

# Prenatal Assessment of Aortic Arch Hypoplasia and Coarctation

Tara Bharucha



# Introduction

- Prenatal diagnosis allows delivery/postnatal planning
- Accuracy of prenatal diagnosis of aortic coarctation/hypoplasia remains a challenge
  - False negative diagnosis can lead to devastating outcomes
  - False positive diagnosis can lead to significant parental anxiety and healthcare resource usage



# Impact of Prenatal Diagnosis

- Prenatal diagnosis facilitates
  - Information giving and counselling
  - Parental choice of course of pregnancy
  - Planning of delivery in a cardiac centre
  - Postnatal management planning
    - Institution of prostaglandin infusion
    - Surgical planning



# Impact of Prenatal Diagnosis

- Duct-dependent lesions diagnosed antenatally\*
  - Improved arterial pH
  - Improved oxygenation
  - Less myocardial dysfunction
  - Less end-organ damage

\*Tworetzky Circulation 2001,  
Tzifa Arch Dis Child 2007

**Table 3** Trend in outcome and patient condition by route of presentation by which congenital heart disease was recognised

Outcome measure	Route			p Value*
	A	B	C	
Ventilation time (median hours)	71	117	90	0.64
Death	21%	12%	8%	<0.05
Cardiovascular compromise	23%	33%	60%	<0.01
End organ dysfunction	5%	11%	29%	<0.01

\* $\chi^2$  test for trend.

Brown *et al.*, *Heart* 2002

- Timing and setting of diagnosis have an impact on preoperative condition, which influences postoperative course and outcome

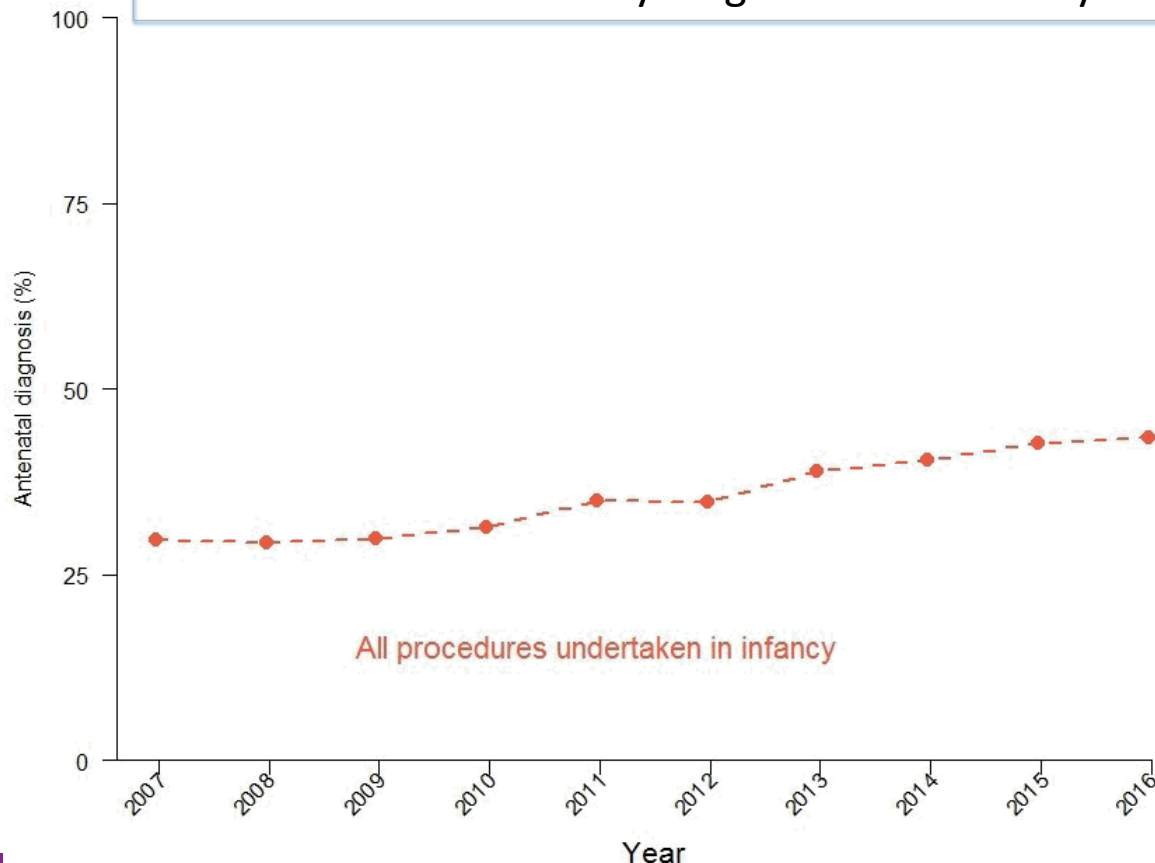
A prenatal diagnosis  
 B diagnosis prior to discharge from maternity unit  
 C diagnosis after discharge home



# Impact of Prenatal Diagnosis

NICOR UK National Congenital Heart Disease Audit 2014-2017

Proportion of patients undergoing procedures in infancy successfully diagnosed antenatally



60% of isolated coarctation diagnosed post discharge\*

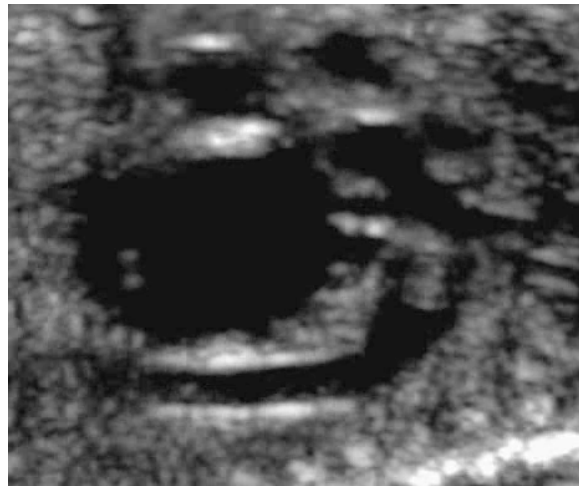
\*Mellander Acta Paediatr 2006, Wren Arch Dis Child 2008

