



Effectiveness and pitfalls of fetal screening for transposition

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- ★ Prenatal detection rates for TGA have been generally low
 - Often four chamber view normal
 - Very low association with other abnormalities
- ★ Has been improvement in prenatal detection TGA with introduction of outflow tract views during obstetric anomaly scans
- ★ But most published series still report less than 50% detection rate for TGA





Bull C on behalf British Paediatric Cardiac Association, Lancet 1999

Current and potential impact of fetal diagnosis on prevalence and spectrum of serious congenital heart disease at term in the UK

- 4799 affected pregnancies
- Prenatal detection overall 23.4%
- Prenatal detection of TGA 3% overall in UK

★ BINOCAR/EUROCAT 2008-2012

- Prenatal detection TGA 44.8%



Ultrasound Obstet Gynecol. 2015 Mar;45(3):320-5. doi: 10.1002/uog.14689. Epub 2015 Jan 27.

Prenatal detection of transposition of the great arteries reduces mortality and morbidity.

van Velzen CL¹, Haak MC, Reijnders G, Rijlaarsdam ME, Bax CJ, Pajkrt E, Hruda J, Galindo-Garre F, Bilardo CM, de Groot CJ, Blom NA, Clur SA.

- ★ Evaluation of prenatal detection of TGA after introduction of Dutch screening programme in 2007

- ★ N = 144 in 10 years (2002-2012)
 - Overall 26.4% diagnosed prenatally
 - 15.7% diagnosed prenatally in the first 5 years study
 - 41.0% diagnosed prenatally in last 5 years of the study period



Ultrasound Obstet Gynecol. 2015 June ; 45(6): 678–682. doi:10.1002/uog.14751.

Prenatal Diagnosis of Transposition of the Great Arteries over a 20-Year Period: Improved but Imperfect

Maria C Escobar-Diaz, MD^{*‡}, Lindsay R Freud, MD^{*‡}, Alejandra Bueno, MD^{*‡}, David W Brown, MD^{*‡}, Kevin Friedman, MD^{*‡}, David Schidlow, MD^{*‡}, Sitaram Emani, MD^{†§}, Pedro del Nido, MD^{†§}, and Wayne Tworetzky, MD^{*‡}

- ★ Evaluated trends in prenatal diagnosis of TGA IVS over 20 year period (1992-2011)

- ★ N= 340
 - Overall 23.8% had prenatal diagnosis
 - 6% 1992-1995
 - 41% 2008-2011



[Birth Defects Res A Clin Mol Teratol](#). 2016 Mar;106(3):178-84. doi: 10.1002/bdra.23474. Epub 2015 Dec 21.

Impact of prenatal diagnosis on the outcome of patients with a transposition of great arteries: A 24-year population-based study.

[Debost-Legrand A](#)^{1,2}, [Ouchchane L](#)^{3,4}, [Francannet C](#)^{5,8}, [Goumy C](#)⁷, [Perthus I](#)^{2,5,6}, [Beaufrère AM](#)⁸, [Gallot D](#)^{9,10}, [Lemery D](#)^{2,9}, [Lusson JR](#)^{4,11}, [Laurichesse-Delmas H](#)^{2,6,9}.

- ★ Evaluated sensitivity of antenatal detection in 2 study periods (1988-1999 and 2000-2012)
 - Data from French population based birth defect registry

- ★ Overall simple TGA n=66, associated TGA n=28
- ★ 1988-1999 prenatal detection
 - 21.1% simple TGA, 4.8% associated TGA

