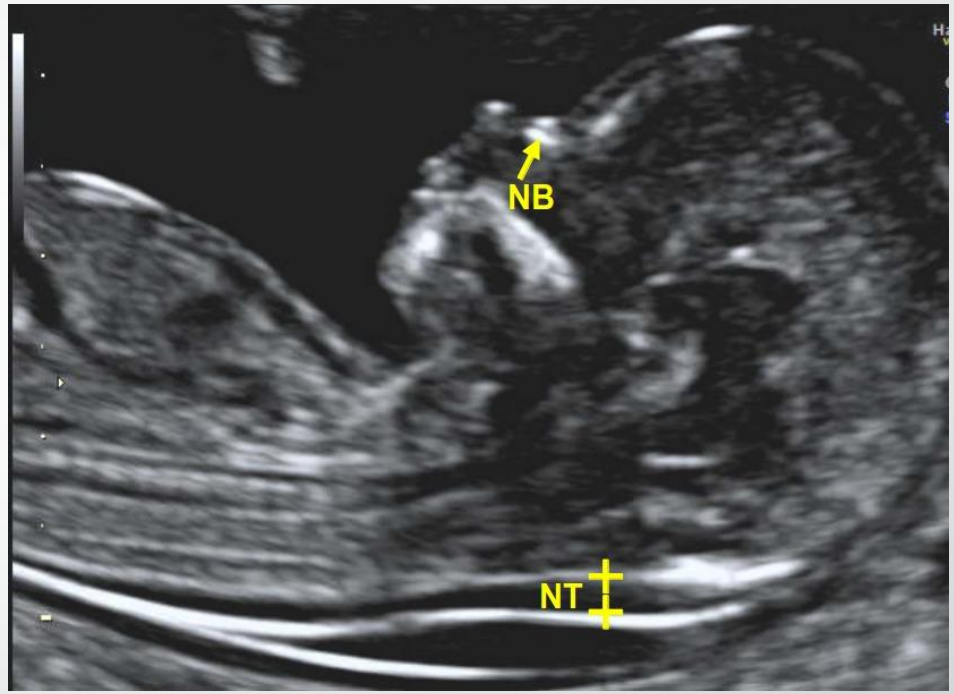






Nuchal Translucency: Ultrasound

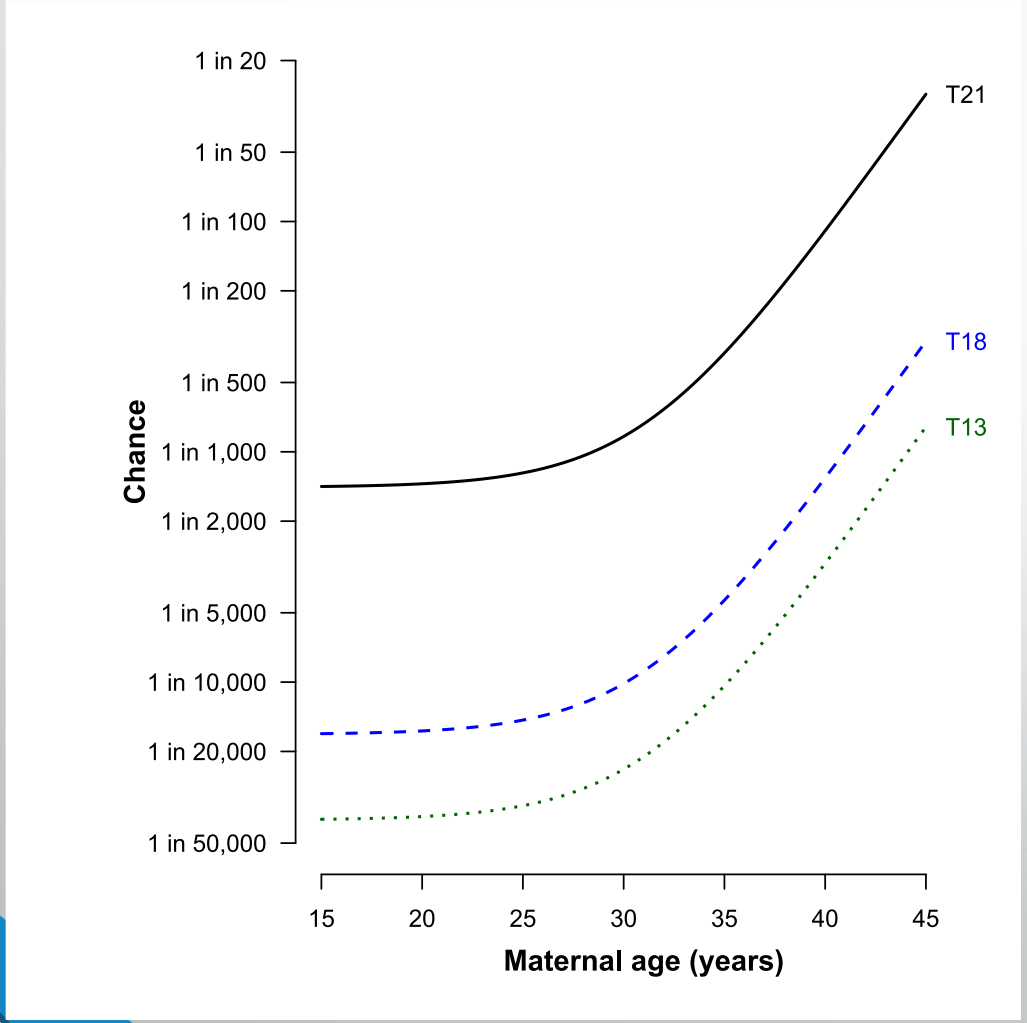
- **11 - 13+6 weeks and CRL 45 to 84mm.**
- Magnify till fetal head and thorax occupy the whole screen.
- **Mid-sagittal view of the face with:**
  - echogenic tip of the nose
  - rectangular shape of the palate anteriorly
  - the translucent diencephalon centrally
  - nuchal membrane posteriorly.
- **Neutral position:**
  - Neck hyperextended falsely increased
  - neck is flexed falsely decreased.