# Challenges of Prenatal Diagnosis

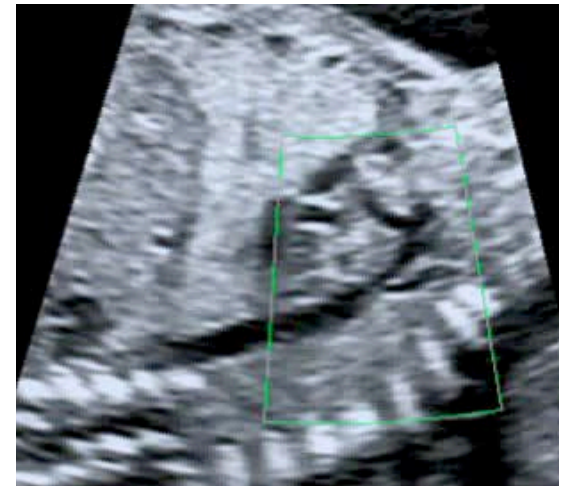
- Coarctation is difficult to diagnose during fetal life
  - Duct is patent
  - Posterior shelf may not be visible
  - No Doppler gradients



Normal



Hypoplastic arch

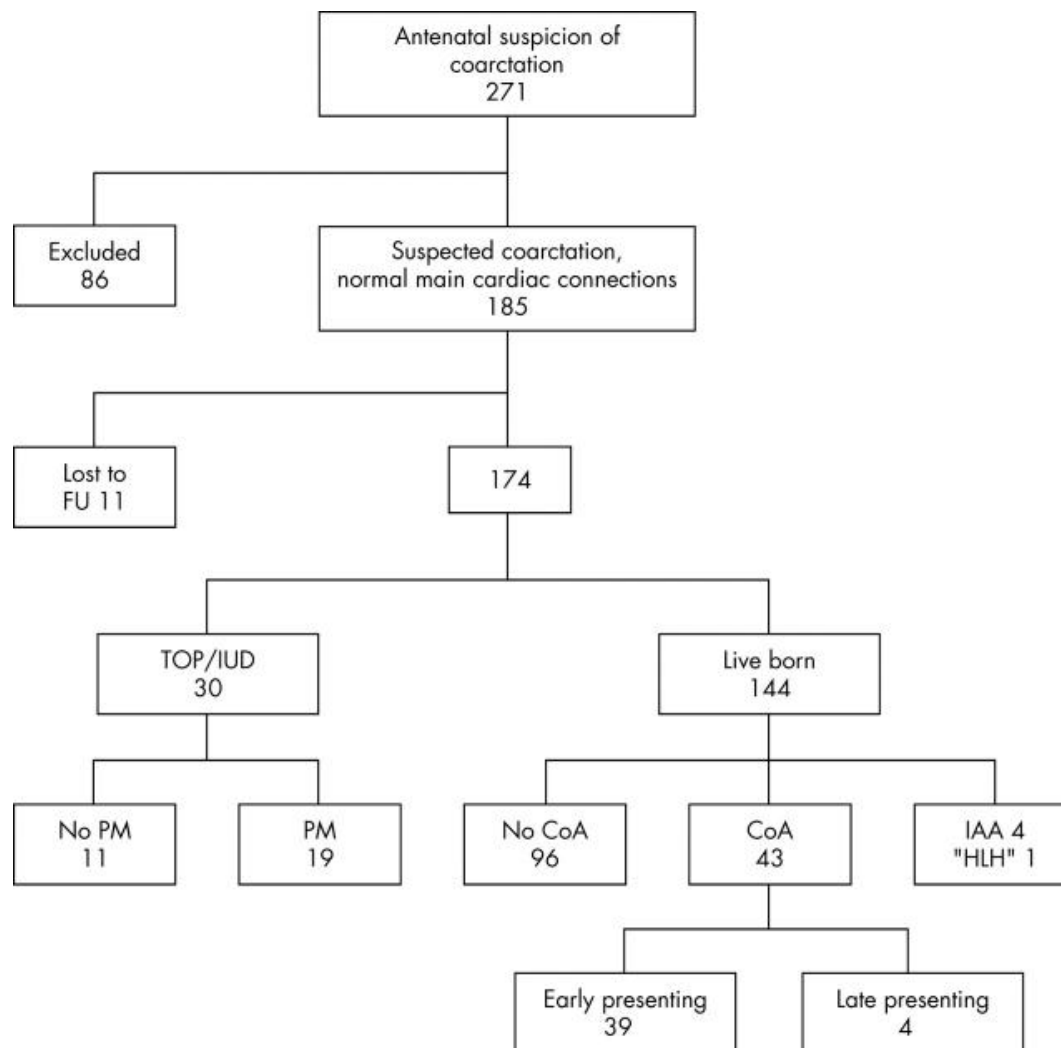


Coarctation

# Timing of presentation and postnatal outcome of infants suspected of having coarctation of the aorta during fetal life

C E G Head, V C Jowett, G K Sharland, J M Simpson

Heart 2005;91:1070-1074.



