Prenatal Counseling for a 24-week GP fetus with severe APVS?

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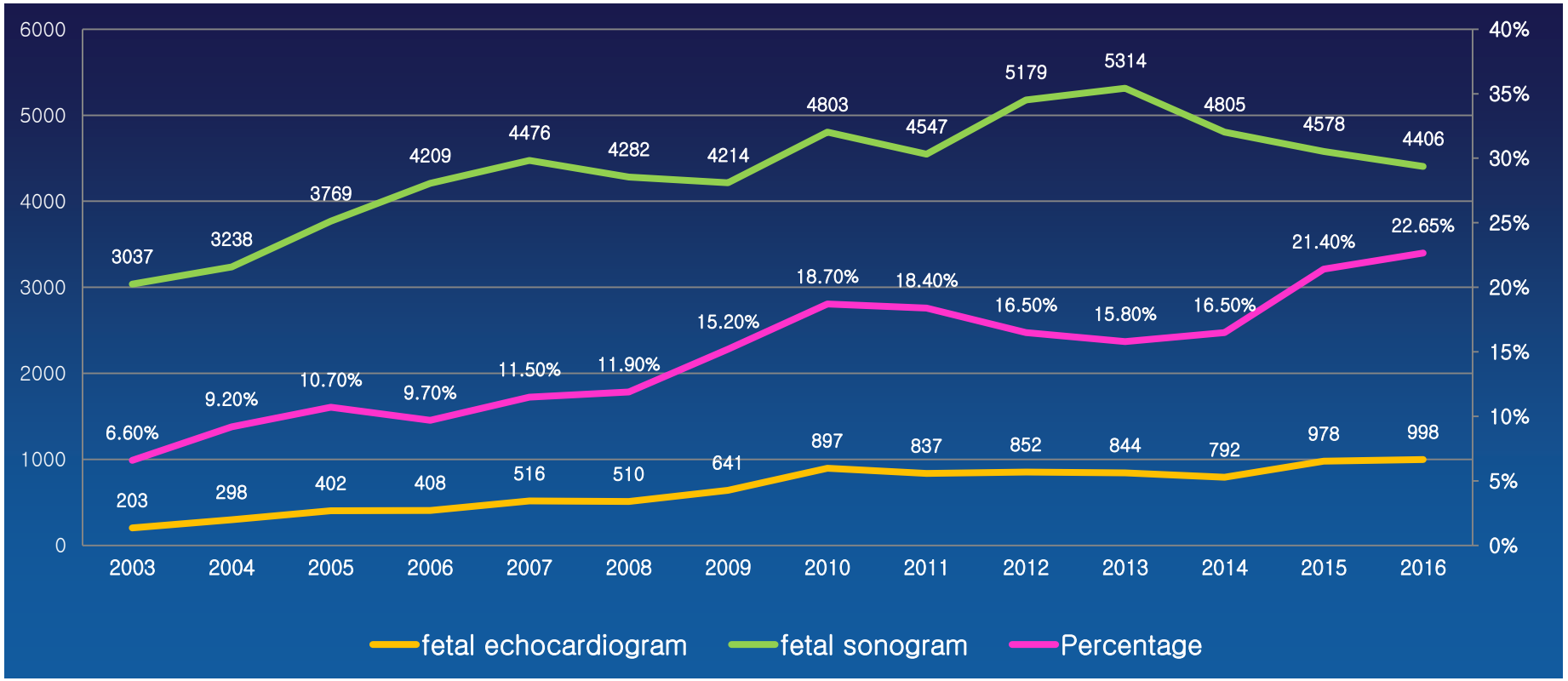
What would you do for a 24-week GP fetus with severe APVS?

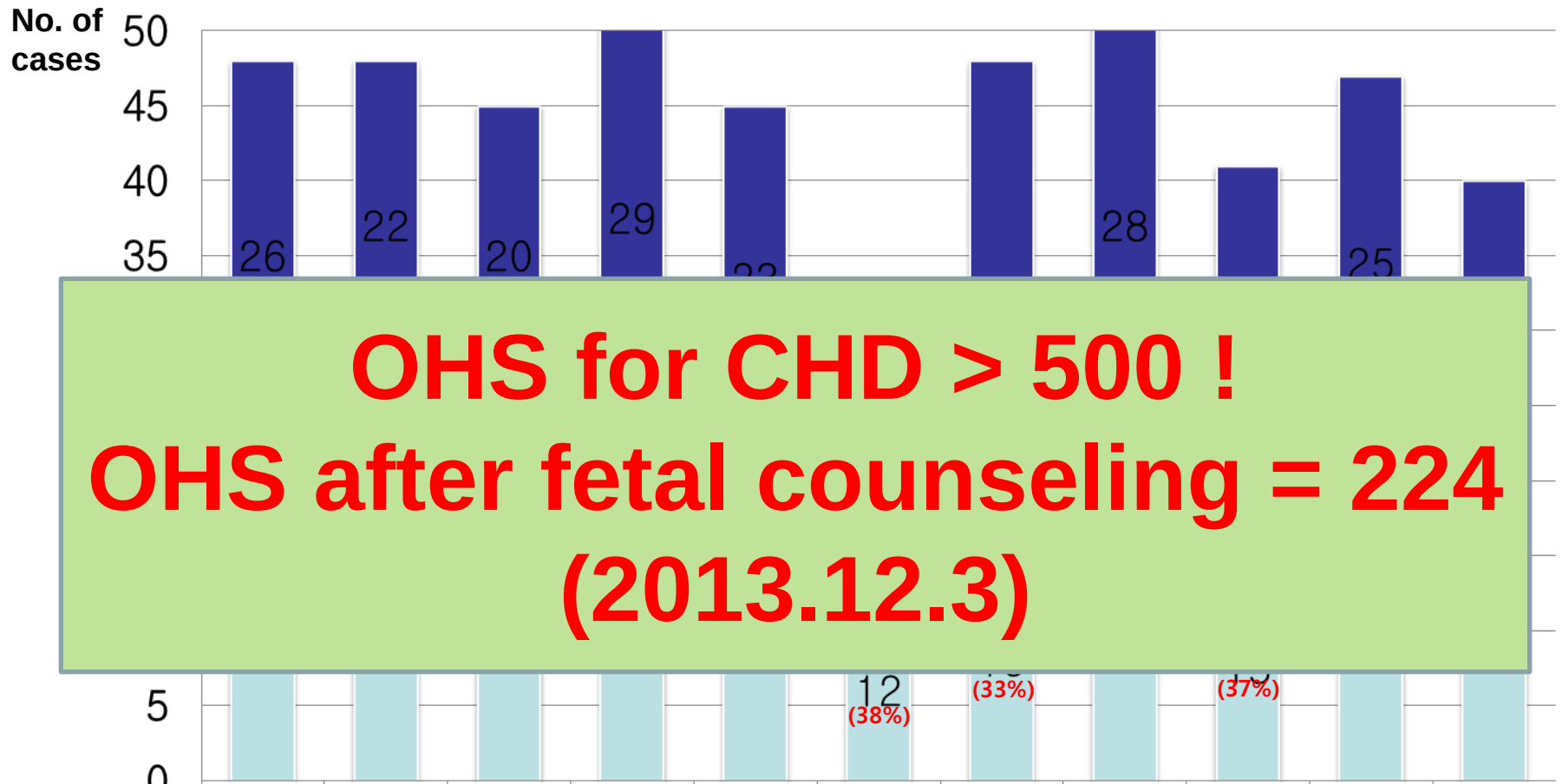
Get the pregnant mother to give birth to the baby!

AMC Experience of fetal echocardiography

Year	2003	2004	2005	2006	2007	2008	2009	2010	2011	2012	2013	2014	2015	2016
Fetal echocardiogram (A)	203	298	402	408	516	510	641	897	837	852	844	792	978	998
Fetal sonogram (B)	3037	3238	3769	4209	4476	4282	4214	4803	4547	5179	5314	4805	4578	4406
A / B x 100 (%)	6.6%	9.2%	10.7%	9.7%	11.5%	11.9%	15.2%	18.7%	18.4%	16.45%	15.8%	16.48%	21.36%	22.65%



Prevalence of fetal Dx among OHS cases (2013)