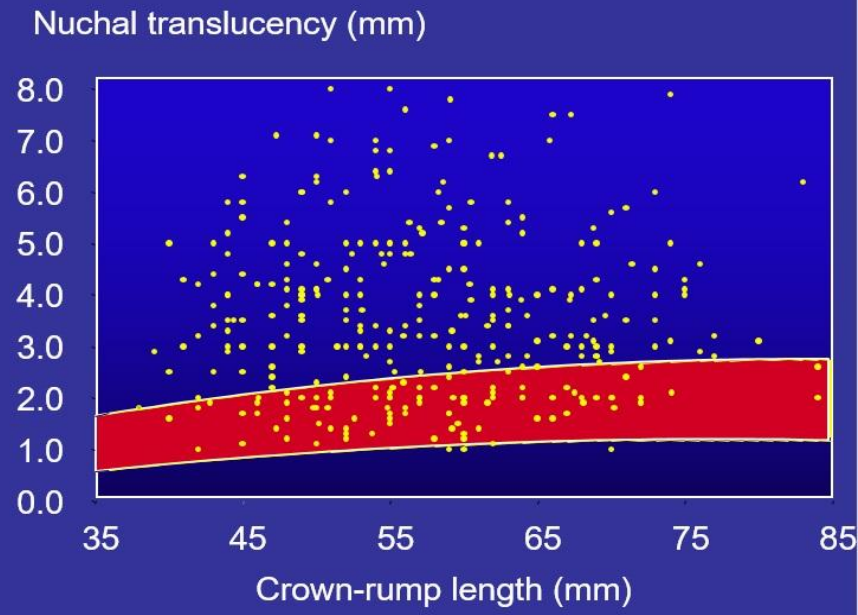


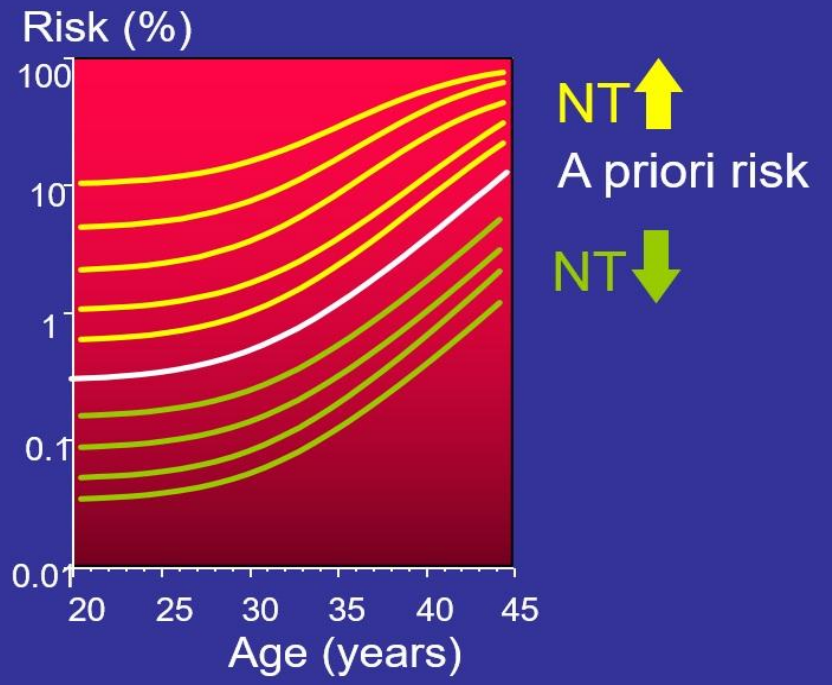
- **Distinguish between fetal skin and amnion.**
- The widest part of translucency must **always** be measured.
- Measurements from inner border of the horizontal line of the calipers placed ON the line that defines the nuchal translucency thickness - the crossbar of the caliper should be such that it is hardly visible as it merges with the white line of the border, not in the nuchal fluid.
- The umbilical cord around the fetal neck: measurements of NT above and below the cord , use the average of the two measurements.