# Prenatal Suspicion of Arch Hypoplasia/Coarctation

- Common reasons for obstetric sonographer referral for fetal echo

## Abnormal 4C view

- Ventricular disproportion

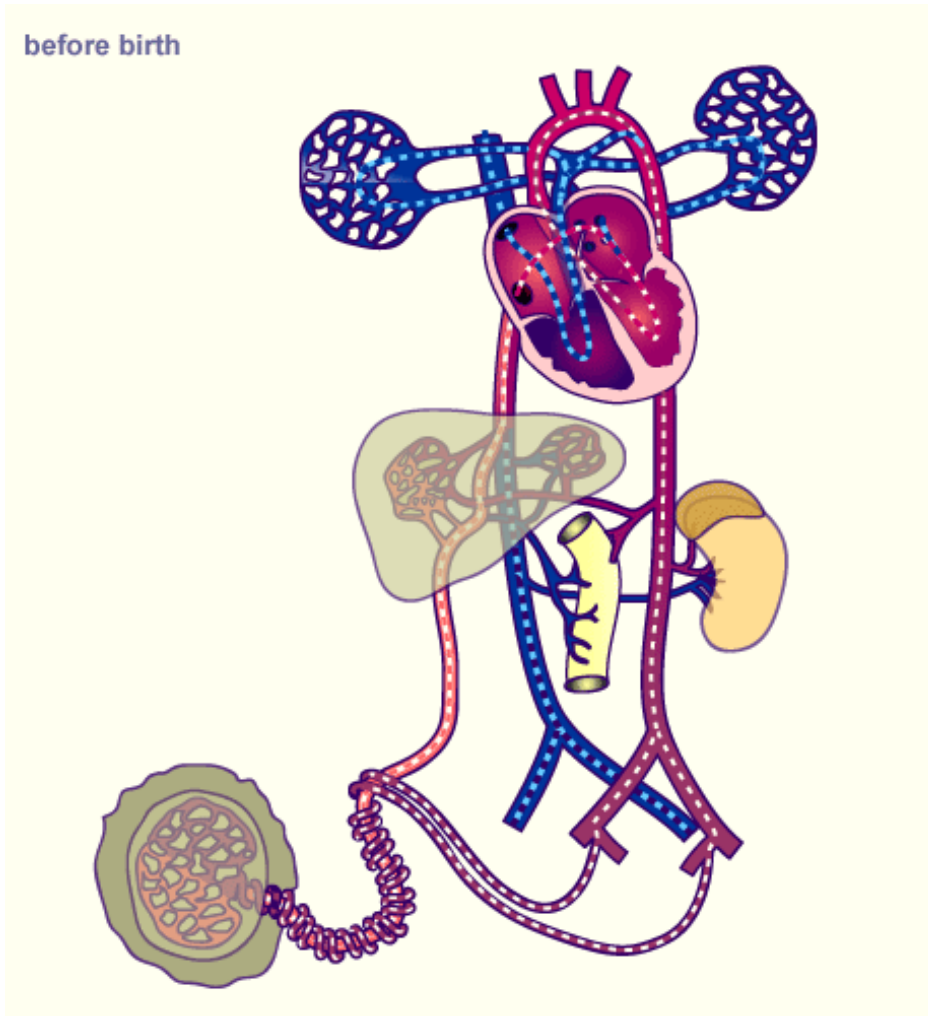


## Abnormal 3VT view

- Aorta:PA disproportion
- L SVC



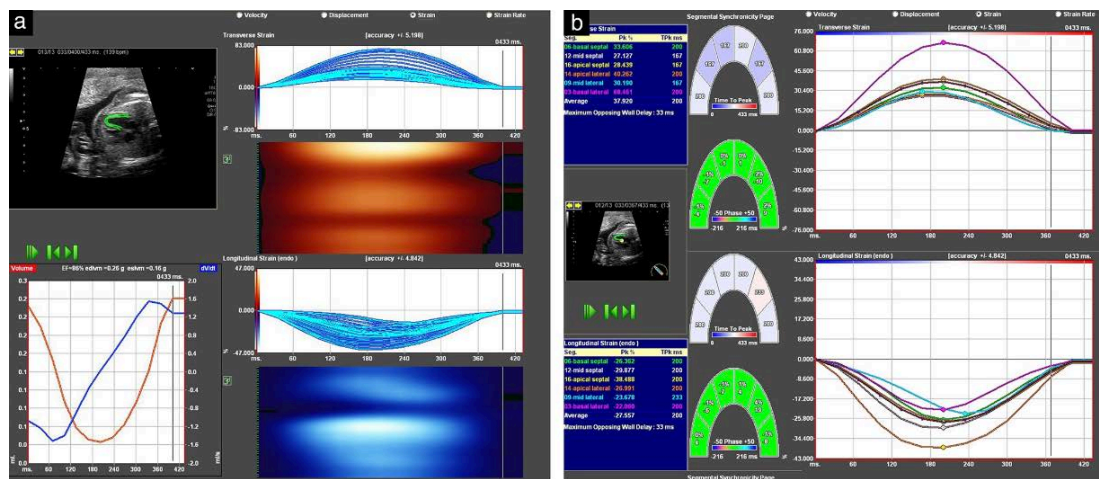
# Foetal Circulation



# Myocardial deformation in fetuses with coarctation of the aorta: a case-control study

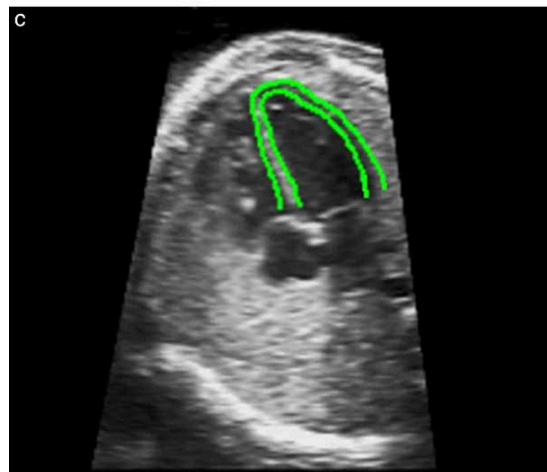
*Ultrasound Obstet Gynecol* 2017;