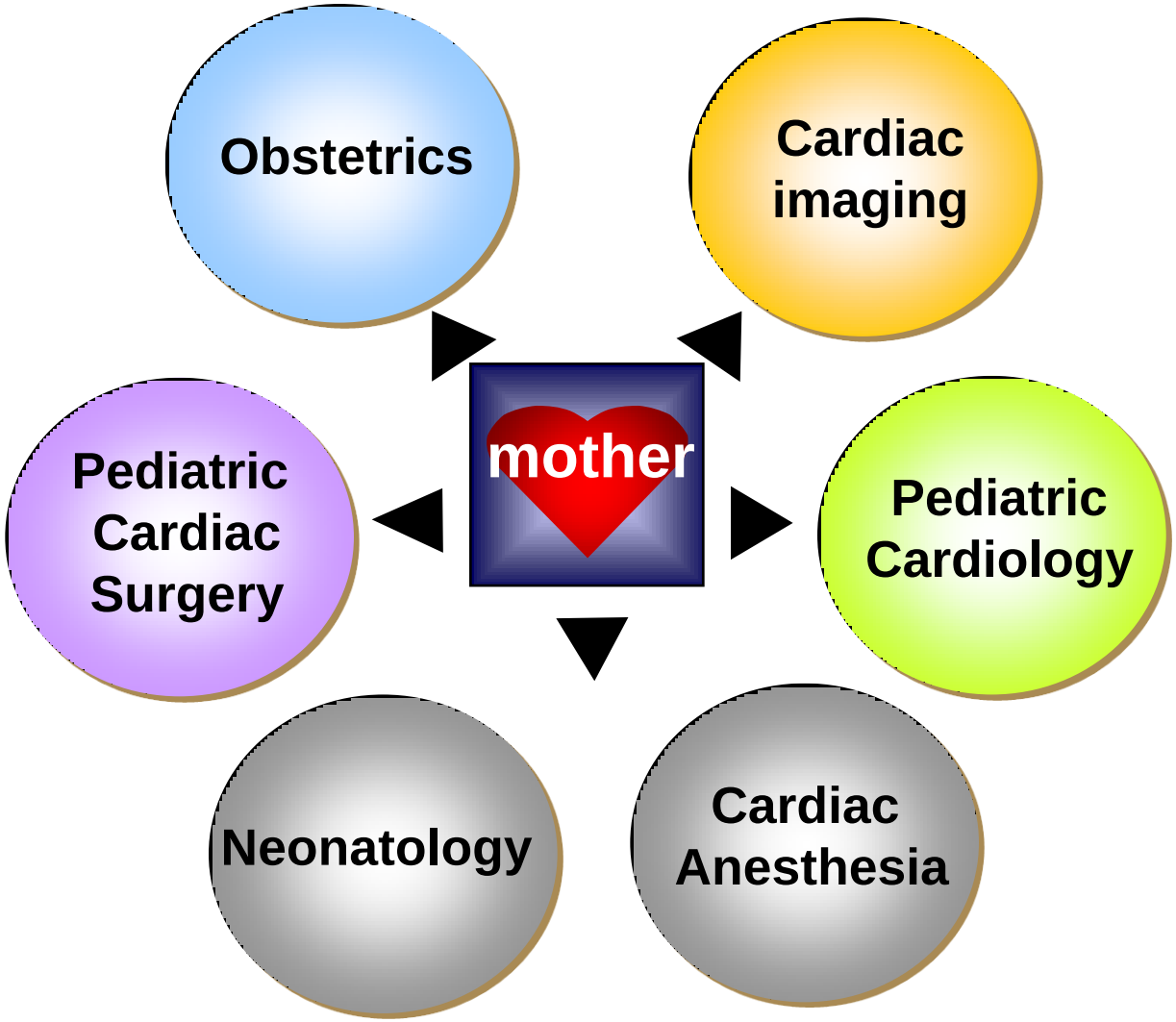


	1월	2월	3월	4월	5월	6월	7월	8월	9월	10월	11월
Fetal Dx (−)	26	22	20	29	23	20	32	28	26	25	20
fetal Dx (+)	22	26	25	23	22	12	16	23	15	22	20

OHS for CHD > 500 !

OHS after fetal counseling = 224 (2013.12.3)

Conventional Fetal Counseling



Mother to Doctors!

Multidisciplinary fetal counseling

Pediatric Cardiology

Pediatric Cardiac Surgery

Diagnositc imaging

NICU

PICU

Cardiac anesthesia

Doctors to mother!

AMC Fetal counseling flow

태아 심장 기형 의심

↓

태아 치료센터 전화예약
(TEL. 02-3010-3654)

↓

태아 치료센터로 산모 방문

↓

정밀 초음파를 통한
정확한 진단

산모 진료, 유전자 검사 시행

↓

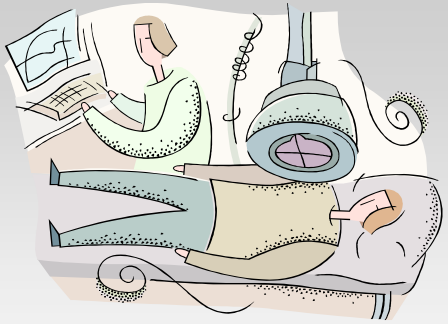
출생 후 치료 및 예후에 관한
상담 (통합진료)