Nuchal Translucency: Results

- NT used for First Trimester Screening





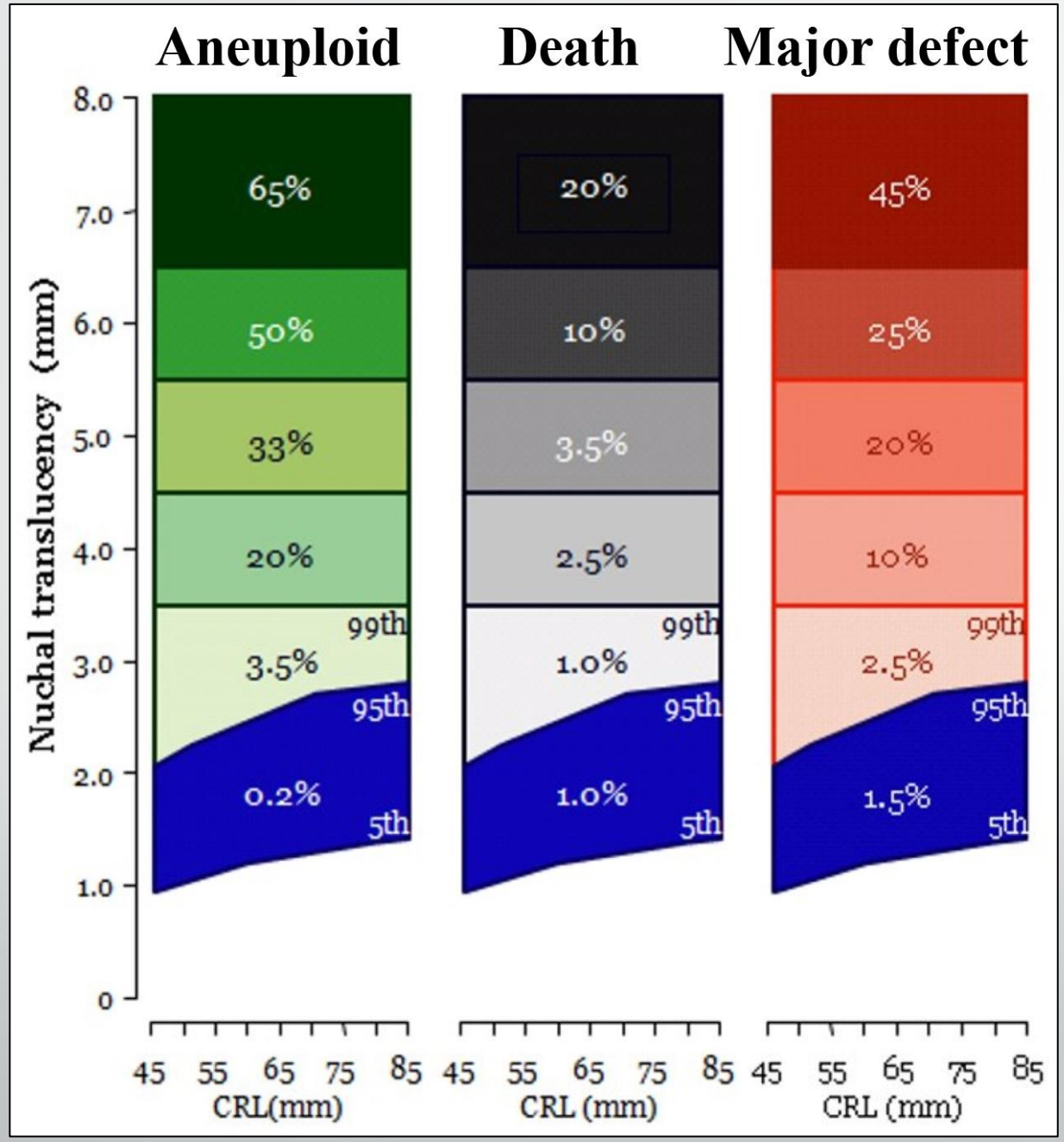


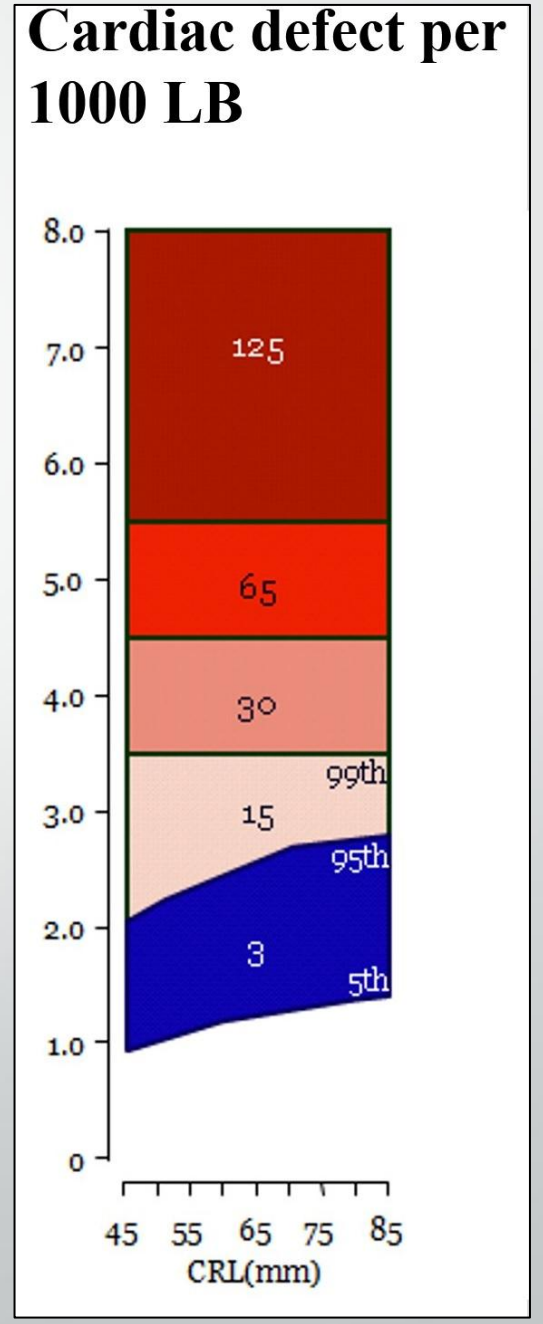
Nuchal Translucency: Increased

- **If NT increased (≥3.5mm):**
  - Offer referral to FMU
  - Still send FTS if consented

Nuchal translucency	Chromosomal defects	Fetal death	Major fetal abnormalities	Alive and well
<95th centile	0.2%	1.3%	1.6%	97%
95th-99th centiles	3.7%	1.3%	2.5%	93%
3.5-4.4 mm	21.1%	2.7%	10.0%	70%
4.5-5.4 mm	33.3%	3.4%	18.5%	50%
5.5-6.4 mm	50.5%	10.1%	24.2%	30%
>6.5 mm	64.5%	19.0%	46.2%	15%

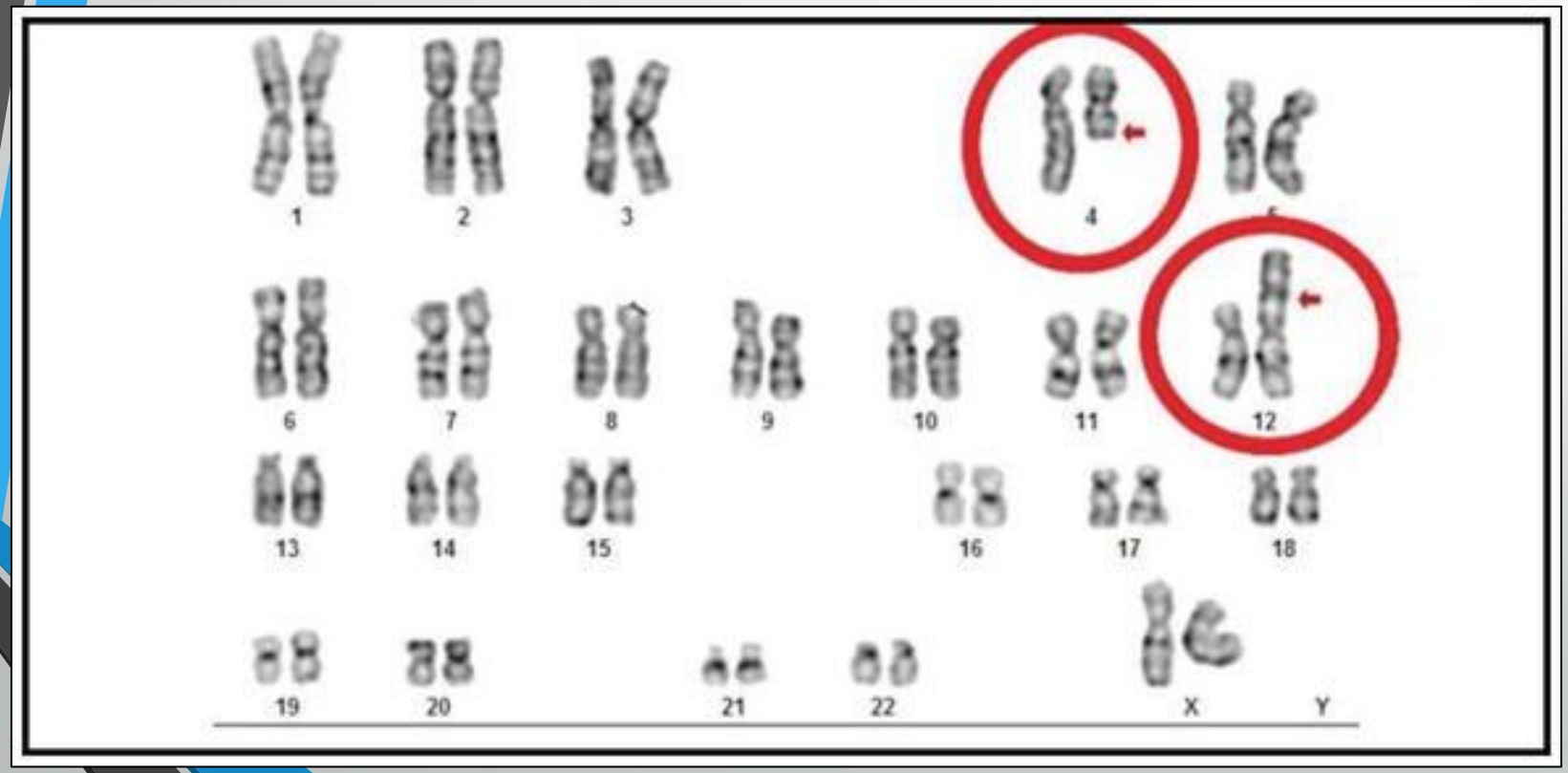
NT:

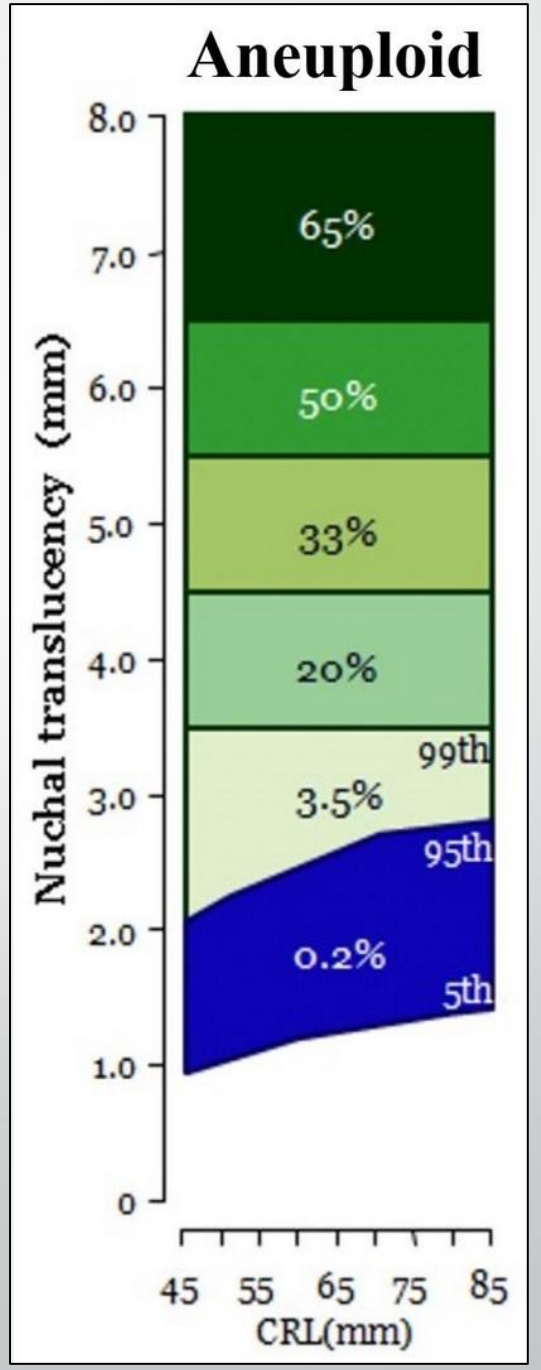

Aneuploid
Death
Major defect
Nuchal translucency (mm)
8.0
7.0
6.0
5.0
4.0
3.0
2.0
1.0
0
65%
50%
33%
20%
99th
3.5%
95th
0.2%
5th
20%
10%
3.5%
2.5%
99th
1.0%
95th
1.0%
5th
45%
25%
20%
10%
99th
2.5%
95th
1.5%
5th
45 55 65 75 85
CRL(mm)
CRL (mm)
CRL (mm)