J. O. MIRANDA<sup>1</sup>, L. HUNTER<sup>1</sup>, S. TIBBY<sup>2</sup>, G. SHARLAND<sup>1</sup>, O. MILLER<sup>1</sup> and J. M. SIMPSON<sup>1</sup>



## Coarctation of controls:

- LV narrower, not shorter in CoA
- LV GLS, systolic strain rate and diastolic strain rate significantly lower
- RV parameters not different



## Possible factors:

Impairment of diastolic filling → ↓ flow through MV & LV  
 → underdevelopment of L heart

- Abnormal differentiation of the myocardium itself  
 → impaired ventricular filling
- Properties of the aorta (abnormal stiffness, increased afterload)



# Ventricular Discrepancy as a Sonographic Sign of Coarctation of the Fetal Aorta:

## How Reliable Is It?

J Ultrasound Med 16:95–99, 1997

Douglas L. Brown, MD, Sara M. Durfee, MD, Lisa K. Hornberger, MD

“Ventricular discrepancy has moderate sensitivity and mediocre positive predictive value for coarctation..... but it is still a useful sign”


*Sensitivity 62%, positive predictive value 33%*



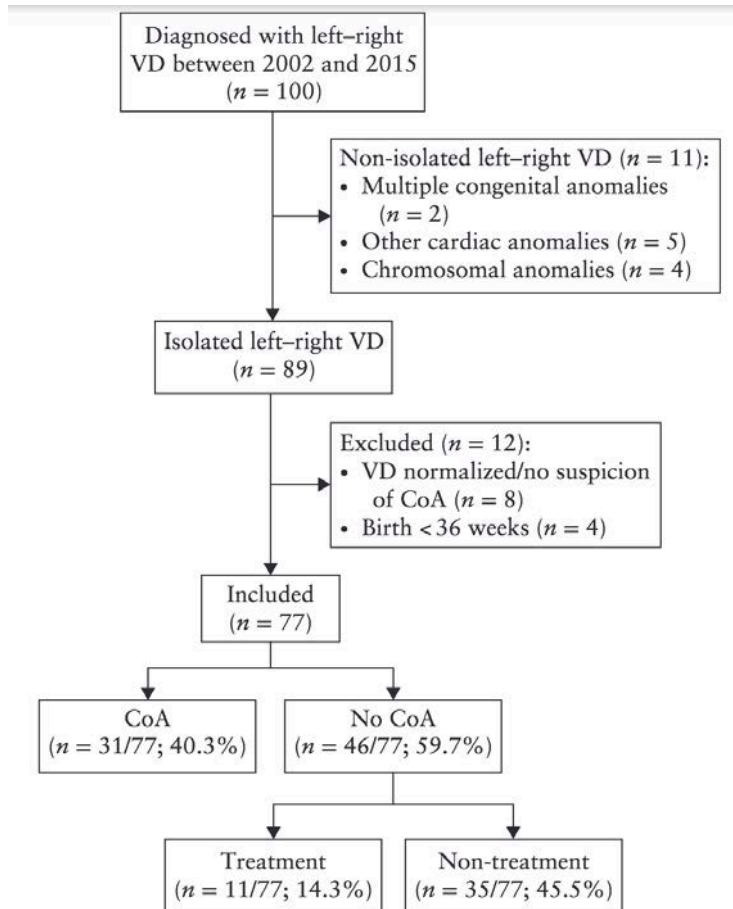
Also associated with

- TAPVD
- VSD
- Bilateral SVCs
- Direct connection of the umbil vein to RA
- Vein of Galen malformation
- IUGR (placental insufficiency)
- Chromosomal abnormalities
- Late gestation

# Postnatal outcome of fetal isolated ventricular size disproportion in the absence of aortic coarctation

A. E. L. VAN NISSELROOIJ<sup>1</sup> , L. ROZENDAAL<sup>2</sup>, I. H. LINSKENS<sup>3</sup>, S. A. CLUR<sup>4</sup>, J. HRUDA<sup>5</sup>, E. PAJKRT<sup>6</sup>, C. L. VAN VELZEN<sup>3</sup>, N. A. BLOM<sup>2</sup> and M. C. HAAK<sup>1</sup>

*Ultrasound Obstet Gynecol* 2018; 52: 593–598



- Fetal finding of ventricular disproportion – No postnatal coarctation:
  - Patients requiring cardiac treatment
    - TAPVD, ASD, VSD, AS MS, PHT
  - Patients not requiring cardiac treatment
    - PLSVC, VSD, ASD, genetic disorders, dysmorphism, PHT



# Antenatal Recognition: Abnormal 3VT View

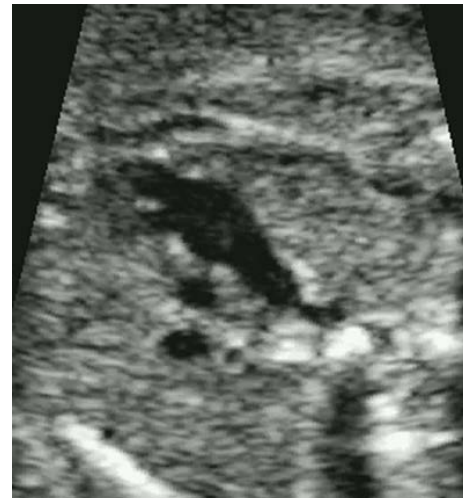
## Three-vessel view of the fetal upper mediastinum: an easy means of detecting abnormalities of the ventricular outflow tracts and great arteries during obstetric screening

*Ultrasound Obstet. Gynecol.* 9 (1997) 173–182

S.-J. Yoo<sup>\*†††</sup>, Y.-H. Lee<sup>\*</sup>, E. S. Kim<sup>†</sup>, H. M. Ryu<sup>†</sup>, M. Y. Kim<sup>†</sup>, H.-K. Choi<sup>†</sup>, K. S. Cho<sup>†</sup> and A. Kim<sup>\*\*</sup>

Suspicion of coarctation if

- Ao:PA disproportion
- L SVC



# Persistent L SVC



# Left superior caval vein: a powerful indicator of fetal coarctation

L Pasquini, A Fichera, T Tan, S Y Ho, H Gardiner

Heart 2005;91:539–540.