↓

환아 출생 및 NICU 입원

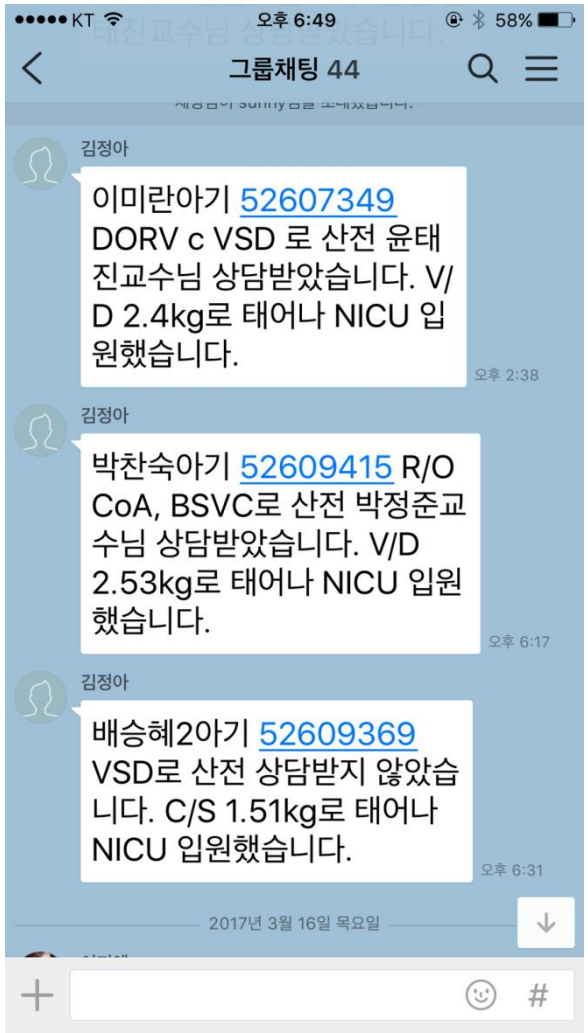
↓

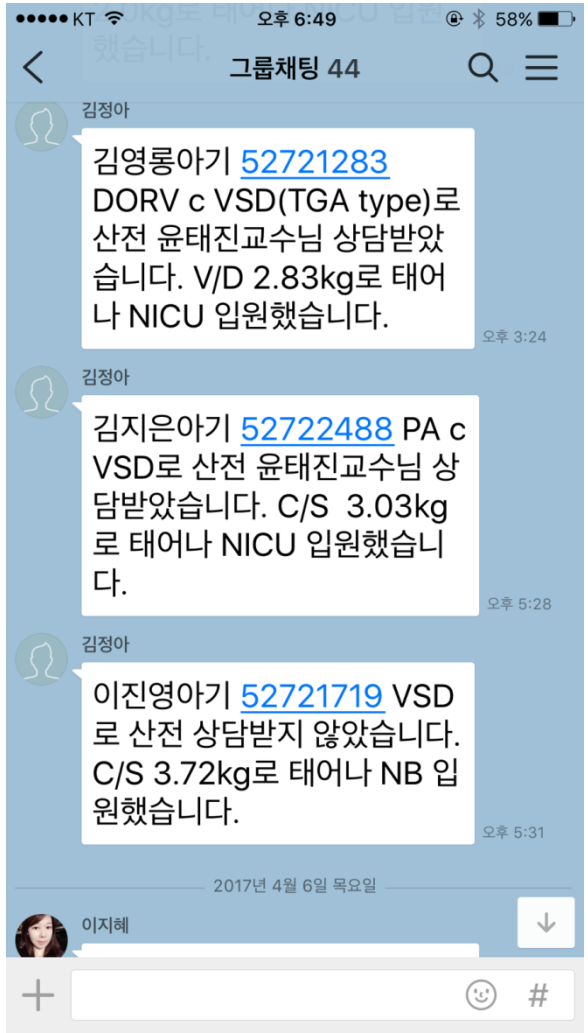
관련과 실시간 협진 시작

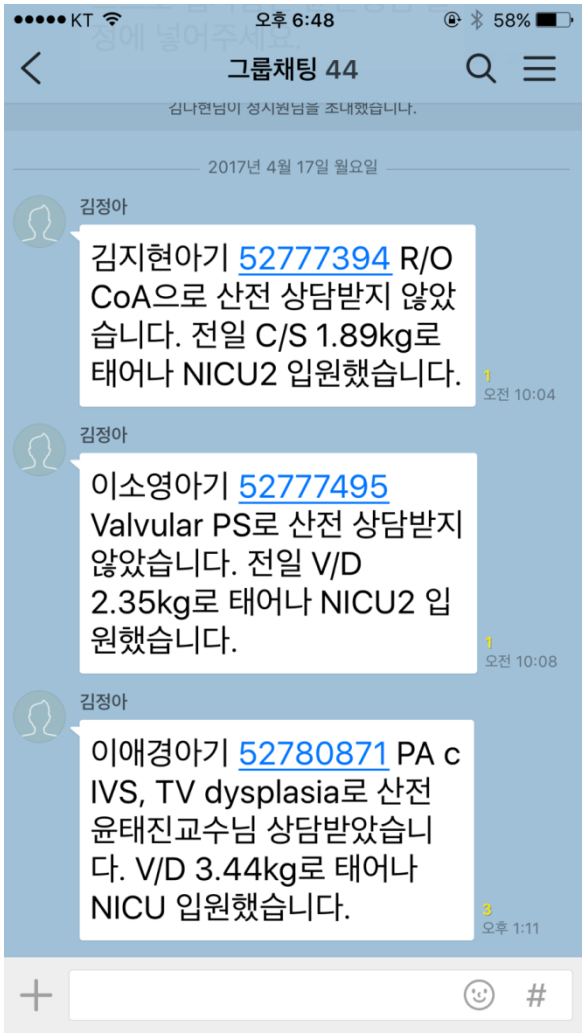


··전문 의료진의 상담 및
질병 & 치료에 대한
정보제공, 정서적 지지

Multidisciplinary patient care





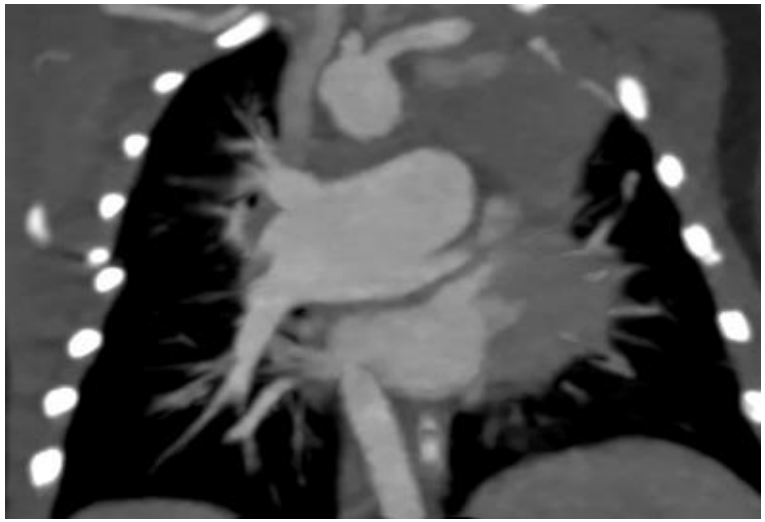


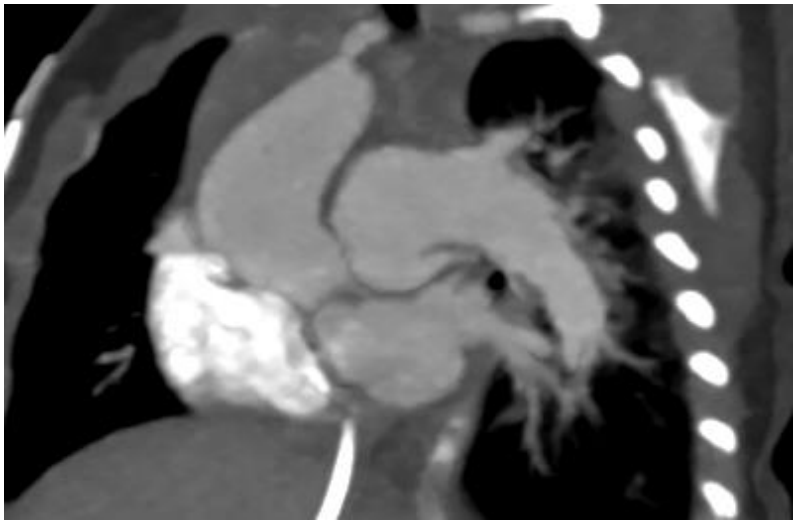
Multidisciplinary fetal counseling

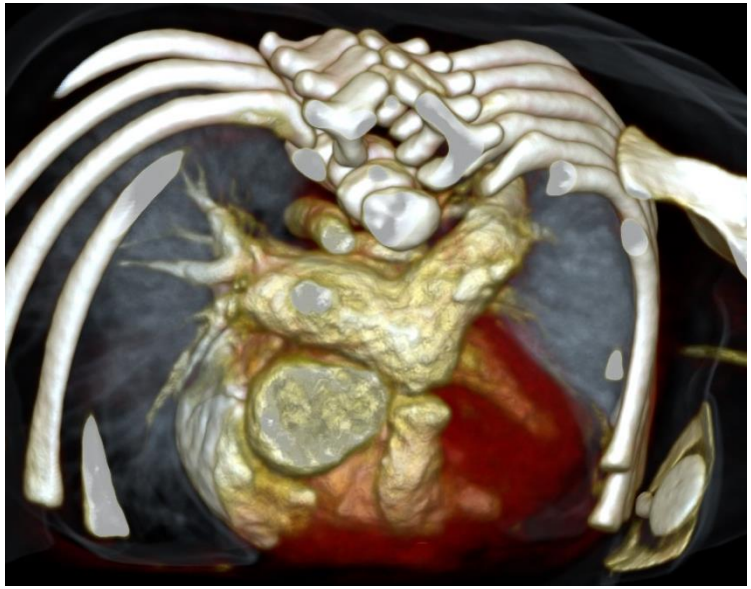
Case

- Prenatal counseling at GA 36^{+1} weeks
- Prenatal Diagnosis: ToF with APVS
- Delivery at GA 38^{+3} weeks, 3.6 kg, A/S 9
- SaO2: 85%
- Preoperative ventilatory support (-)
- Preoperative inotropic support (-)
- Elective Rastelli op at age 16 days using 10 mm hand-made Gore-tex valved conduit
- RV-PA conduit change at age 24 months
- Currently 3 years old, asymptomatic

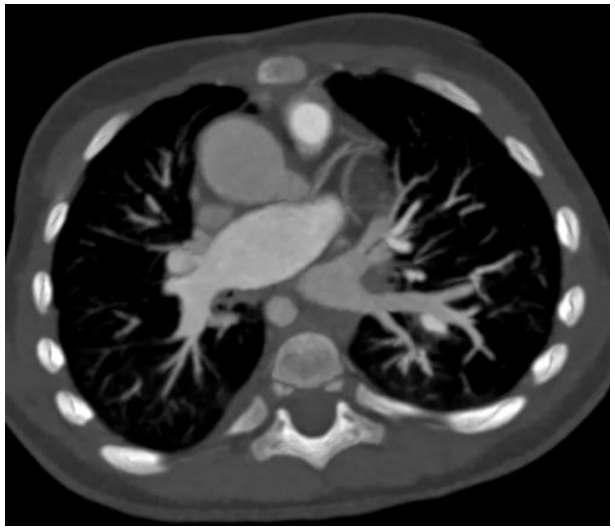
Preoperative CT

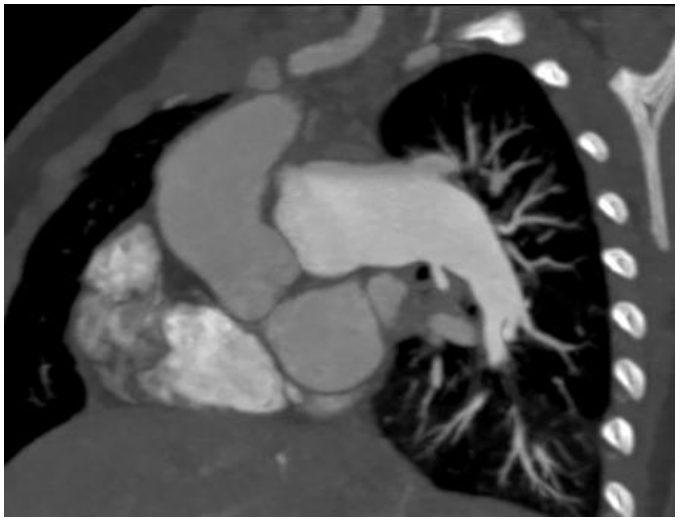


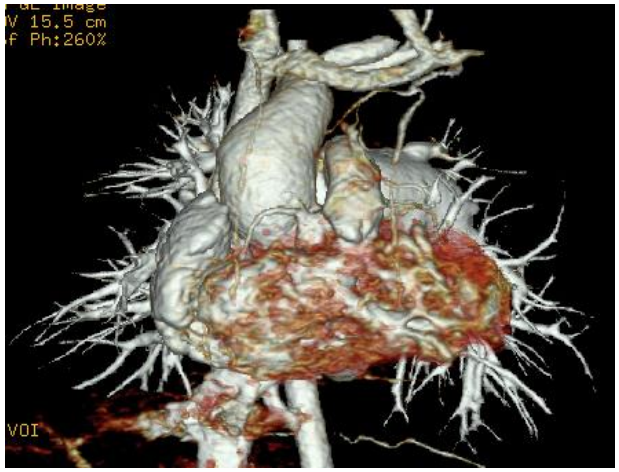


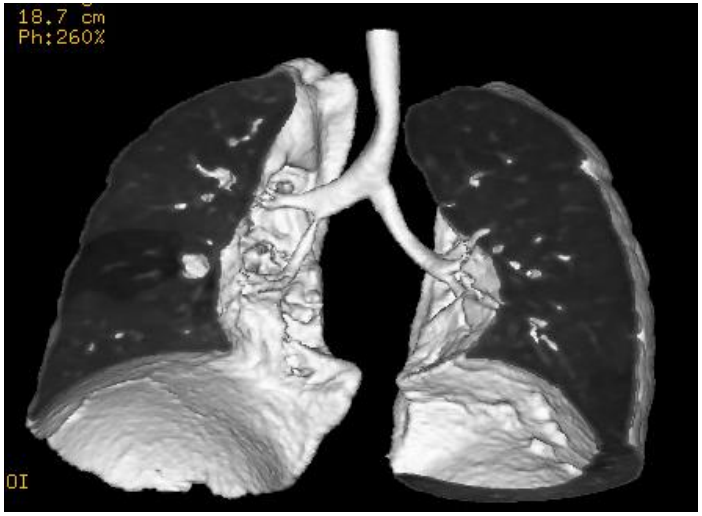


Postoperative CT

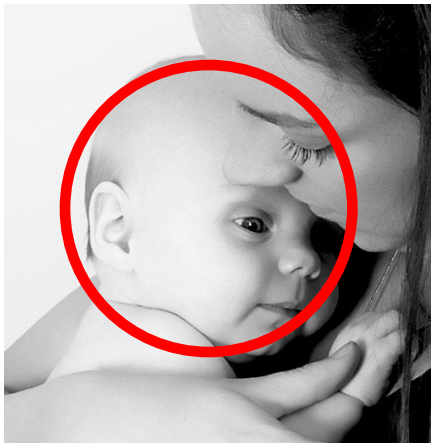








Prenatal awareness of CHD



Mothers
Psychological trauma

Babies
Better chance for better care

Appropriate fetal counseling is important!