Cardiac defect per 1000 LB
8.0
7.0
6.0
5.0
4.0
3.0
2.0
1.0
0
125
65
30
99th
15
95th
3
5th
45 55 65 75 85
CRL(mm)

Increased NT:

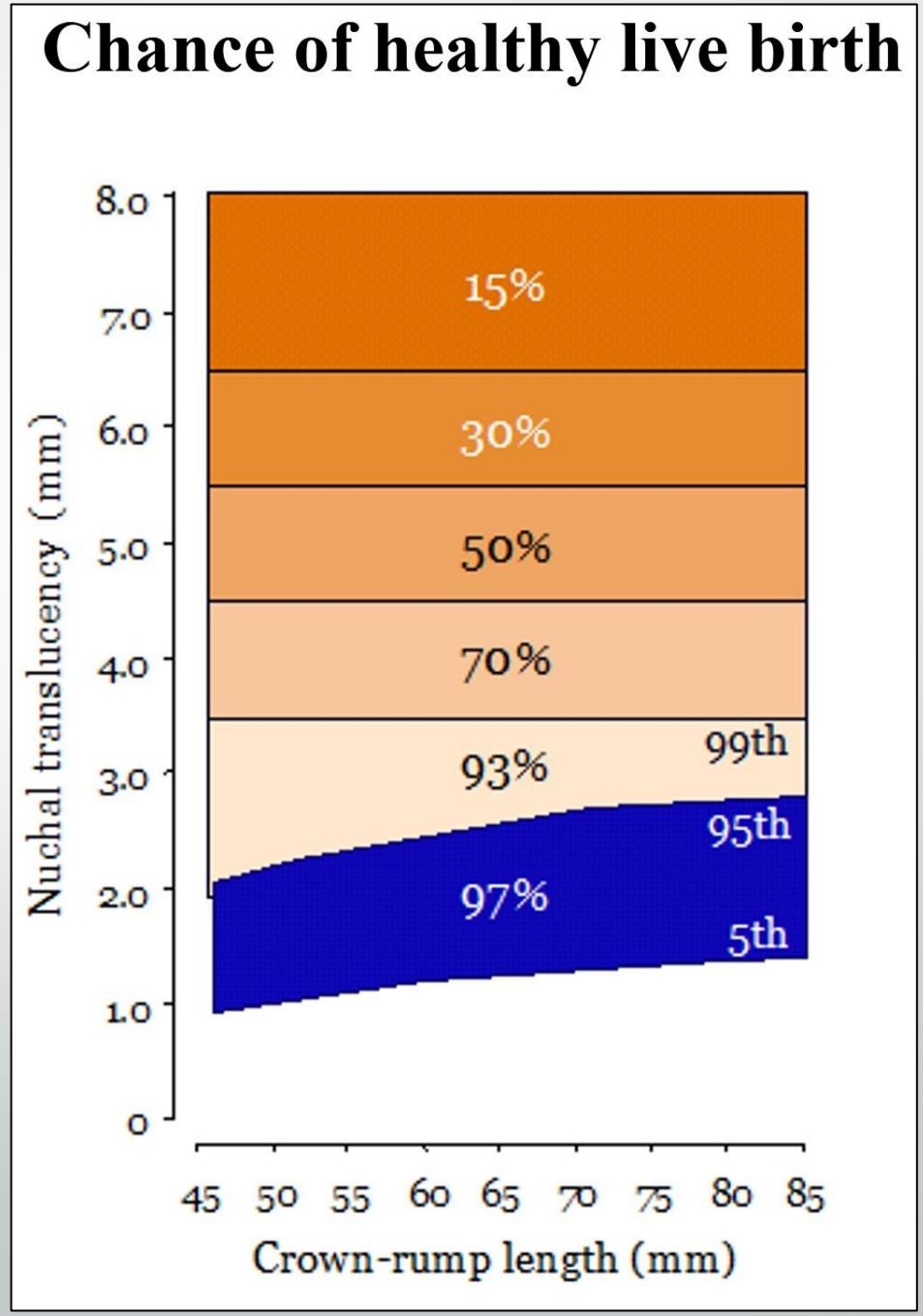
- Risk aneuploidy:
  - not only T13/18/21
  - NIPT not reassuring





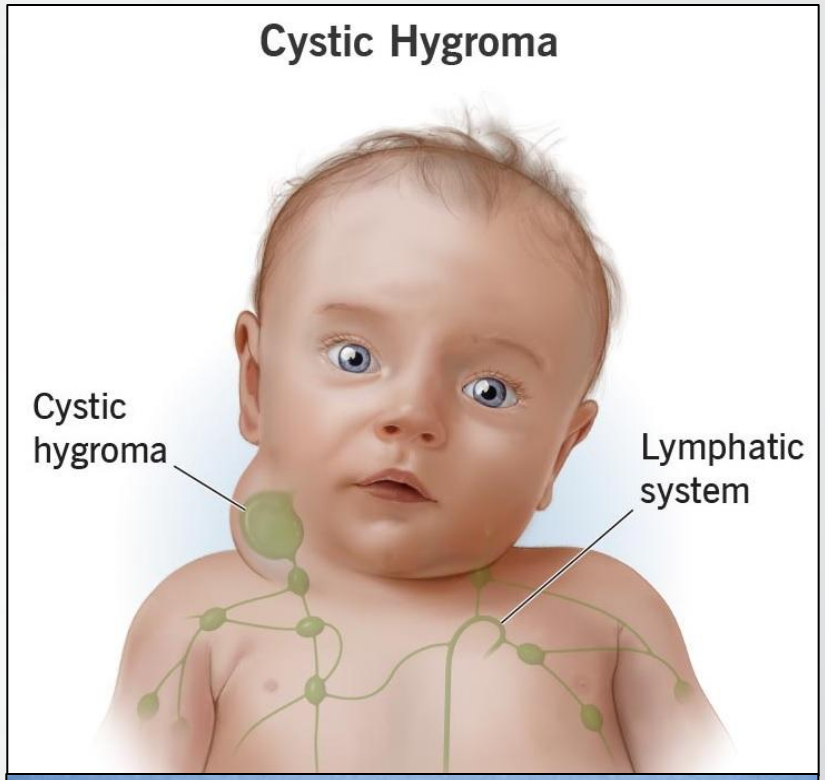
Increased NT:

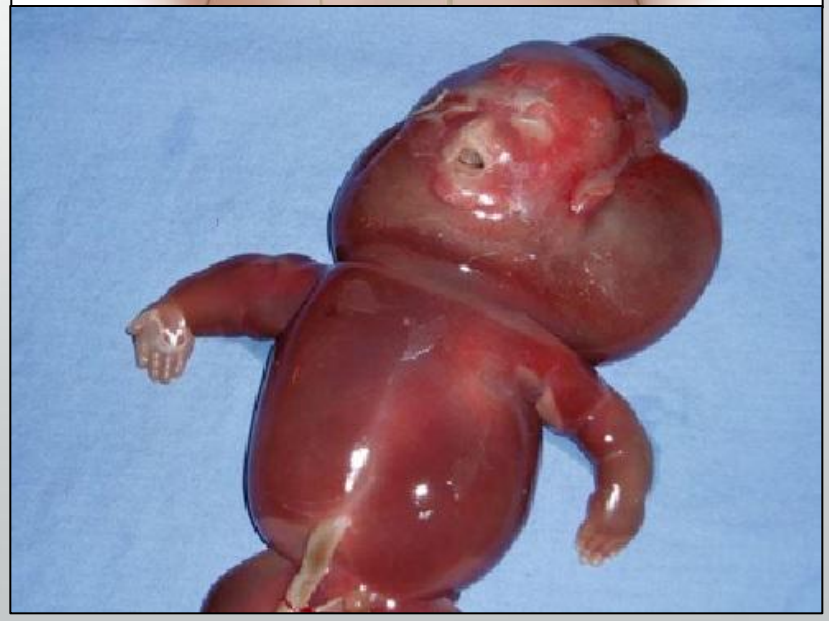
- Other reasons:
  - Genetic syndromes
  - Infection
  - TTTS
  - Normal



Cystic Hygroma

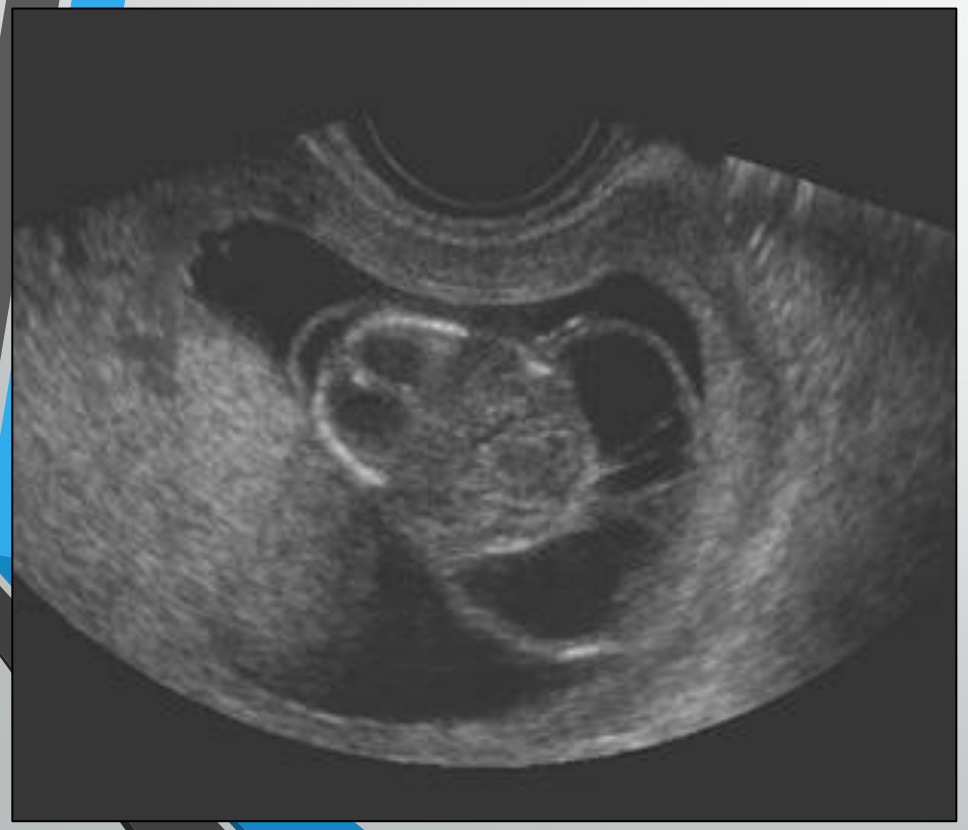
- **Prevalence**
  - 1 in 800 Pregnancies
  - 1 in 8000 live births
- **Associated abnormalities**
  - Chromosomal abnormalities ~50% (Turner Syn.)
  - Genetic syndromes in ~40%:
    - Noonan Syn.
    - Multiple-pterygium S
    - Fryns Syn
    - Neu-Laxova S
  - **Hydrops in 60-80%**

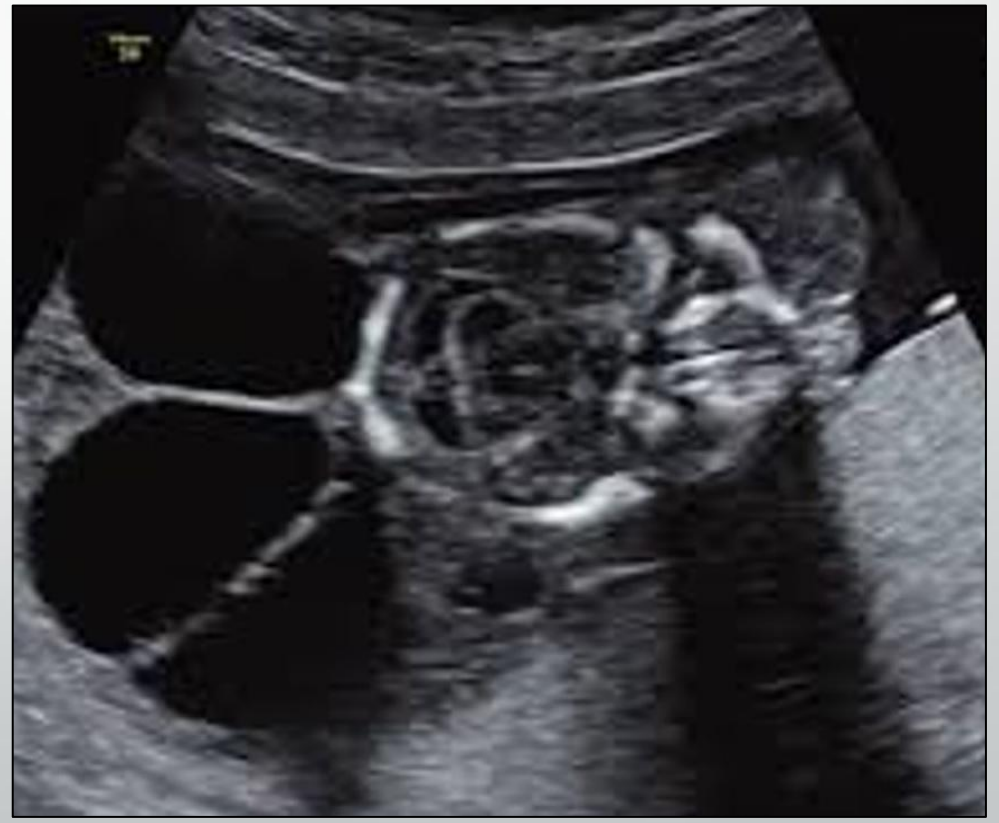




Cystic Hygroma: ultrasound

- Bilateral cystic structures in occipital-cervical region of the neck.
- Nuchal ligament (midline septum) is visible
- Caused by defect in neck lymphatics