**Table 1** Cardiac diagnosis, extracardiac malformations, karyotype, and outcome in 16 fetuses with left superior caval vein

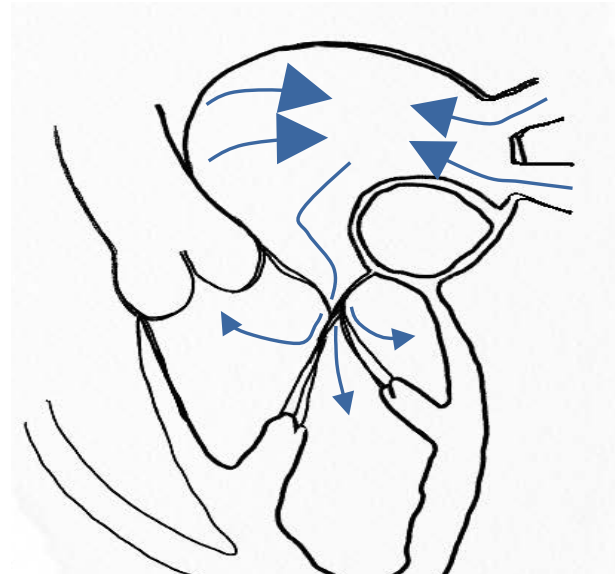
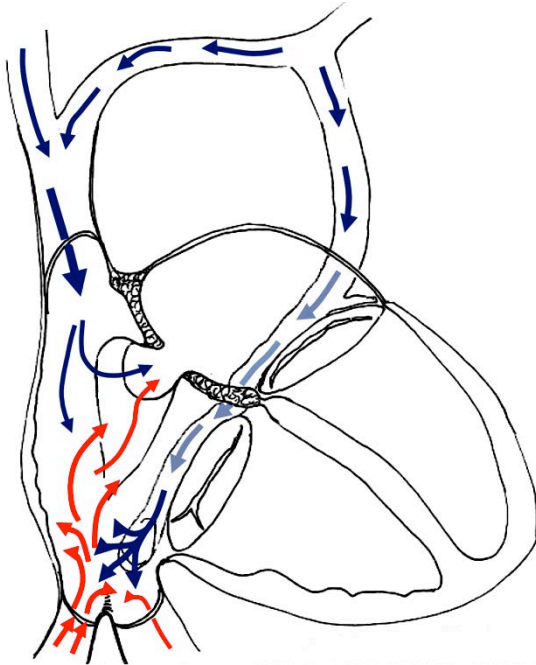
Case	Fetal cardiac diagnosis	Situs	Karyotype	Extracardiac malformations	Outcome
CHD	S,C,C, unbalanced AVSD, dysplastic LAVV	N	46XY	Soft marker (two vessel cord)	Surgery, well
CHD	S,C,C, sub-Ao VSD, CoA	N	46XY	Renal agenesis, SGA, ventriculomegaly, talipes	NND
CHD	S,C,C, CoA (postnatal hypoplasia only), PE	N	45X0	None	Well
CHD	S,C,C, muscular VSD, CoA	N	46XX	None	Surgery, well
CHD	S,C,C, PMVSD, CoA	PV	46XY	SGA, postnatal malrotation and biliary atresia, right sided stomach	Surgery, well
CHD	S,C,C, CoA	PV	46XX	None	Surgery, well
CHD	S,C,D, large VSD, PS, criss-cross heart	PV	46XY	Soft marker (mild hydronephrosis)	Surgery, well
CHD	SI,C, DORV, restrictive VSD, straddling MV	SI	46XY	None	Postoperative death
CHD	S,C,C, infradiaphragmatic TAPVC	PV	46XY	Massive bilateral pulmonary sequestration	NND
CHD	S,C,C, VSD and TAPVC to confluence	N	T18	Two vessel cord, SGA, rocker-bottom feet, microcephaly	NND
CHD	S,C,C, TOF	N	46XY	None	Surgery, well
CHD	S,C,C, dextrocardia, unbal. AVSD, aortic atresia	PV	46XY	None	NND
Normal	S,C,C, right aortic arch	N	46XX	None	Well
Normal	S,C,C, small PE	N	46XX	None	Well
Normal	S,C,C, biventricular hypertrophy	N	46XX	None	Well
Normal	S,C,C	N	45X0	None	Well

45X0, Turner syndrome; AVSD, atrioventricular septal defect; C, concordant atrioventricular or ventriculoarterial connection; CHD, congenital heart disease; CoA, coarctation of the aorta; D, discordant atrioventricular or ventriculoarterial connection; DORV, double outlet right ventricle; LAVV, left atrioventricular valve; N, normal situs; NND, neonatal death without cardiac surgery; PE, pericardial effusion; PMVSD, perimembranous ventricular septal defect; PV, posterior vein (azygous/hemiazygous) and absent inferior vena cava at situs; S, situs solitus; SGA, small for gestational age; SI, situs inversus; T18, trisomy 18; TAPVC, total anomalous pulmonary venous connections; TOF, tetralogy of Fallot; unbal, unbalanced; VSD, ventricular septal defect.