- ★ 2000-2012 prenatal detection
 - 100% simple TGA, 33.3% associated TGA

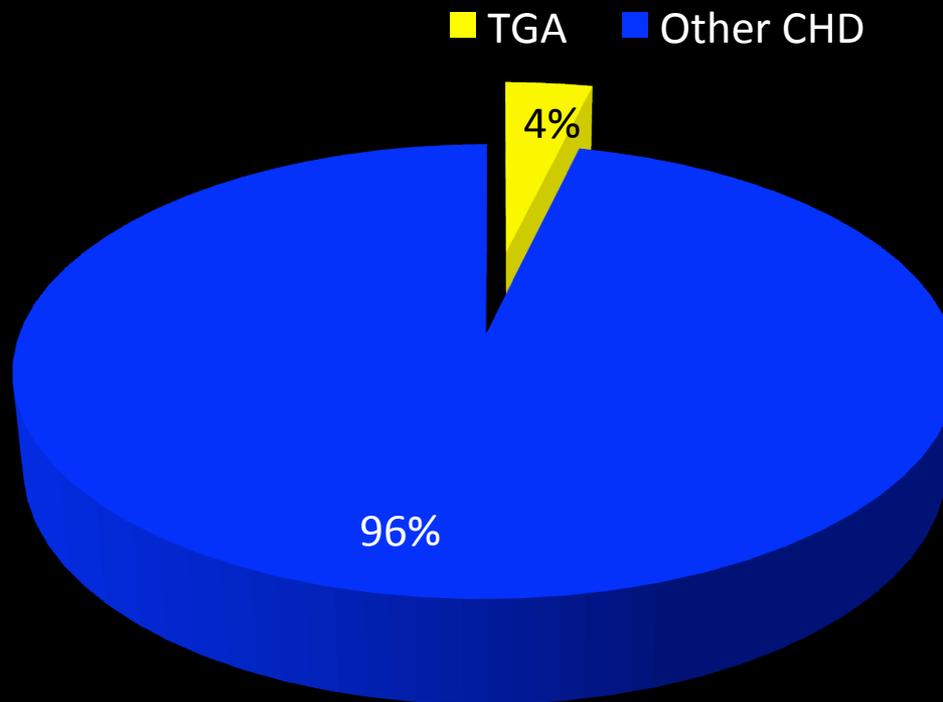
Evelina London



Transposition of the great arteries in fetal series

TGA simple and TGA VSD

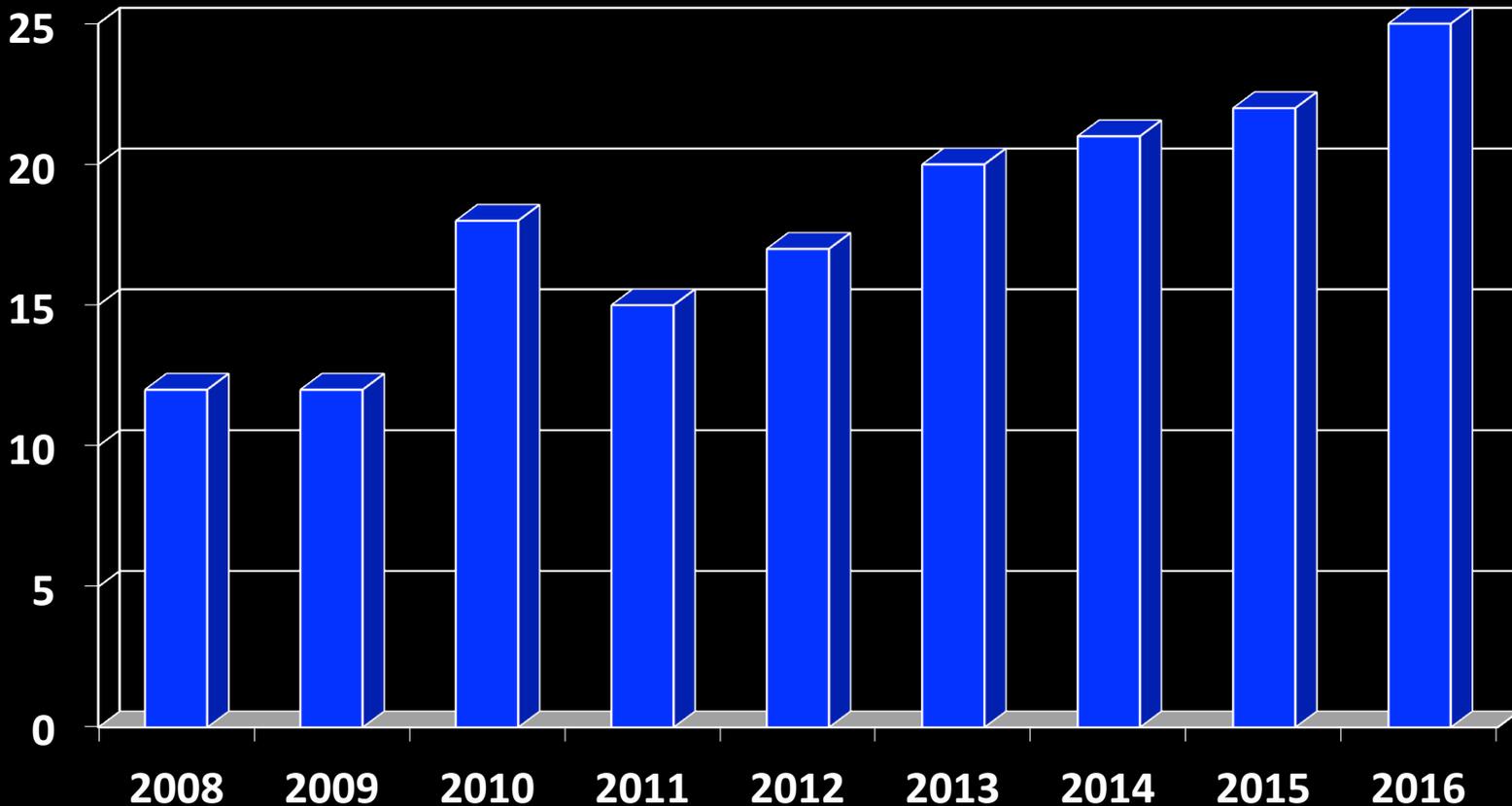
n=>5000



4-7% of postnatal CHD



Evelina Children's Hospital Numbers TGA diagnosed prenatally





NHS Fetal Anomaly Screening Programme (FASP)

18+0 to 20+6 Weeks Fetal Anomaly Scan

National Standards and Guidance for England

★ All pregnant women offered anomaly scan 18-21 weeks

★ Cardiac views must be included

- abdominal situs
- 4 chambers
- aorta/left ventricular outflow
- pulmonary artery/right ventricular outflow
- 3 vessel tracheal view



Teaching Obstetric Sonographers

- ★ First vessel seen when moving cranially from four chamber view is aorta
- ★ Scan further cranially to view PA and three vessel view
- ★ Aorta and PA cross over at origin





Teaching Obstetric Sonographers



Normal 4
chamber view



First vessel seen
arising from heart
is PA (branching
artery)