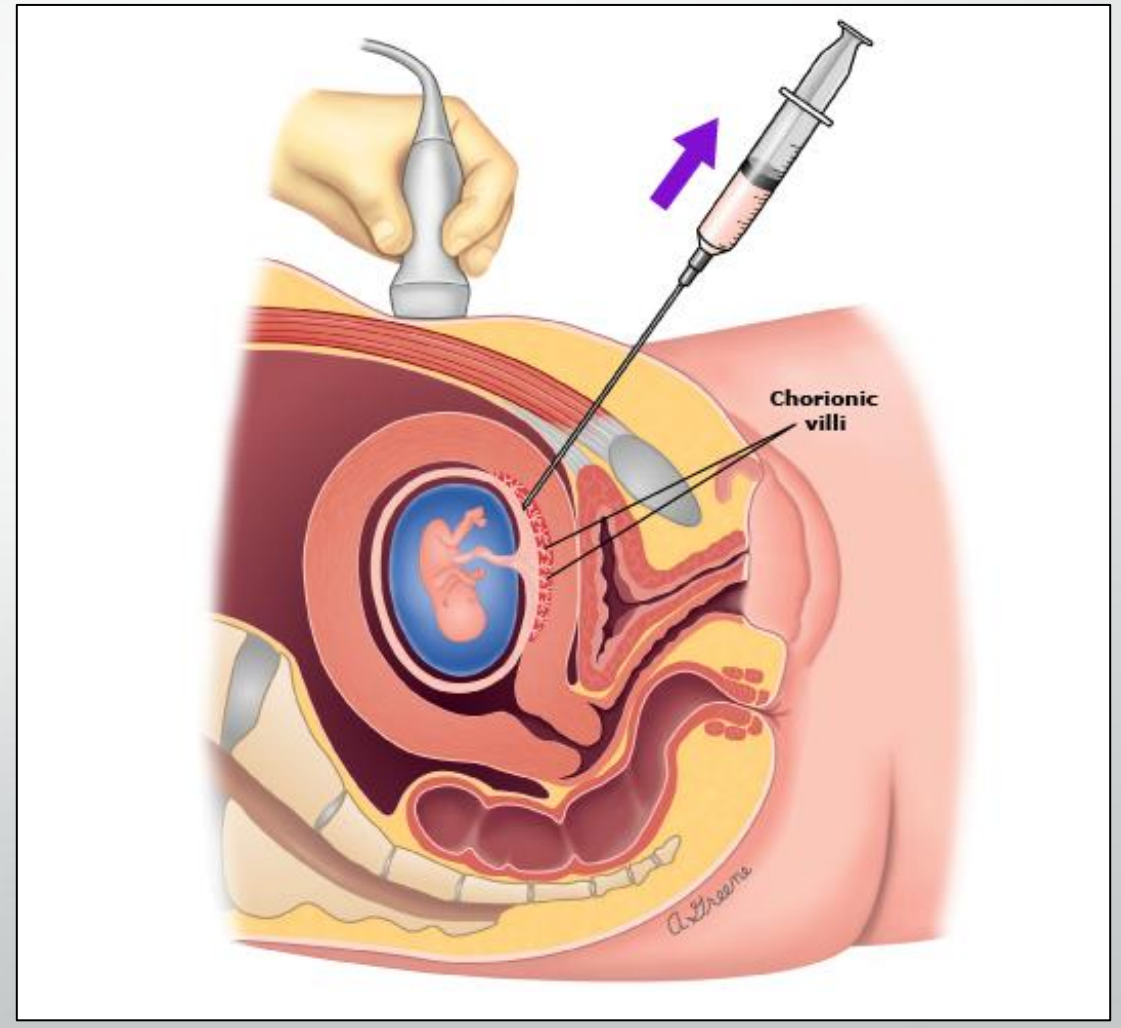




Cystic Hygroma:

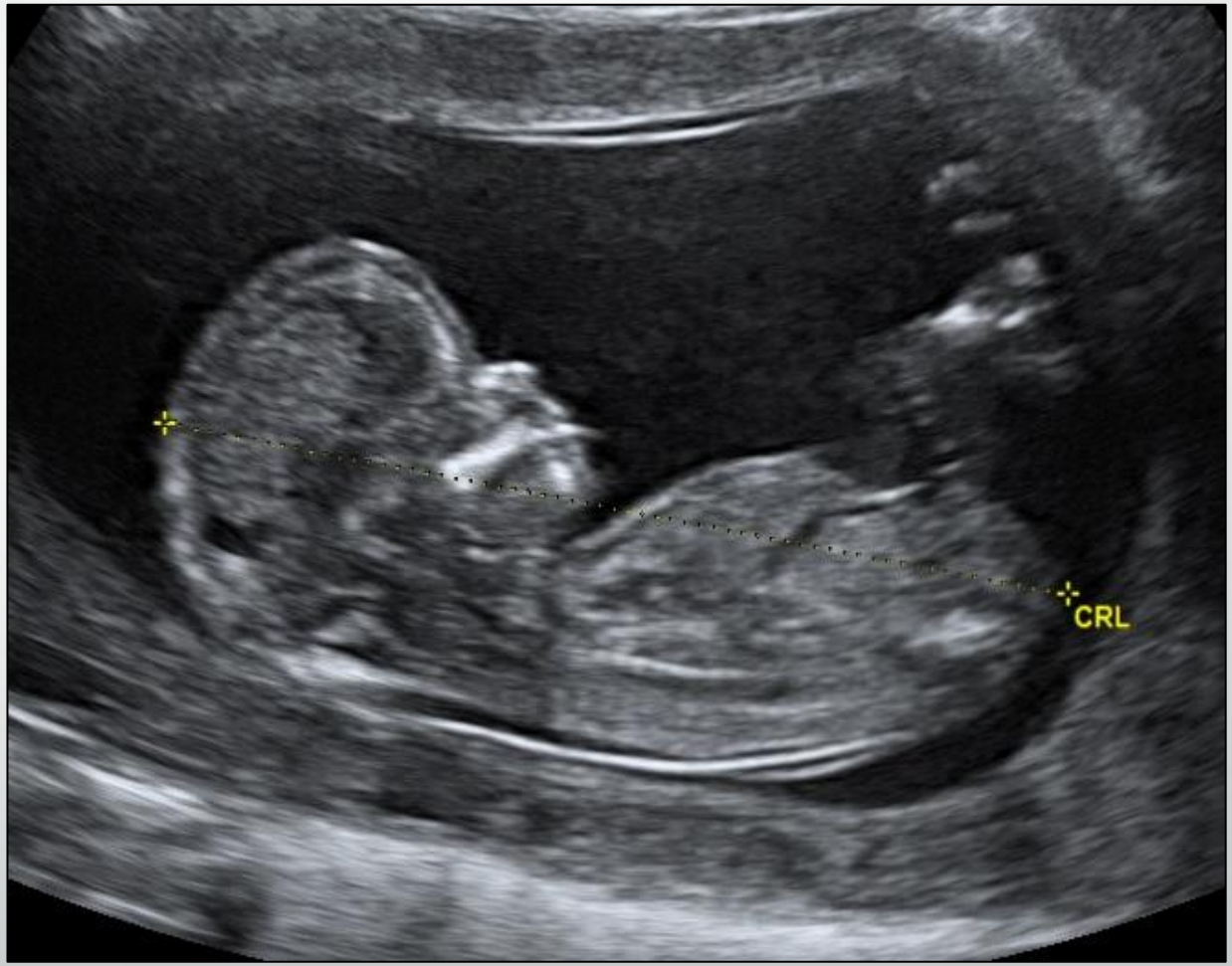
Investigation + Prognosis

- Detailed ultrasound
- Offer:
  - Fetal cardiac echo
  - Invasive testing for SNP array +/- FAGP
- Fetal death in ~90%
- Good prognosis if:
  - Genetic testing normal
  - No associated anomalies
  - CH resolves



First trimester ultrasound scan

- 11 - 13+6 weeks
- Dating
- Early detection anomalies
- Multiple pregnancies:
  - Chorionicity
- Measure NT and first trimester combined screening