Prenatal diagnosis of CHD

- **Prenatal awareness of the disease by the medical staff:**

  → Rapport establishment by appropriate counseling

- **Prenatal preparation for specific care for the baby**
- **Delivery in the hospital where the baby shall be treated**
- **Immediate postnatal NICU care.**

Should lead to a better outcome

Prenatal diagnosis of CHD

Improved Surgical Hypoplas

Wayne Tworetzky, MD; Doff B. McE
Frank L. Ha

Background—Hypoplastic left heart syndrom
improve surgical outcome.

Methods and Results—We reviewed patients
of prenatal diagnosis on preoperative clinic
Of 88 patients, 33 were diagnosed prenatall
and pregnancy was terminated in 11. Of 2
and parents elected to forego treatment in 8
because of parental decisions or clinical
surgery than patients diagnosed after bi
nonintervention. Among patients who und
diagnosis and underwent surgery survived,
Patients diagnosed prenatally had a low
(P=0.001), and ventricular dysfunction
medications or bicarbonate (P=0.005). Prec
(P=0.009), more severe acidosis (P=0.03
dysfunction (P=0.05).

Conclusions—Prenatal diagnosis of HLHS w
survival after first-stage palliation in com
1273.)

Key Words: prenatal diag

Heart disease

Prenatal diagnosis of structural heart disease: does it make a difference to survival?

I D Sullivan

Is there a survival advantage conferred by the prenatal diagnosis of congenital heart disease?

There are two potential advantages of mid trimester diagnosis of fetal heart disease. Perinatal management in an environment where the appropriate expertise is available and prepared might result in improved outcomes after intervention in the newborn period in situations where this is required. Alternatively, prenatal diagnosis of congenital heart disease allows consideration of termination of pregnancy. Termination of pregnancy for fetal anomaly is an emotive subject and has a wide range of acceptability in different societal contexts. Nearly 20% of all pregnancies in the UK were terminated for social indications in 1994 whereas the proportion in which the indication for termination was detection of fetal cardiac abnormality was about 0.02%.[1]

SURVIVAL ADVANTAGE?

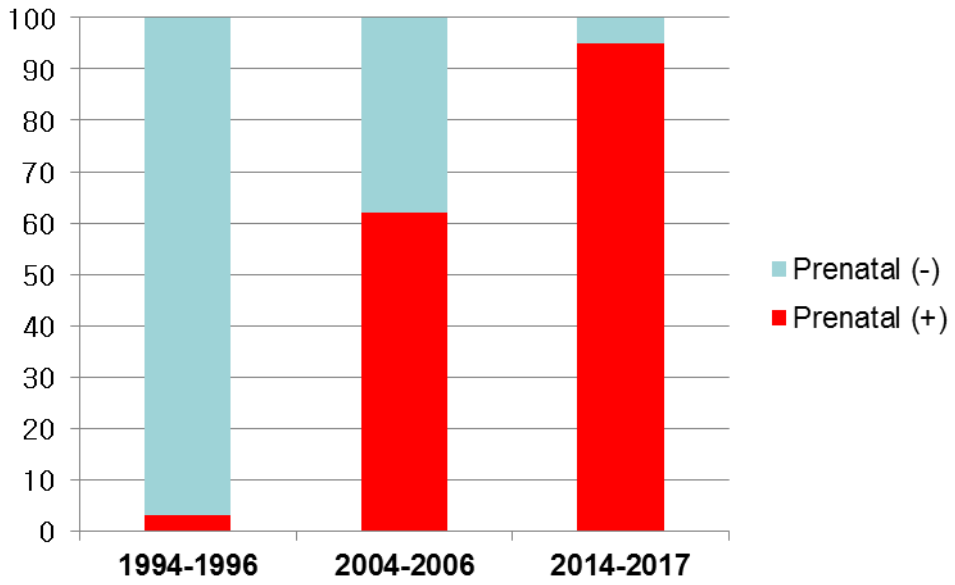
If there is a survival advantage conferred by the prenatal diagnosis of congenital postnatal survival for live born infants with pulmonary atresia and intact ventricular septum was similar regardless of the timing of diagnosis in a large population based study.[4] The hypothesis that costs and duration of initial hospitalisation and survival to discharge home would be improved by prenatal diagnosis was examined in a cohort analysis of neonates with structural heart disease in the absence of other life threatening conditions.[5] There was no difference in survival among those requiring palliative surgery because of a functionally single ventricle. Moreover, the costs and duration of hospitalisation were greater in the group diagnosed prenatally, regardless of whether or not surgery was performed postnatally. The results were similar when the analysis was limited to infants whose survival was dependent on continued patency of the arterial duct.

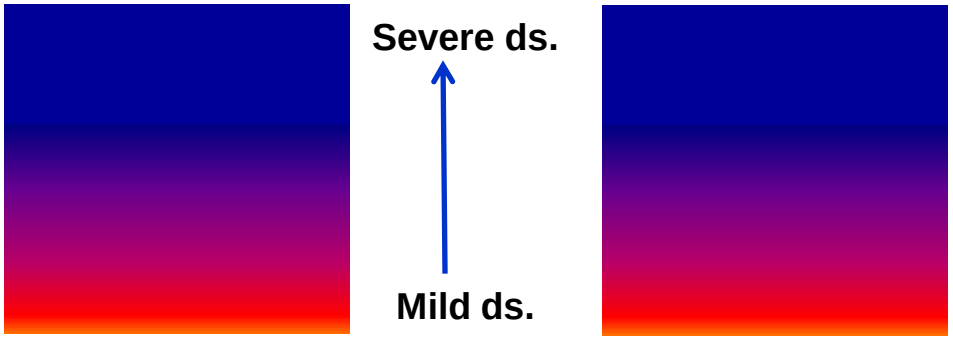
to newborn infants in whom the same cardiac diagnosis was known prenatally. These factors may also explain why other studies of surgical patients have failed to demonstrate improved postoperative survival after prenatal diagnosis of hypoplastic left heart.[9–11] A recent report indicating improved hospital survival after initial staged palliative surgery for hypoplastic left heart following prenatal diagnosis[12] is more encouraging, but may not be directly comparable with previous data[6 8] as infants with birth weight less than 2 kg or serious extracardiac abnormality were excluded. In a report from a single surgical institution it is also difficult to exclude a selection bias in referral of prenatally diagnosed cases of hypoplastic left heart perceived to be good risk surgical candidates. Population based data may be better able to address this question.[1]

None of the studies cited above was designed to detect mortality resulting from neonatal death with congenital heart disease not diagnosed during life. Detection of this occult cardiac mortality allowed demonstration of improved survival after prenatal diagnosis of transposition of the great arteries in Paris.[13] By analogy, four critically ill newborn infants with respiratory failure and undiagnosed transposition of the great arteries have been referred to Great Ormond Street Hospital for ECMO (extracorporeal membrane oxygenation) support since 1994. However, the impact of prenatal diagnosis on improved postnatal survival will not be great in population terms while as little as 3% of all

Prenatal diagnosis of CHD in AMC

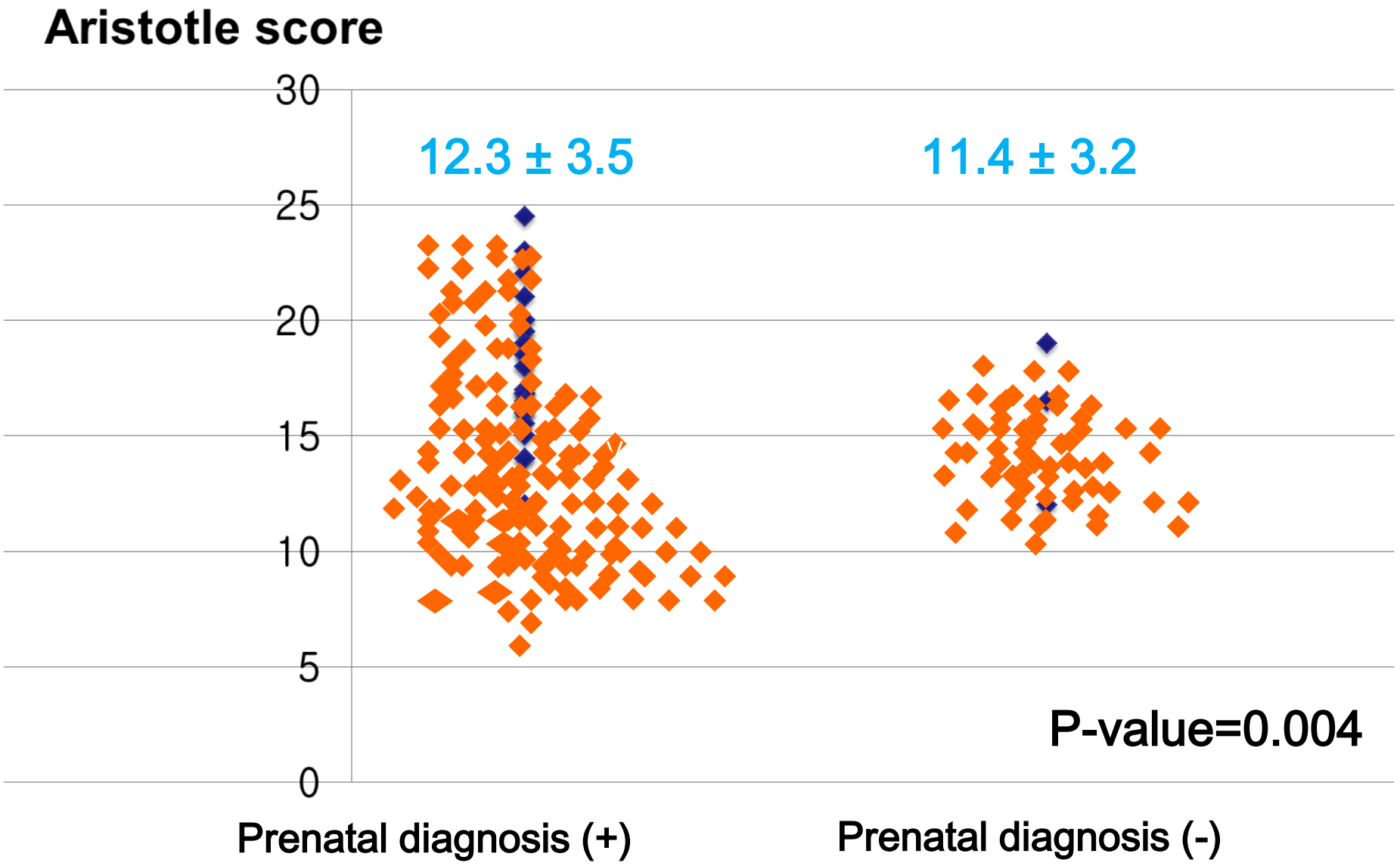
NICU admission of the babies with CHD





We are facing more and more difficult cases !