PA from LV

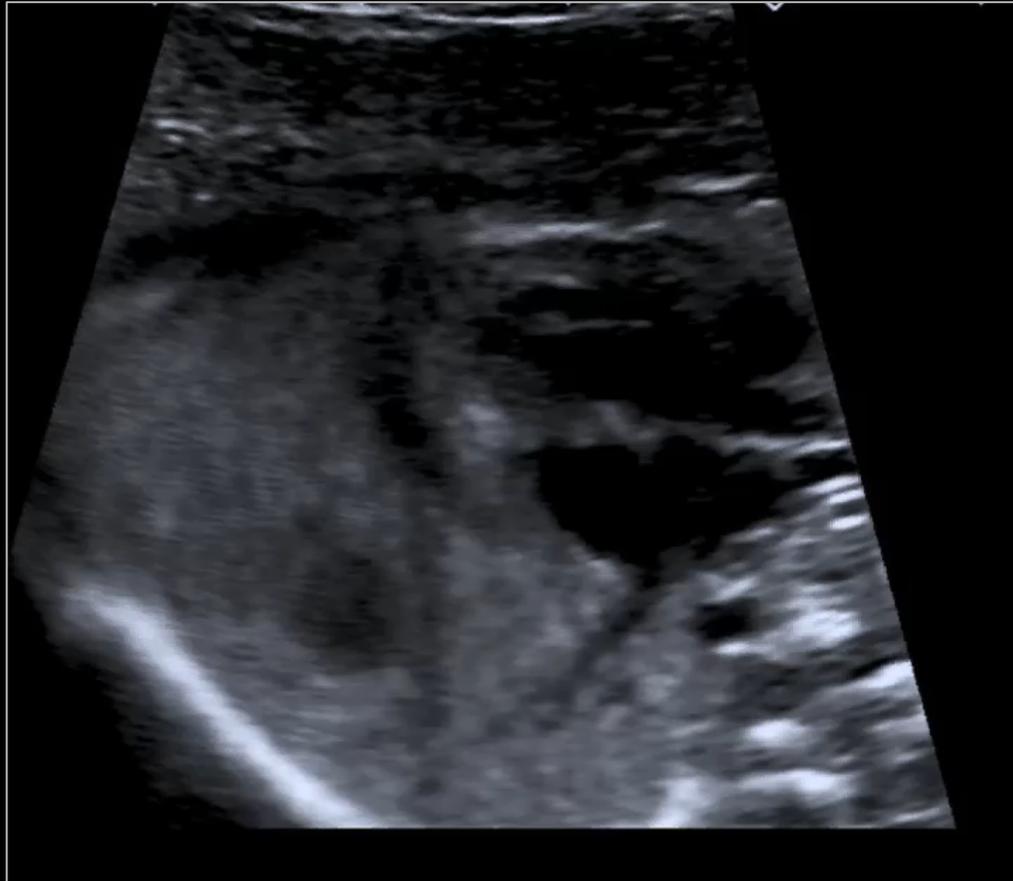


Aorta from RV
Aorta arises more
superiorly to PA

Prenatal detection TGA



- Teaching Obstetric Sonographers
- Loss of cross over great arteries
 - Parallel great arteries