EUROCAT

DAVID TUCKER
CARIS Team Manager

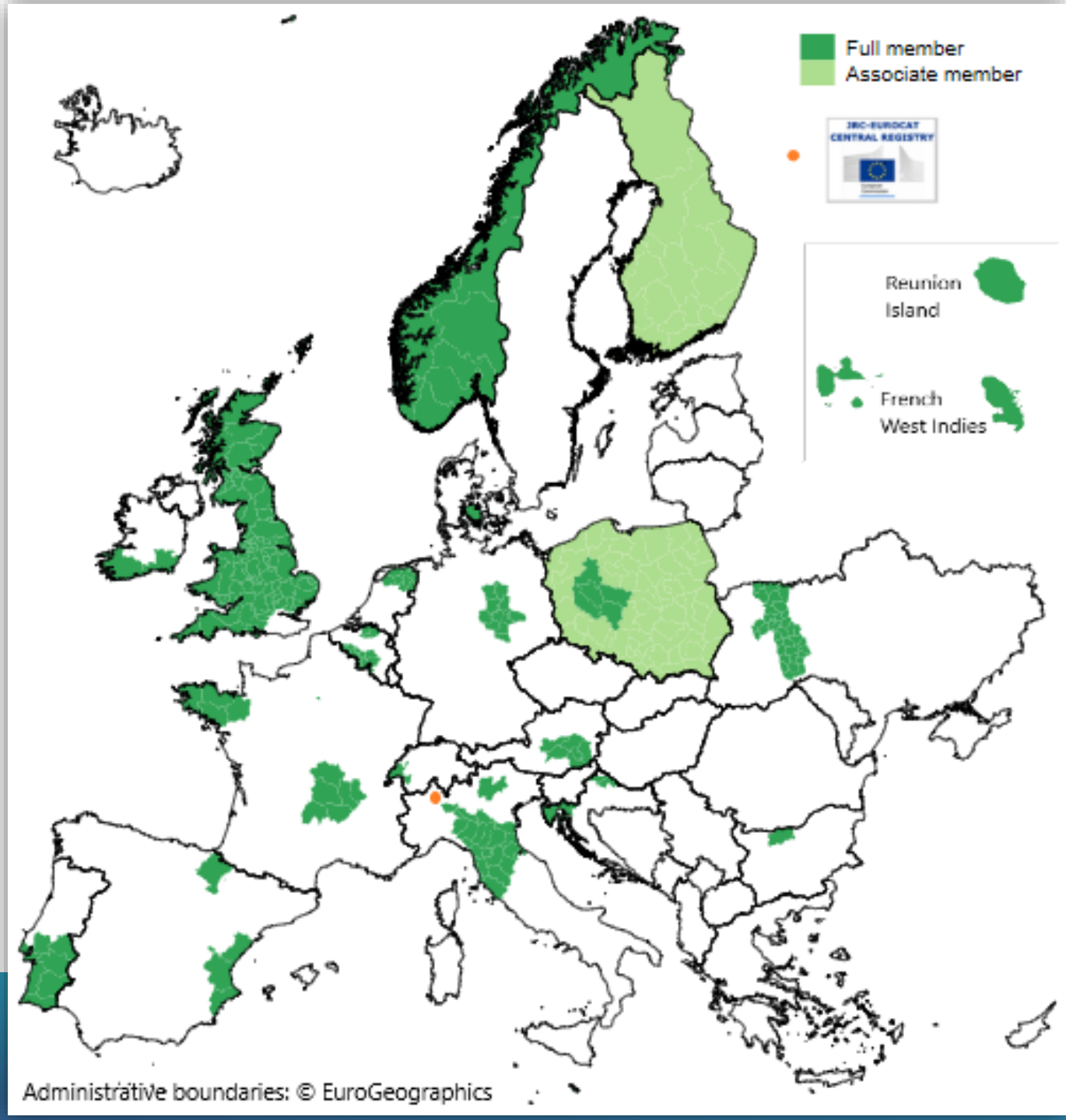
GIG CYMRU NHS WALES
Iechyd Cyhoeddus Cymru
Public Health Wales

EUROCAT

NEWS

European network of population – based registries for the epidemiological surveillance of congenital anomalies.

[EUROCAT | European Platform on Rare Disease Registration]