# Persistent Left SVC

## Theories of association with coarctation



- Interference with IVC flow -> FO -> LA
- Obstruction to flow LA -> LV
- Likely to be multifactorial

# Risk Factors for Coarctation of the Aorta on Prenatal Ultrasound

## A Systematic Review and Meta-Analysis

Alessandra Familiari

*Circulation.* 2017;135:772–785.

**Table 2.** Quality Assessment of the Included Studies According to the Newcastle-Ottawa Scale

Author	Year	Selection	Comparability	Outcome
Arya <sup>14</sup>	2016	★★★	★★	★★
Toole <sup>15</sup>	2016	★★	★★	★★
Mărginean <sup>16</sup>	2015	★★★	★	★
Durand <sup>17</sup>	2015	★★★	★★	★★
Sivanandam <sup>18</sup>	2015	★★	★	★
Gomez-Montes <sup>19</sup>	2015	★★★	★★	★★
Rizzo <sup>20</sup>	2014	★★	★	★
Slodki <sup>21</sup>	2010	★★	★	★
Axt-Flidner <sup>22</sup>	2009	★★	★	★
Matsui <sup>23</sup>	2008	★★★	★★	★★
Head <sup>24</sup>	2008	★★★	★★	★★
Hornung <sup>25</sup>	2002	★★	★	★

- 12 studies, 922 fetuses undergoing echo *for suspicion of CoA*
- Mean GA ranged from 22 – 36 weeks
- Reference standard postnatal echocardiography and/or surgery



**Table 3. Results of the Meta-Analyses Comparing the Echocardiographic Parameters of Different Cardiac Structures in Fetuses With Diagnosis of Coarctation of the Aorta Versus Normal Fetuses**

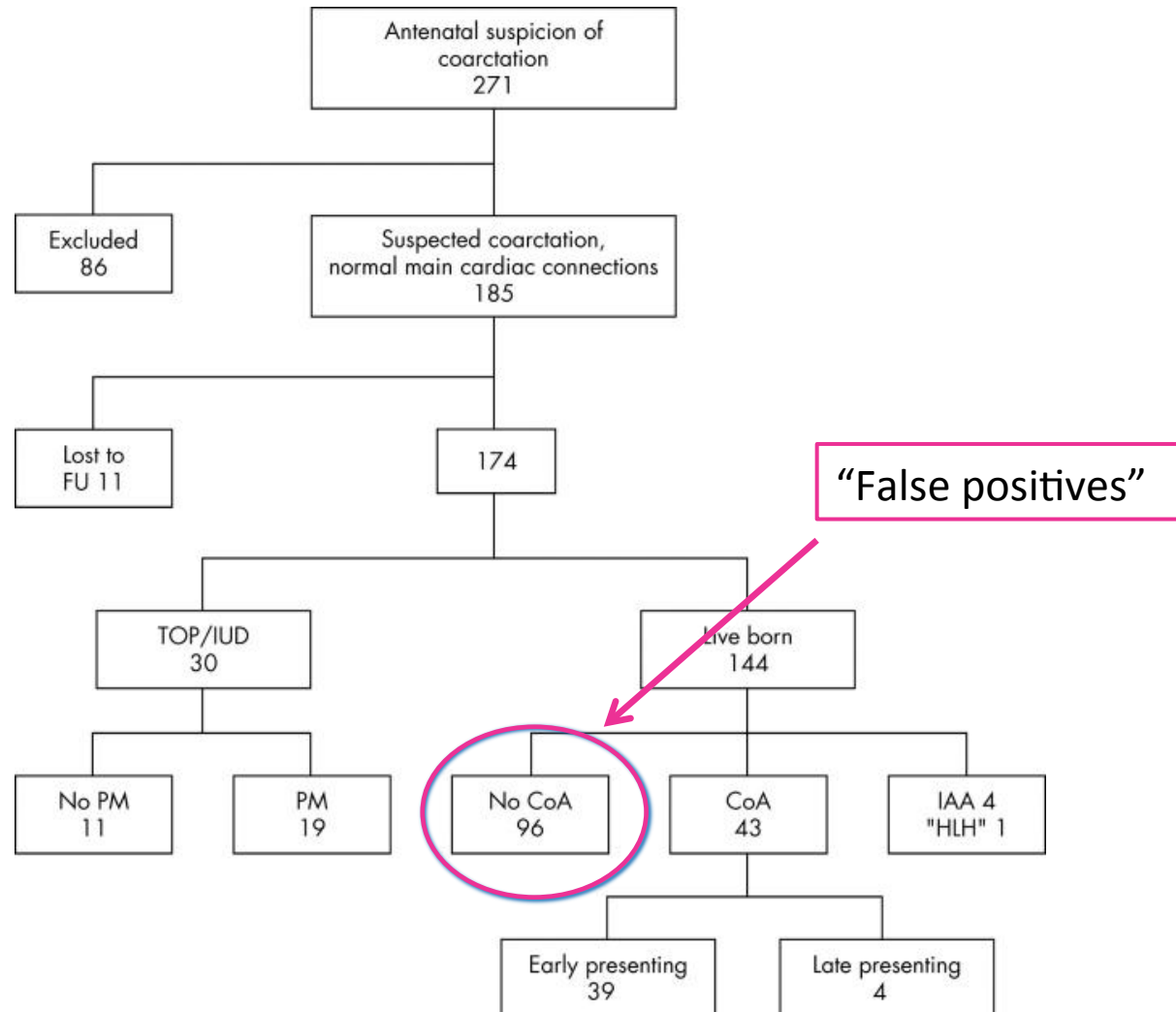
Continuous Parameters	No. Studies (Total Sample)	Reference	n/N*	Mean Difference (95% CI)	P Value	P, %
Atrioventricular valves						
→ Tricuspid valve z score	2 (177)	15,19	79/98	0.40 (0.09 to 0.71)	0.01†	0
→ Mitral valve z score	3 (208)	15,18,19	90/118	-0.97 (-1.43 to -0.51)	<0.001†	0
Outflow tracts						
→ Aortic valve z score (based on GA)	4 (383)	15,17-19	130/253	-1.19 (-1.56 to -0.82)	<0.001†	32
Ascending aorta z score (based on GA)	2 (177)	15,19	79/98	-0.95 (-1.97 to 0.07)	0.07	85
Ascending aorta diameter, mm	2 (84)	16,21	21/63	-0.78 (-2.86 to 1.31)	0.47	87
→ Aortic isthmus z score (sagittal view)	4 (248)	14,15,18,19	110/138	-1.24 (-2.27 to -0.22)	0.02†	82
→ Aortic isthmus z score (3-vessel view)	3 (178)	16,18,19	72/106	-1.47 (-2.27 to -0.68)	<0.001†	74
→ Aortic isthmus diameter, mm	2 (84)	16,21	21/63	-0.99 (-1.21 to -0.77)	<0.001†	0
→ Pulmonary valve z score	2 (177)	15,19	79/98	0.73 (0.32 to 1.13)	<0.001†	0
Ratios						
→ Right ventricle/left ventricle (diameters, mm)	2 (72)	14,16	29/43	0.21 (0.04 to 0.39)	0.02†	0
Right ventricle/left ventricle (areas, mm <sup>2</sup> )	2 (102)	14,15	47/55	0.25 (-0.01 to 0.51)	0.06	0
Pulmonary artery/ascending aorta (diameters, mm)	4 (217)	15,19-21	81/136	0.43 (0.07 to 0.78)	0.02†	90
→ Aortic isthmus/arterial duct (diameters, mm)	2 (161)	15,19	63/98	-0.13 (-0.19 to -0.08)	<0.001†	0
Arterial duct/aortic isthmus (diameters, mm)	2 (70)	14,16	28/42	0.24 (-0.17 to 0.65)	0.25	82