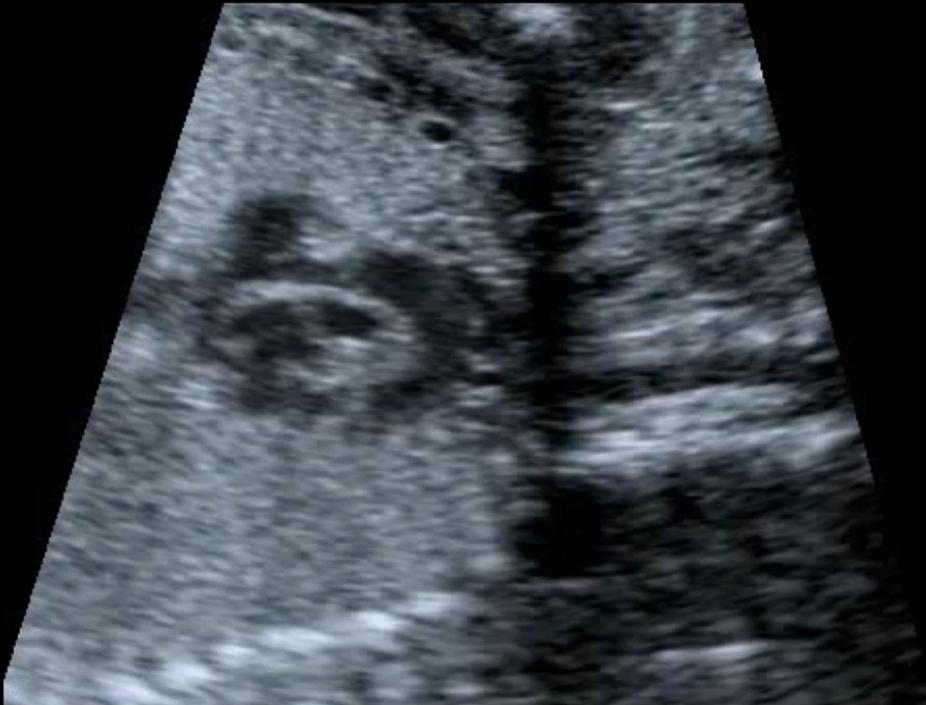


Prenatal detection TGA



Teaching Obstetric Sonographers
Wide sweeping aortic arch

Normal



TGA



Prenatal detection TGA



Teaching Obstetric Sonographers
Abnormal 3 vessel tracheal view

Normal



TGA





Do we need to know before birth?

- ★ Parental choices at time of diagnosis
- ★ Parents prepared for likely management after birth
 - Possible atrial septostomy
 - Arterial Switch
 - Other surgical management in more complex forms
 - Meet with surgeons and other team members
- ★ Plan optimal perinatal management
 - Site of delivery
 - Timing of delivery
- ★ Predict cases that may require early intervention
- ★ Other considerations



Pitfalls in diagnosis

- ★ Unable to predict coronary artery anatomy
- ★ Predicting which babies will require emergency atrial septostomy after birth is possible but can be challenging



Predicting which babies with TGA will require immediate treatment after birth



Sensitivity and Specificity of Prenatal Features of
Physiological Shunts to Predict Neonatal Clinical Status
in Transposition of the Great Arteries
Jouannic et al, *Circulation*. 2004;110:1743-1746

- ★ Restriction of the Foramen Ovale and of the Ductus Arteriosus has a high specificity to predict the need for emergency neonatal care in fetuses with TGA, but the sensitivity is too low to detect all high-risk fetuses



Fetal Predictors of Urgent Balloon Atrial Septostomy in Neonates with Complete Transposition

Rajesh Punn, MD, and Norman H. Silverman, MD, DSc (Med), FASE, Palo Alto, California

J Am Soc Echocardiogr 2011;24:425-30.

- ★ Hypermobile septum and reverse diastolic flow in duct help predict need for urgent balloon atrial septostomy

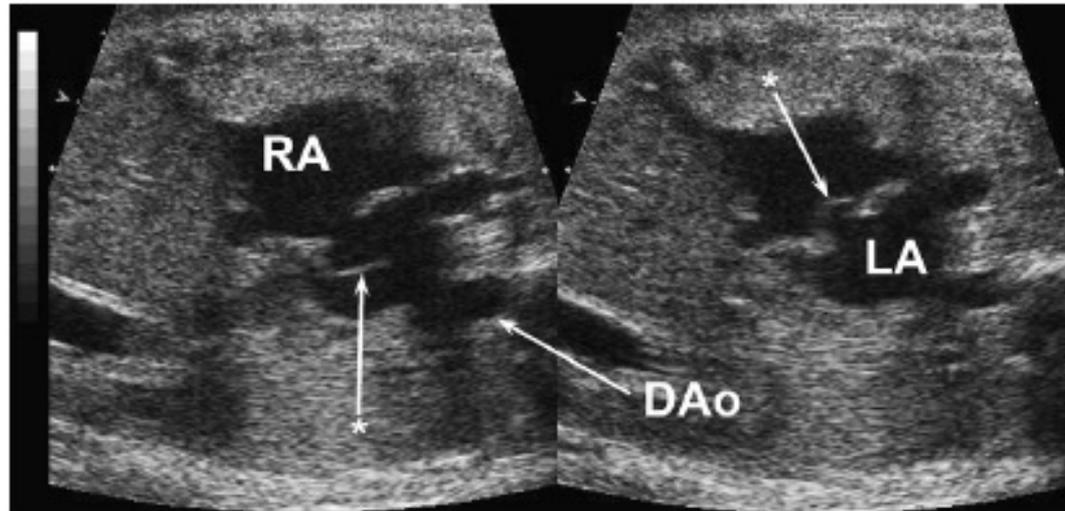


Figure 1 Hypermobile atrial septum. Batrial view demonstrating the septum primum flap (*) oscillating between the left and right atrium during various phases of the cardiac cycle (patient 7 in Table 1). See Video (📺) view video clip online). RA, Right atrium; LA, left atrium; DAo, descending aorta.