Disease complexity



What would you do for a 24-week GP fetus with severe APVS?

What happened to the all babies diagnosed as having APVS prenatally?

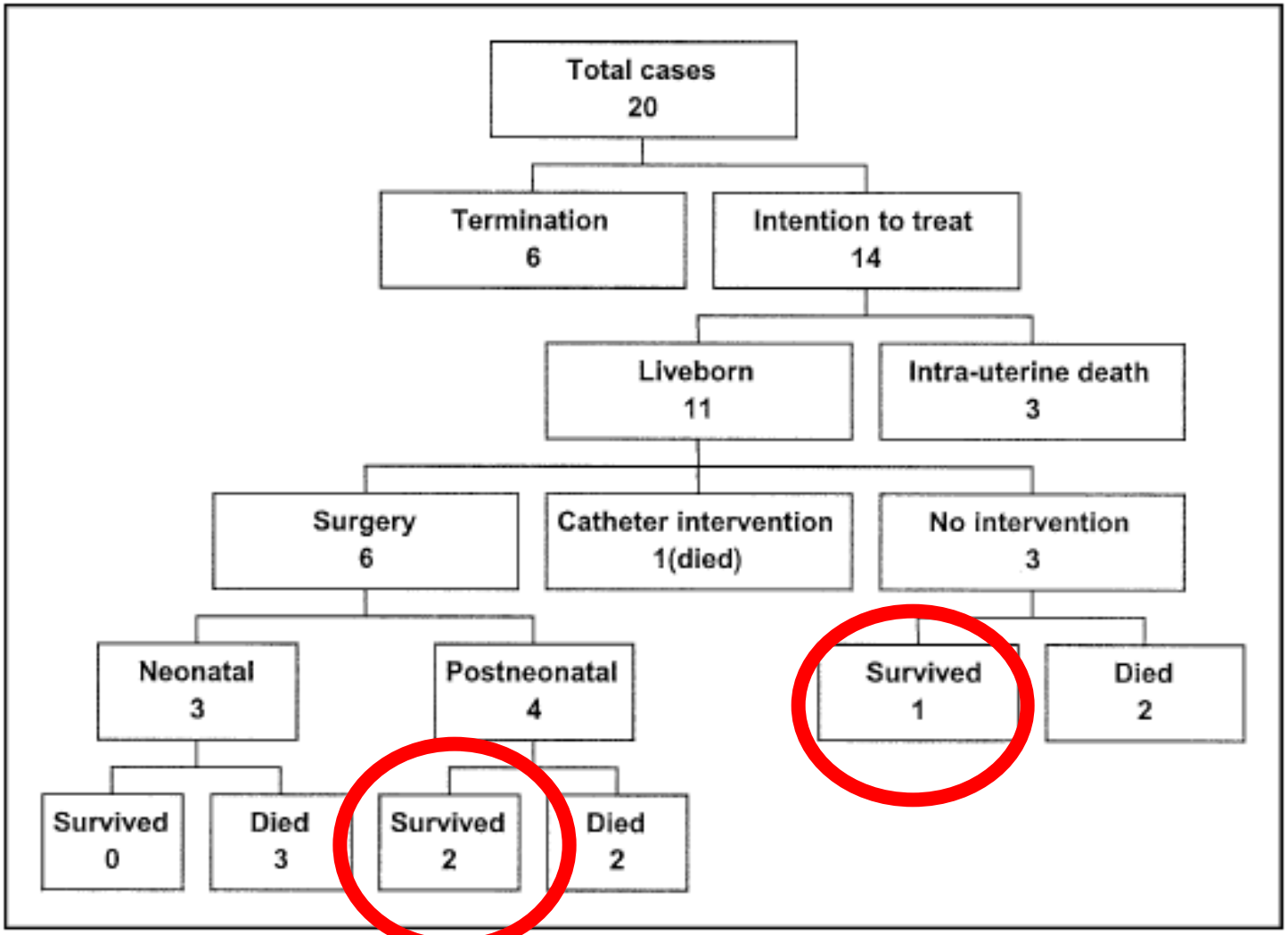
Prenatal Diagnosis by Echocardiogram and Outcome of Absent Pulmonary Valve Syndrome

Reza S. Razavi, MRCP, Gurleen K. Sharland, MD, and John M. Simpson, MD

We reported echocardiographic findings and outcomes of fetuses with absent pulmonary valve syndrome diagnosed during fetal life. Cases were identified from a prospectively acquired computerized database of 18,308 pregnancies referred to a fetal cardiology center between January 1988 and July 2000. Twenty fetuses were identified with a median gestation of 23 weeks (range 18 to 36) at presentation. In 18 cases (90%), there was an associated ventricular septal defect. Eighteen cases (90%) had branch pulmonary artery diameters above the normal range. In four cases (20%), an arterial duct was present. A chromosome 22q11 deletion was identified in 2 of 9 cases (22%) in which this deletion was sought. There were 6 terminations of pregnancy (30%), 3 intrauterine deaths (15%), 5 neonatal deaths (25%), 3 infant deaths (15%), and 3 patients who did not die (15%). Ten of the 11 "liveborn" infants required early ventilation. The outcome of absent pulmonary valve symdrome diagnosed prenatally appears poor. The high morbidity and mortality is due to both cardiac disease and associated bronchomalacia. ©2003 by Excerpta Medica, Inc.

(Am J Cardiol 2003;91:429–432)

Prenatal Diagnosis by Echocardiogram and Outcome of Absent Pulmonary Valve Syndrome



Prenatal Diagnosis by Echocardiogram and Outcome of Absent Pulmonary Valve Syndrome

TABLE 1 Details of Fetuses With Absent Pulmonary Valve Syndrome (APVS)

Case No.	Gestation (wk)	DA	VSD	Karyotype	Surgery, Age at Surgery	Outcome
1	23	0	+	Normal	+, 1 yr	Alive
2	23	0	+	Normal	0	Alive
3	25	0	+	Normal	+, 2 yrs	Alive
4	27	0	+	22q11 deletion	+, 2 wk	InfD
5	36	0	+	Normal	+, 3 mo	InfD
6	32	0	+	Normal	+, 9 wk	InfD
7	20	0	+	Normal	+, 19 d	NND
8	20	0	+	22q11 deletion	+, 5 d	NND
9	34	0	+	Normal	0	NND
10	25	0	+	Failed	—	IUD
11	21	0	+	Normal	—	IUD
12	20	0	+	—	—	TOP
13	25	0	+	—	—	TOP
14	18	0	+	—	—	TOP
15	20	0	+	—	—	TOP
16	19	0	+	—	—	TOP
17	31	+	+	Normal	Catheter, 4 d	NND
18	20	+	+	—	—	IUD
19	26	+	0	—	0	NND
20	19	+	0	Normal	—	TOP

DA = ductus arteriosus; InfD = infant death; IUD = intrauterine death; NND = neonatal death; TOP = termination of pregnancy; VSD = ventricular septal defect.

Prenatal Diagnosis by Echocardiogram and Outcome of Absent Pulmonary Valve Syndrome

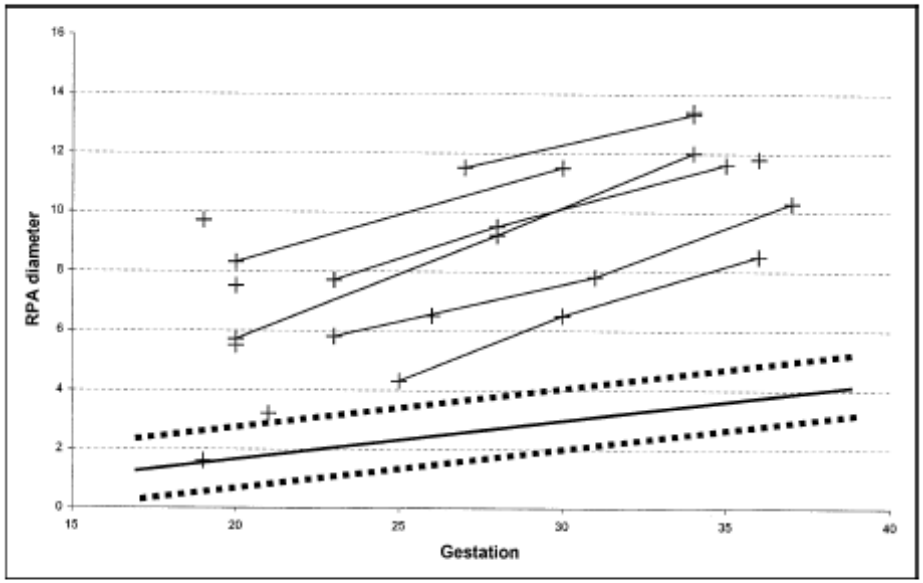


FIGURE 2. Right pulmonary artery (RPA) measurements plotted against gestational ages are shown. The fifth and ninety-fifth percentiles are indicated by the *lower and upper dashed lines,* respectively.[19]