# Timing of presentation and postnatal outcome of infants suspected of having coarctation of the aorta during fetal life

C E G Head, V C Jowett, G K Sharland, J M Simpson

Heart 2005;91:1070-1074.



# Impact of False Positive Diagnosis

## Impact of prospective measurement of outflow tracts in the prediction of coarctation of the aorta

Vigneswaran et al.,  
*Ultrasound Obs Gynae* 2019

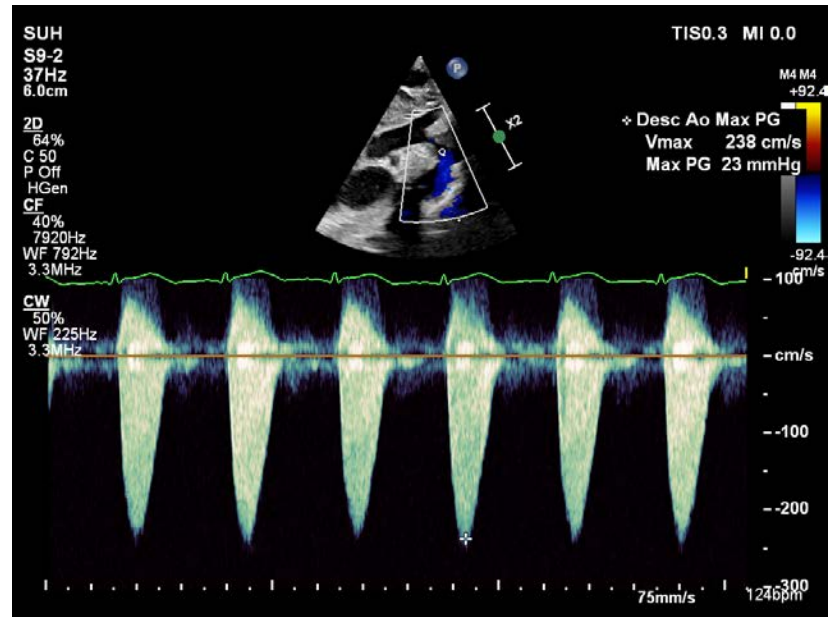
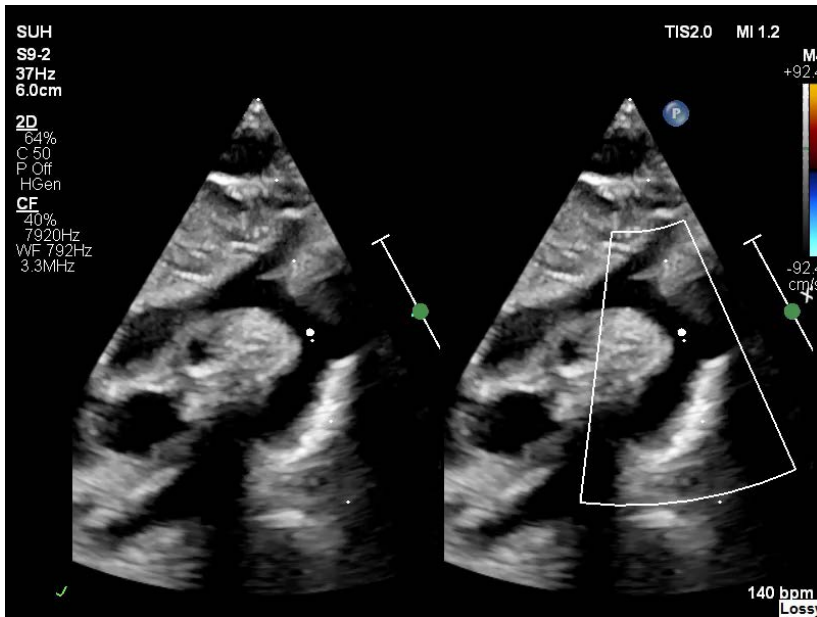
	False positive (n=72)	True coarctation (n=77)	P value
Median gestational age (range) in weeks	23 (19-35)	23 (15-37)	0.76
Nuchal translucency >95 <sup>th</sup> centile	12/47	13/41	0.52
Persistent left SVC	20/72 (28%)	16/77 (21%)	0.32
No. of patients with VSD	20/72 (28%)	34/77 (44%)	0.038*
No. of confirmed cases of bicuspid aortic valve	18/59 (30%)	34/60 (57%)	0.004*