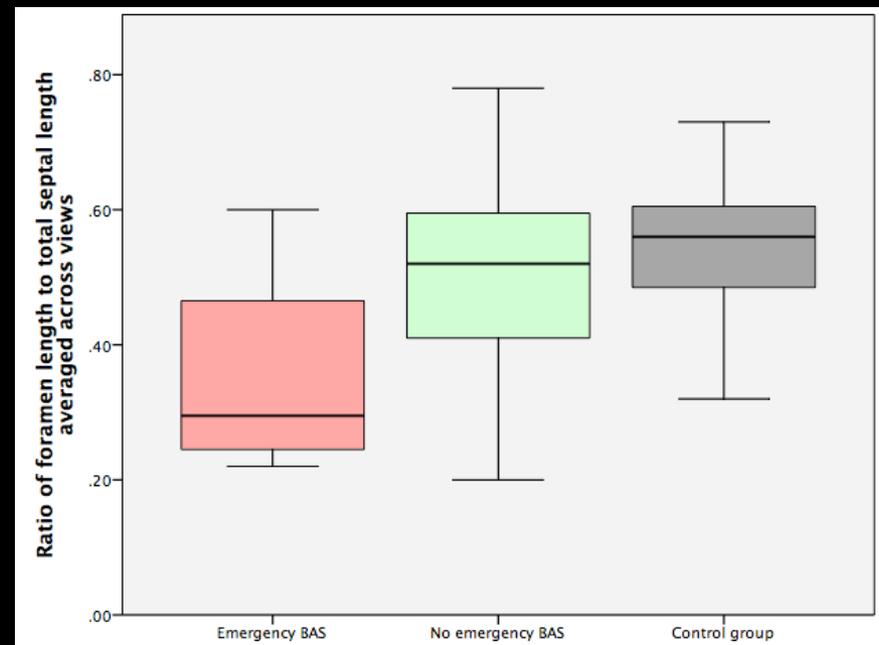


Vigneswaran TV et al, *Am J Cardiol* 2017

Usefulness of the Prenatal Echocardiogram in Fetuses With Isolated Transposition of the Great Arteries to Predict the Need for Balloon Atrial Septostomy

Likelihood of an emergency BAS is increased by FO:TSL <0.5 and a fixed appearance of the flap valve.

Hypermobile and/or aneurysmal atrial septum did not indicate inadequate postnatal mixing in this series.