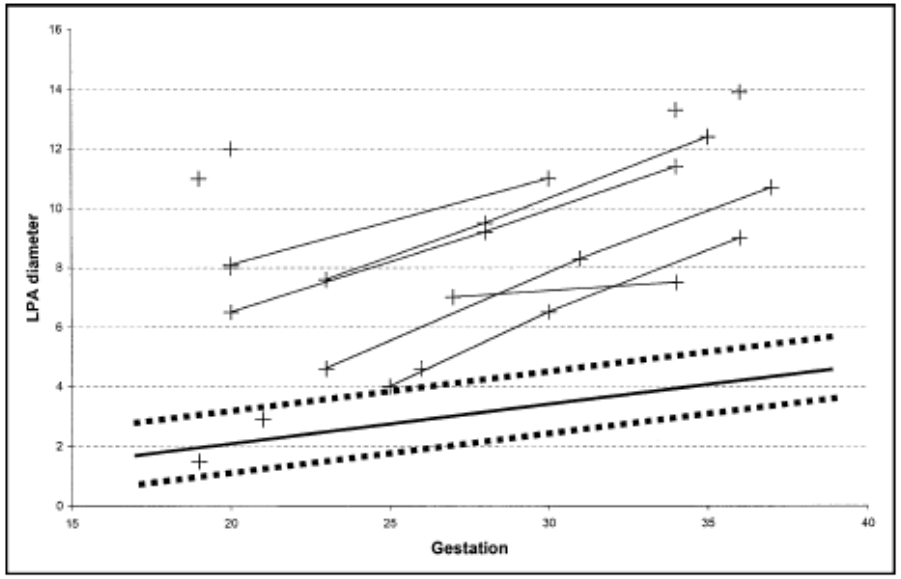
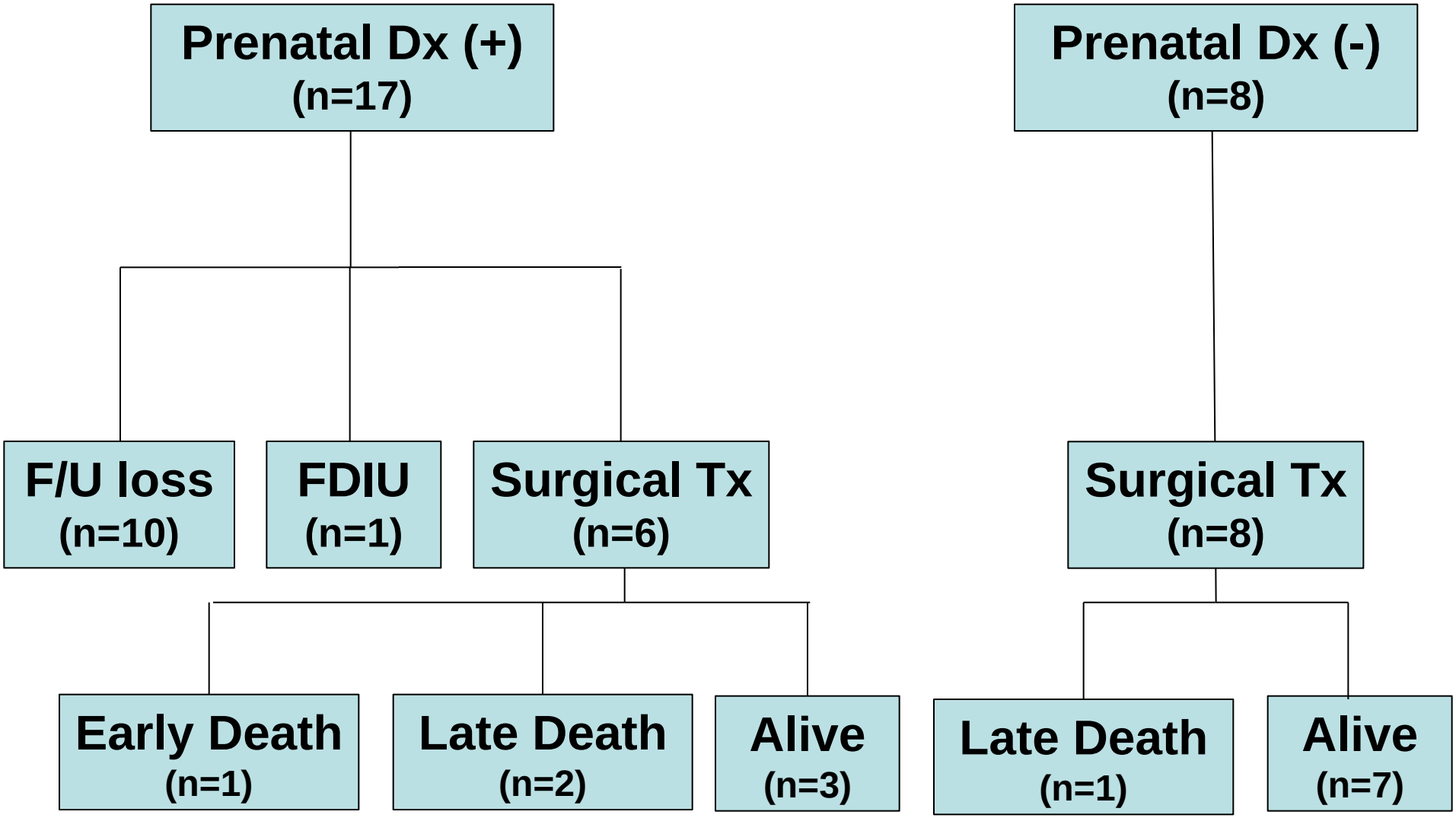


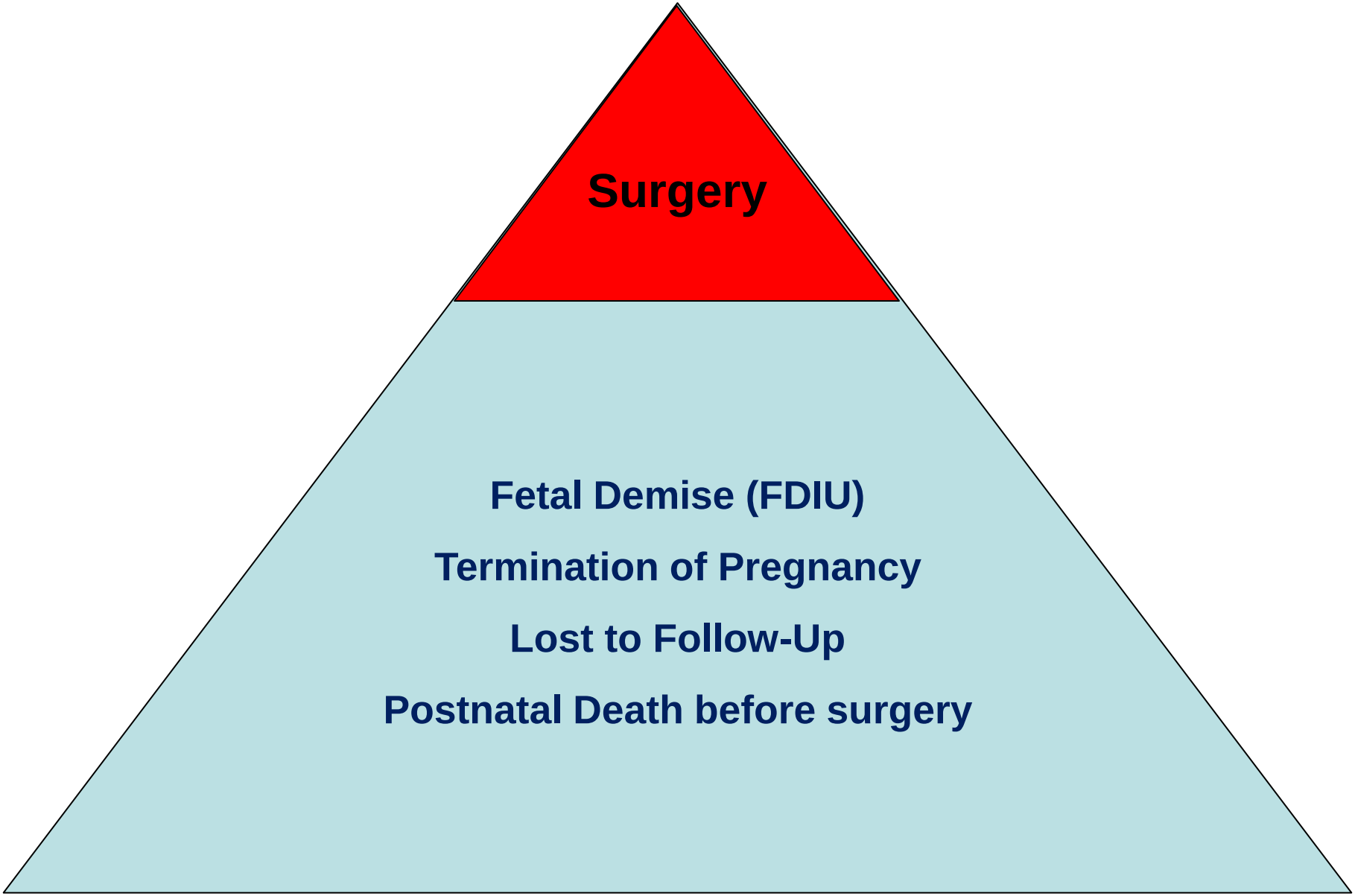
FIGURE 3. Left pulmonary artery (LPA) measurements plotted against gestational ages are shown. The fifth and ninety-fifth percentiles are indicated by the *lower and upper dashed lines,* respectively.[19]