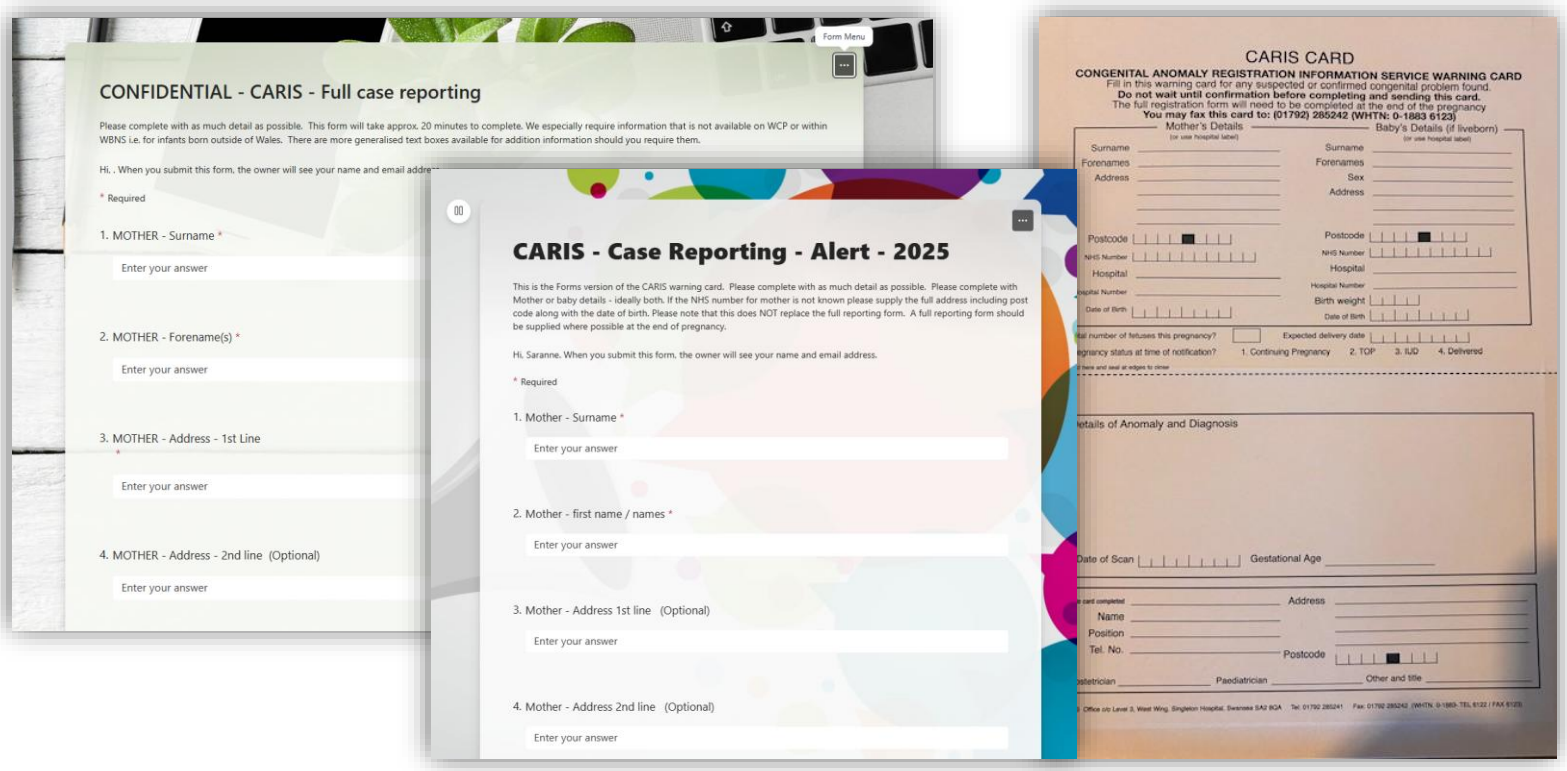
GOOD ASCERTAINMENT OF DATA

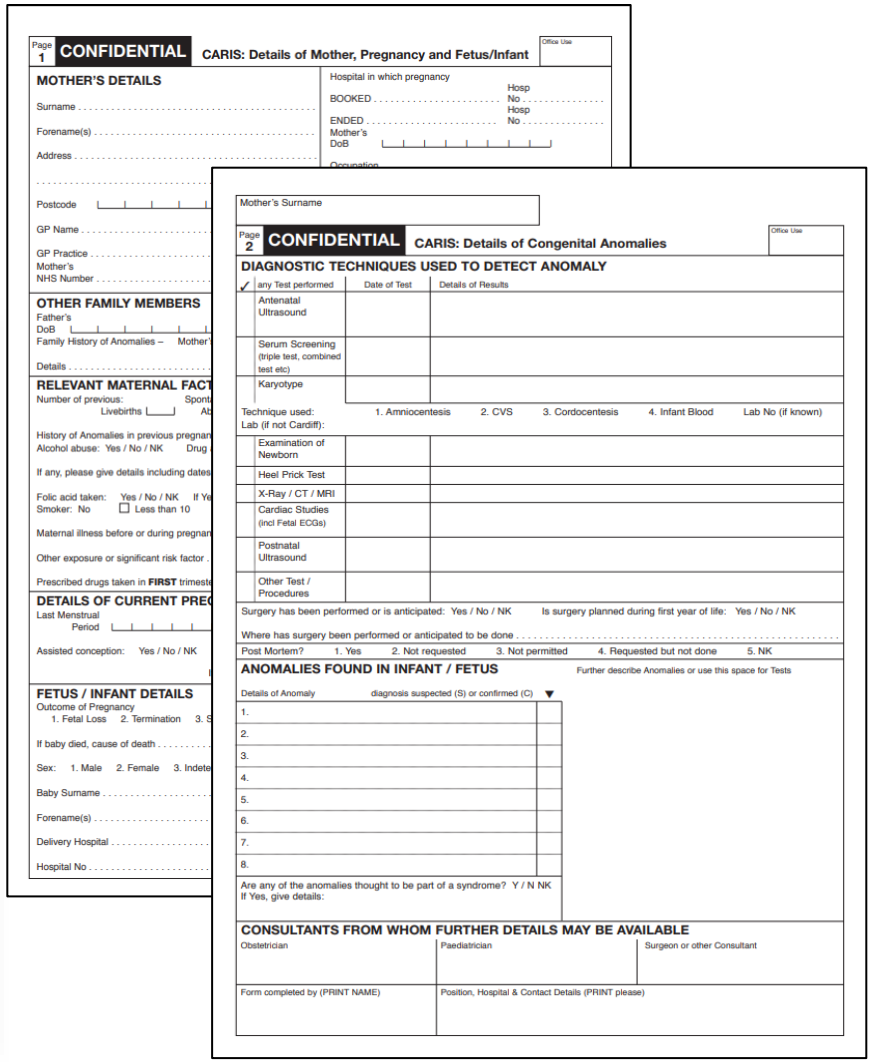
CARIS can't magically make registrations from the clinical portal...

We need YOU.

PLEASE REMEMBER TO…

Continue **reporting cases** to CARIS





Caris.SafehavenMailbox@wales.nhs.uk

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Public Health Wales

Fetal Cardiac clinic letters

Blood Spot results

NIPE Findings

Ophthalmology clinic letters

Newborn Hearing test

Newborn Eye test

Maternal Infections

Hip USS Results

Antenatal Scan Results

Genetics testing results

PM Report Results

Maternal Syphilis / HIV

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Public Health Wales

PLEASE REMEMBER TO…

Continue **reporting cases** to CARIS

- By email to: caris.safehavenmailbox@wales.nhs.uk
- **Digitally: by visiting our report to CARIS QR links OR page**
- **https://phw.nhs.wales/services-and-teams/caris/contact-caris/**
- **Reporting to CARIS - Public Health Wales**
- Traditional mail (Full case data) with downloadable forms
- E-Forms – for quick flagging of cases or full case reports
- Maternal infections – HIV and Syphilis case reporting.

ENGLISH

CYMRAEG

CARIS Links

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

Annual Report and Data 2025

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Rare Disease Data 2025

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Antenatal Screening 2025

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Report to CARIS – caris.safehavenmailbox@wales.nhs.uk

General communications – caris@wales.nhs.uk

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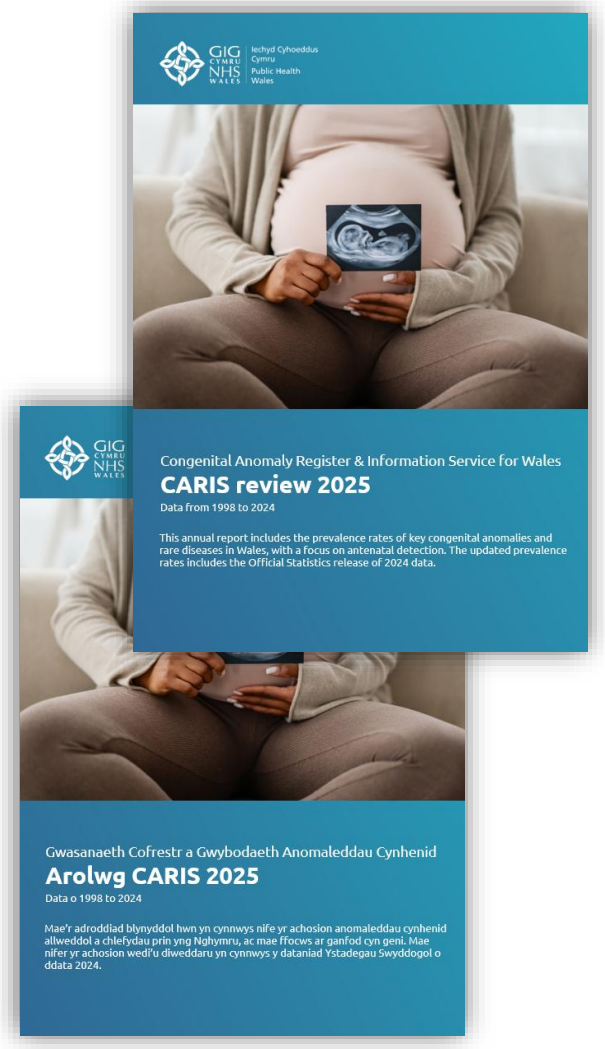
AND FINALLY….

We will provide CPD certificates upon receipt of completed feedback, so if you are viewing as part of a single login (i.e. room viewers) please contact us with your NHS emails!

We design our reports and presentations with **YOU** in mind, so every bit of feedback helps shape CARIS future content.



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FEEDBACK

ENGLISH

CYMRAEG

caris@wales.nhs.uk

Thank you for attending today

Diolch am fod yn bresennol heddiw.

caris

Gwasanaeth Cofrestr a Gwybodaeth Anomaleddau Cynhenid Cymru
Congenital Anomaly Register & Information Service for Wales

caris@wales.nhs.uk

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