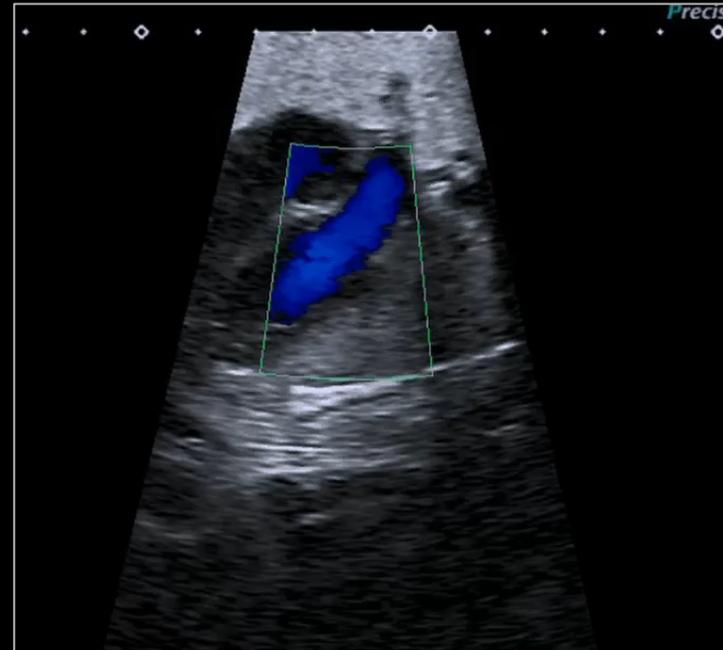


Atrial septum beginning to show signs restriction





Baby required emergency septostomy





Aneurysmal foramen flap common



Vigneswaran TV et al, *Am J Cardiol* 2017

An aneurysmal AS was identified in 31/40 cases of TGA

- seen more frequently in those cases who did not require an emergency BAS

An aneurysmal atrial septum and/or a hypermobile atrial septum were noted in normal fetuses (n=40) and those with TGA (n=40), with no significant difference in the frequencies between the groups



Things can change as pregnancy advances...
Baby required emergency septostomy

20 weeks



35 weeks



Importance of assessment in late pregnancy



Detection of Transposition of the Great Arteries in Fetuses Reduces Neonatal Morbidity and Mortality

Bonnet et al, *Circulation*. 1999

- ★ Compared preoperative and postoperative morbidity & mortality TGA over 10 years
 - 68 neonates with prenatal diagnosis
 - 250 neonates with a postnatal diagnosis
- ★ Clinical condition at arrival, including metabolic acidosis and multi-organ failure, worse in the neonatal group ($P < 0.01$)
- ★ Preoperative mortality
 - 15 of 250 in the neonatal group
 - 0 of 68 in the prenatal group ($P < 0.05$)
- ★ Postoperative mortality
 - 20 of 235 in neonatal group
 - 0 of 68 in prenatal group ($P < 0.01$)



Prenatal detection of transposition of the great arteries
reduces mortality and morbidity

van Velzen et al, Ultrasound Obstet Gynecol, 2015

N=144 cases TGA

- 26.4% prenatal diagnosis
- ★ First-year mortality significantly lower in cases with prenatal diagnosis of TGA than in those without (0.0% vs 11.4%)
- ★ Pre-surgical mortality (4.9%) only occurred in undetected simple TGA cases
- ★ Closure of the duct before treatment, renal dysfunction and hypoxia occurred significantly more often in the group without a prenatal diagnosis



Prenatal diagnosis: impact on outcome

Prenatal diagnosis of transposition of the great arteries over a 20-year period: improved but imperfect

Escobar-Diaz MC et al Ultrasound Obstet Gynecol 2015

340 patients with TGA IVS

- 81 (23.8%) had prenatal diagnosis

- ★ Mortality rate not statistically significantly different between prenatally and postnatally diagnosed patients
- ★ Significant preoperative differences with regard to earlier balloon atrial septostomy and fewer neonates that required mechanical ventilation



Other important considerations

Congenital Heart Defects and Indices of Fetal Cerebral Growth
in a Nationwide Cohort of 924 422 Liveborn Infants

Matthieson NB et al, Circulation 2016

- ★ Neurodevelopmental disorders common and distressful comorbidities associated with CHD
- ★ Head circumference at birth (HC), a proxy for prenatal cerebral growth, is an established risk factor for neurodevelopmental disorders
- ★ Infants with transposition of the great arteries had smaller HC relative to birth weight



Other important considerations

Severity of Fetal Brain Abnormalities in Congenital Heart Disease in Relation to the Main Expected Pattern of in utero Brain Blood Supply

Masoller et al Fetal Diag Ther 2015

- ★ 58 fetuses with CHD divided into two functional classes and compared to 58 controls:
 - **class A** with an expected severe reduction in oxygenated brain blood supply (**left outflow tract obstruction and TGA**)
 - class B with theoretically near-normal or mildly impaired oxygenated brain blood supply (other CHD)
- ★ Both class A and B CHD fetuses had significant differences in head biometry, brain perfusion, cortical development and brain metabolism compared with controls
- ★ **However, there were signs of more severe brain alterations in type A CHD fetuses**