APVS (AMC experience: 2002-2016)

No	APVS category	Prenatal Dx	Op age (days)	Op name	Outcome
1	With IVS	+			FDIU
2	With ToF	+			F/U loss
3	With ToF	+			F/U loss
4	With ToF	+			F/U loss
5	With ToF	+			F/U loss
6	FSV	+			F/U loss
7	With ToF	+			F/U loss
8	With ToF	+			F/U loss
9	With ToF	+			F/U loss
10	With IVS	+			F/U loss
11	With IVS	+			F/U loss
12	With IVS	-	9	PDA ligation / division	alive
13	With IVS	-	16	RPA anterior translocation	LD
14	With ToF	+	31	TOF total correction	alive
15	With ToF	+	20	TOF total correction	LD
16	With ToF	+	16	Rastelli with valved conduit	alive
17	With ToF	+	44	Rastelli with valved conduit	alive
18	FSV	+	40	Central shunt	ED
19	With ToF	+	38	Rastelli with valved conduit	LD
20	With ToF	-	414	TOF total correction	alive
21	With ToF	-	125	TOF total correction	alive
22	With ToF	-	141	TOF total correction	alive
23	With ToF	-	80	TOF total correction	alive
24	With ToF	-	34	Rastelli with valved conduit	alive
25	With ToF	-	77	Rastelli with valved conduit	alive

APVS (AMC experience: 2002-2016)




Surgery
Fetal Demise (FDIU)
Termination of Pregnancy
Lost to Follow-Up
Postnatal Death before surgery

What would you do for a 24-week GP fetus with severe APVS?

~~Get the pregnant mother to give birth to the baby!~~

Give the pregnant mother precise information!

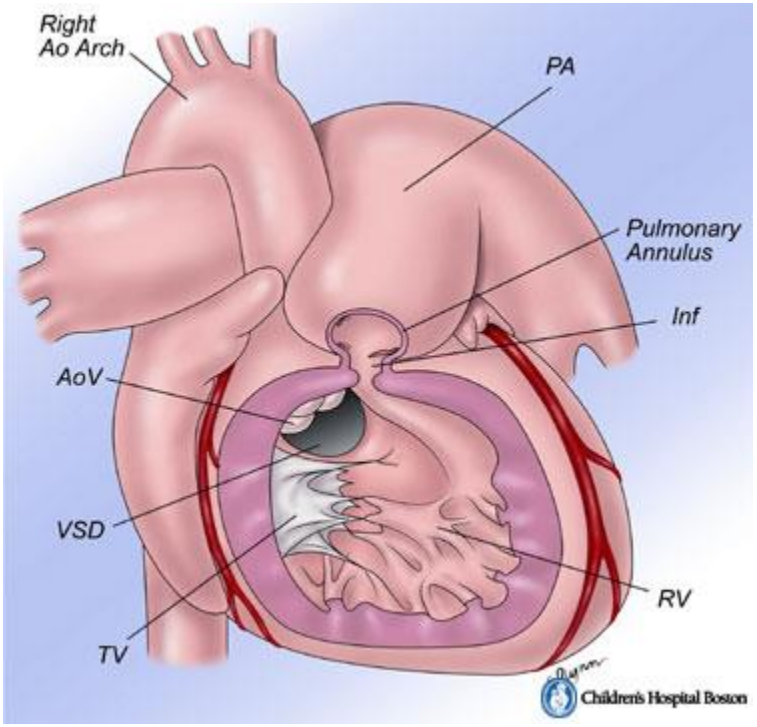
What would you do for a 24-week GP fetus with severe APVS?

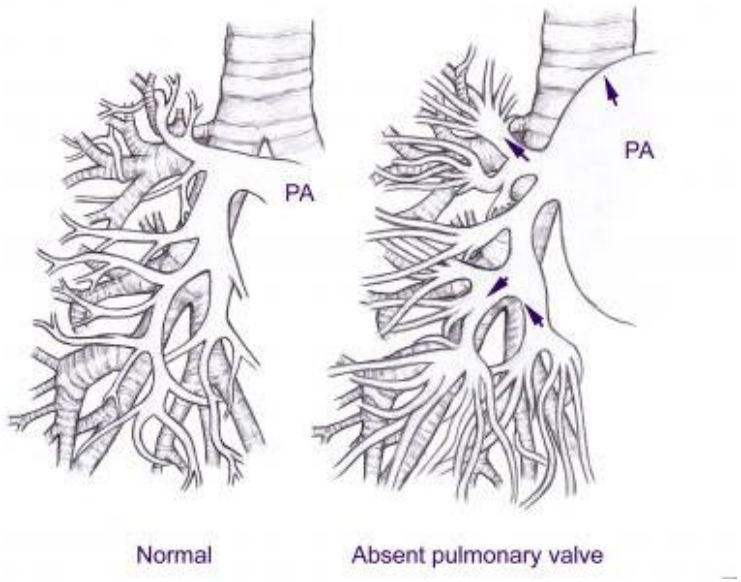
What are the bad prognostic factors for a 24-week GP fetus with severe APVS?

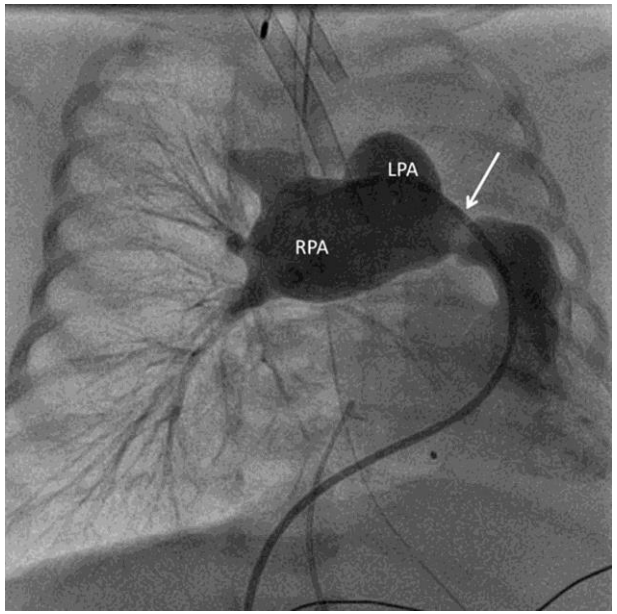
Absent PV syndrome (APVS)

- **APVS with ToF**
- **APVS with intact ventricular septum**
- **APVS in FSV**

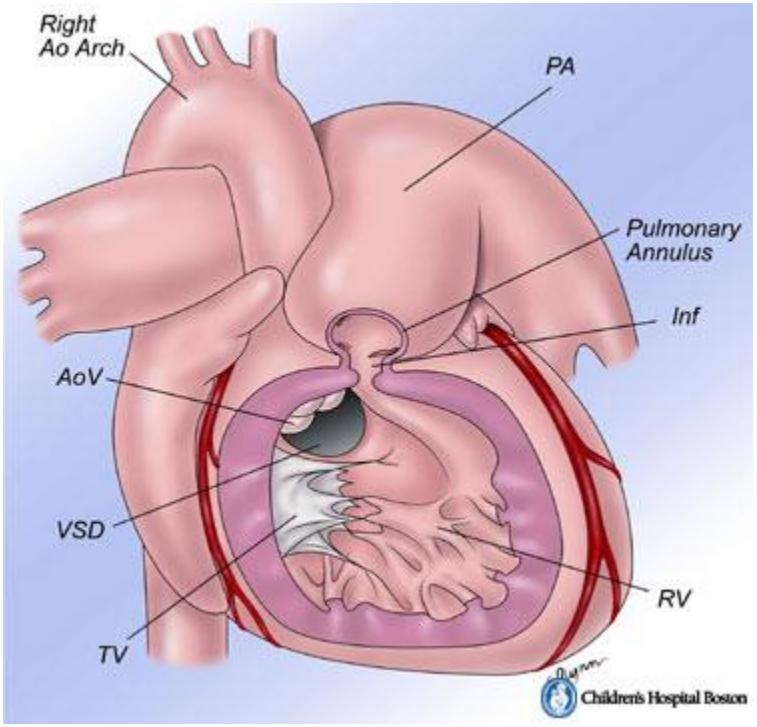
Absent PV syndrome







Absent PV syndrome



PS
High pressure pulsatile flow

PR
High volume ‘to and fro’ flow

Tetralogy of Fallot with Absent Pulmonary Valve: Echocardiographic Morphometric Features of the Right-Sided Structures and their Relationship to Presentation and Outcome

Mary T. Donofrio, MD, Marshall L. Jacobs, MD, and Jack Rychik, MD, *Richmond, Virginia*

Respiratory symptoms in tetralogy of Fallot with absent pulmonary valve are believed to be due to bronchial compression secondary to dilated pulmonary arteries; however, not all patients are born compromised. Echocardiographic morphometry of the right-sided structures was investigated to determine the possible relationship between anatomy and clinical presentation. Twenty-five patients were identified, and 15 had preoperative echocardiograms. Patients were divided into two groups: those with respiratory distress (group I, $n = 9$) and those without (group II, $n = 6$). No difference was noted in branch pulmonary artery diameters between groups; however, the pulmonary valve/aortic valve ratio, reflecting the dimension of the narrowest pathway from the right ventricle, was larger in group I (0.74 ± 0.15 versus 0.60 ± 0.07, $p < 0.05$). Pulmonary valve diameter correlated with main and right pulmonary artery diameters. We conclude that patients with tetralogy of Fallot with absent pulmonary valve and respiratory compromise have a greater pulmonary valve/aortic valve ratio but do not have greater dilatation of proximal branch pulmonary arteries. This suggests that the pathophysiology is not due solely to compression of the bronchi but is also related to the blood flow dynamics in the pulmonary vessels. (J Am Soc Echocardiogr 1997;10:556-61.)