# Antenatal Suspicion of Coarctation



# Postnatal Echo



- Early postnatal surveillance until duct closes
- Ongoing follow up until < 1 year of age




# Impact of False Positive Diagnosis

- Repeat fetal scans
- Impact on birth plan and delivery centre
- Delay in discharge home after birth
- Repeat hospital visits
- Parental distress during pregnancy and in postnatal period
- Financial cost to service and family



# Impact of False Positive Diagnosis

## Parental reactions, distress, and sense of coherence after prenatal versus postnatal diagnosis of complex congenital heart disease

Ewa-Lena Bratt<sup>1,2</sup> , Stina Järholm<sup>3</sup>, Britt-Marie Ekman-Joelsson<sup>2</sup>,  
Antje Johannsmeyer<sup>4</sup>, Sven-Åke Carlsson<sup>5</sup>, Lars-Åke Mattsson<sup>3</sup> and  
Mats Mellander<sup>2,6</sup>

*Cardiology in the Young* **29**: 1328–1334.

Compared to parents with a postnatal diagnosis, parents with a prenatal diagnosis of CHD report

- More depression
- Similar anxiety levels
- Low sense of coherence during pregnancy which decreases during FU
- Lower satisfaction with life



# Perinatal Maternal Mental Health, Fetal Programming and Child Development

*Healthcare 2015,*

**Andrew J. Lewis <sup>1,2,\*</sup>, Emma Austin <sup>1</sup>, Rebecca Knapp <sup>1</sup>, Tina Vaiano <sup>1,3</sup> and Megan Galbally <sup>3,4</sup>**

- Developmental Origins of Health and Disease (DOHaD model)
  - Multiple complex and interacting maternal factors can have an impact on fetal and later life development
- 
- Depressed mothers more likely to take antidepressants, smoke, drink alcohol
  - Maternal psychological distress → metabolic and functional changes
    - Disrupt maternal/placental endocrine function
    - Induce fetal oxidative stress
    - Reduce fetal circulation of IGF1 and IGF2



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- Hormonal imprinting and placental programming – fetal exposure to maternal/placental glucocorticoids has a range of impacts:
  - Growth restriction
  - Hormonal programming of the stress response
  - Vulnerability to range of cognitive and emotional disorders
- Epigenetic programming – changes in epigenetic markers associated with exposures such as stress, depression, postnatal caregiving
  - Altered epigenetic profiles could link between specific intrauterine and postnatal exposures and later child health and mental health outcomes



# Impact of False Negative Diagnosis

- Prenatal diagnosis of coarctation improves survival and decreases morbidity\*
  - Improved haemodynamic stability pre-operatively
  - Duct patency virtually universal
  - No or little collapse and death

\*Franklin *et al.*, *Heart* 2002

**Table 1** Outcome variables

	Antenatal diagnosis	Postnatal diagnosis	p Value	M-H odds ratio (p)
Femoral pulse				
Good	10	1		
Weak	0	6	<0.0001	<0.0001
Absent	0	15		
Collapse/death				
Yes	0	10	<0.05	
No	10	15		
Left ventricular function				
Good	10	13	>0.05	>0.05
Poor	0	9		
Respiratory rate (mean) (breaths/min)	37.9	52.6	<0.05	
Arterial duct				
Open	8	6		
Restricted	2	9	<0.05	<0.05
Closed	0	7		
Heart rate (mean) (beats/min)	134.9	146.5	>0.05	
Systolic blood pressure (mean), R upper limb (mm Hg)	75.3	90.04	>0.05	
PT/APTT (mean)	1.2	1.3	>0.05	
Plasma potassium (mean) (mmol/l)	5.02	4.9	>0.05	
Plasma creatinine (mean) (µmol/l)	93.7	64.7	>0.05	
ITU stay (mean) (days)	6.9	6.9	>0.05	
Hospital stay (mean) (days)	22	18.2	>0.05	
Total score	3.00	6.59	<0.01	

APPT, activated partial thromboplastin time; ITU, intensive care unit; M-H, Mantel-Haenszel test; PT, prothrombin time; R, right.



# Conclusions

- Accurate antenatal diagnosis remains challenging
  - Overall detection rate at the time of the anomaly scan is poor
- Accurate antenatal diagnosis of aortic coarctation/hypoplasia allows planned postnatal management and reduces morbidity
- Cardiovascular disproportion using several parameters is the best fetal marker
- False positive diagnosis
  - raises parental anxiety and distress
  - has an unquantified effect on childhood development



THANK YOU



# Neck Web and Congenital Heart Defects: A Pathogenic Association in 45 X-O Turner Syndrome?

EDWARD B. CLARK

TERATOLOGY 29:355-361 (1984)

- Association between web neck and coarctation in Turner syndrome
- Hypothesis that increased lymphatic pressure associated with jugular sac obstruction distends thoracic ducts
- Compression of ascending aorta – increased resistance to left sided blood flow