Tetralogy of Fallot with absent pulmonary valve (TOF/APV) is a rare form of congenital heart disease with a wide spectrum of clinical presentations. Despite the continued improvement in surgical techniques and

cause for these findings in the face of a primary cardiovascular deformity are unsettled and speculative. Pulmonary artery dilatation has been hypothesized to be related to an intrinsic defect in the pulmonary vascula-

Value of Clinical and Echocardiographic Features in Predicting Outcome in the Fetus, Infant, and Child With Tetralogy of Fallot With Absent Pulmonary Valve Complex

Anita J. Moon-Grady, MD, Theresa A. Tacy, MD, Michael M. Brook, MD, Frank L. Hanley, MD, and Norman H. Silverman, MD, DSc (Med)

We describe clinical and echocardiographic features of tetralogy of Fallot with absent pulmonary valve complex (TOF/APVC) and hypothesized that outcome might be related to pulmonary artery enlargement or severity of illness. We examined the clinical records of all 23 patients evaluated at our institution before death or surgical correction of TOF/APVC between 1990 and 2000. Echocardiograms for 16 patients (including 5 fetuses) were also reviewed, and measurements of the semilunar valves and pulmonary arteries were obtained and compared with patient's aortic annulus size and with established normal subjects. Actuarial survival was 15 of 23 patients (68%) at 4 years. Four fetuses were hydropic and none survived; 7 patients were ventilator dependent at operation and only 3 survived. No difference was noted in pulmonary artery diameters in survivors versus nonsurvivors. Pulmonary valve annulus size was larger in nonsurvivors (103 ± 25% vs 71 ± 24% of normal, $p = 0.03$); however, when fetal examinations were excluded, this difference did not persist. Thus, only hydrops and ventilator dependence at diagnosis predicted mortality. There was no correlation between postnatal measurements of pulmonary arteries and outcome. Larger pulmonary annulus size in hydropic fetuses and poor survival among patients diagnosed in utero suggests that the pathophysiology in TOF/APVC is not due entirely to the aneurysmal dilation of the pulmonary arteries but may be related to right-sided cardiac dysfunction. ©2002 by Excerpta Medica, Inc.

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Anatomic Variability and Outcome in Prenatally Diagnosed Absent Pulmonary Valve Syndrome

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Background. We sought to describe current outcomes and risk factors for mortality for fetuses diagnosed with absent pulmonary valve syndrome (APV). Fetuses with APV were divided into two cohorts, those with underlying tetralogy of Fallot (TOF/APV) and those without underlying TOF and either an intact ventricular septum or small ventricular septal defect (APV/IVS).

Methods. The fetal echocardiographic database was reviewed from January 1, 2001, until June 1, 2010, and all subjects with a diagnosis of APV were included. Multiple clinical and fetal echocardiographic measurements were recorded. Statistical analysis was performed by χ^2 analysis and t tests. Survival analysis was performed by Kaplan-Meier analysis. Significant relationships between variables were explored by regression analysis. Significance was set at $p = 0.05$.

Results. The cohort consisted of 15 fetuses with TOF/APV and 6 fetuses with APV/IVS. There were no fetal demises in either cohort. Survival to birth was 71% in the TOF/APV cohort and 83% in the APV/IVS cohort ($p = 0.62$). Of subjects born alive, survival was 80% for both cohorts ($p = 0.95$). However, in the APV/IVS cohort, transplantation-free survival was only 20%. Underlying single-ventricle physiology strongly predicted those who underwent heart transplantation ($p = 0.003$, $R^2 = 0.50$). For the entire APV cohort, left ventricular dysfunction ($p = 0.005$, $R^2 = 0.41$) and a higher pulmonary artery valve–to–aortic valve ratio ($p = 0.02$, $R^2 = 0.34$) predicted mortality.

Conclusions. Postnatal outcomes continue to improve for fetuses with APV syndrome. Left ventricular dysfunction and higher pulmonary artery valve–to–aortic valve ratio accurately predict postnatal mortality for fetuses with APV.

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Table 1. Prenatal Echocardiographic Variables for the TOF/APV Cohort and the APV/IVS Cohort

Variables	TOF/APV	APV/IVS	*p* Value
Cardiothoracic area ratio	0.44 ± 0.44 (n = 12)	0.54 ± 0.07 (n = 6)	0.039
Pulmonary artery valve / aortic valve annular ratio	0.95 ± 0.27 (n = 12)	0.65 ± 0.15 (n = 6)	0.008
MPA *z* score	8.82 ± 6.02 (n = 12)	3.09 ± 4.42 (n = 5)	0.053
LPA *z* score	7.25 ± 7.45 (n = 13)	0.43 ± 1.84 (n = 5)	0.008
RPA *z* score	7.38 ± 5.44 (n = 12)	1.84 ± 2.84 (n = 5)	0.016
Severe LV dysfunction	2/15	2 /6	0.29
Severe RV dysfunction	3/15	6/6	0.001
LV enlargement	6/14	3/6	0.77
RV enlargement	12/14	2/6	0.019
Patent ductus arteriosus	2/15	6/6	<0.001
Hydrops fetalis	0/15	2/6	0.019

APV = absent pulmonary valve; IVS = intact ventricular septum or small ventricular septal defect; LPA = left pulmonary artery; LV = left ventricle; MPA = main pulmonary artery; RPA = right pulmonary artery; RV = right ventricle; TOF = tetralogy of Fallot.

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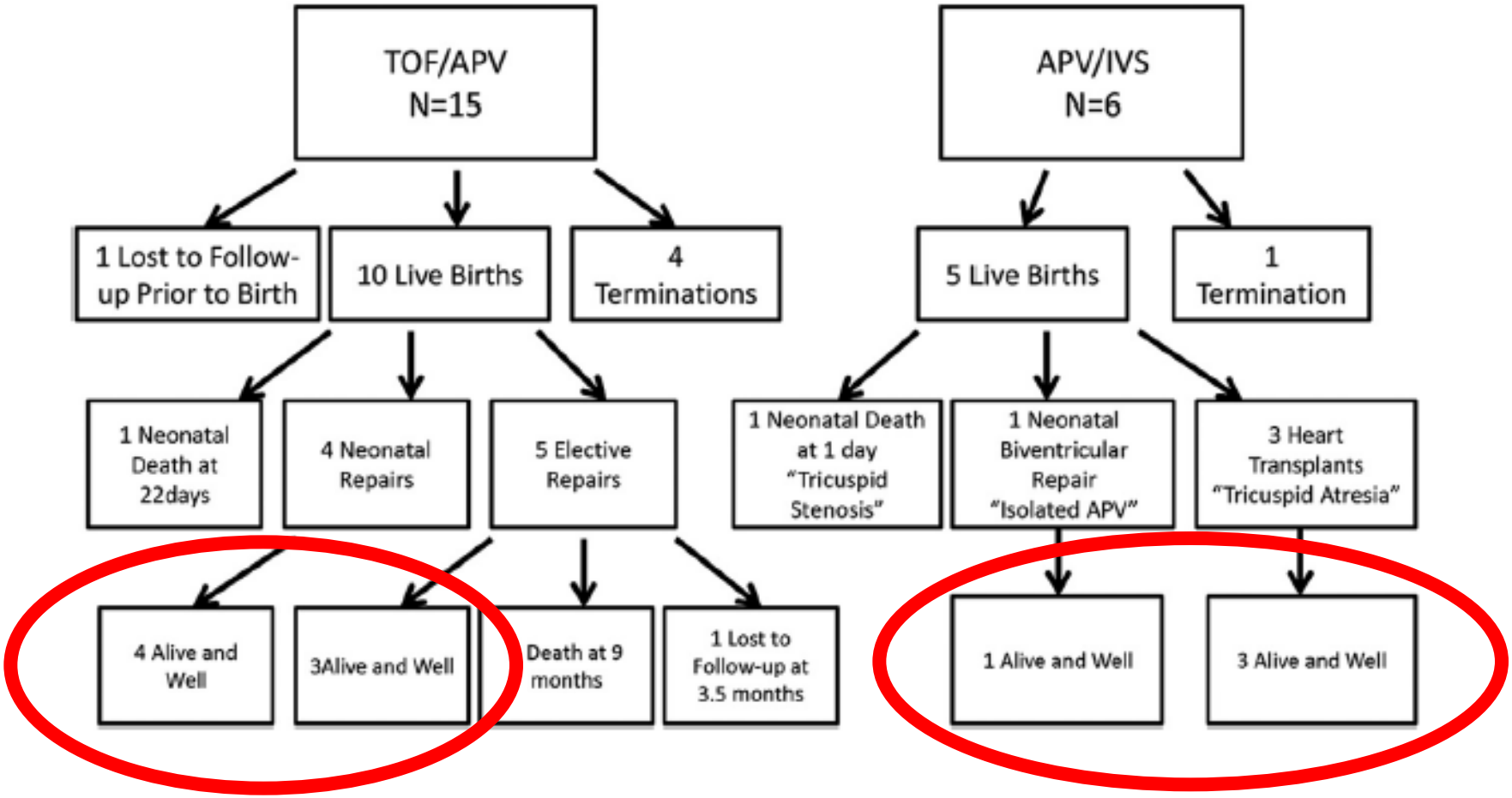

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Table 3. Postnatal Survivors vs Postnatal Nonsurvivors for the TOF/APV Cohort

Variables	Survivors	Nonsurvivors	*p* Value
Pulmonary artery valve / aortic valve annular ratio	0.82 ± 0.27 (n = 6)	1.23 ± 0.11 (n = 2)	0.022
MPA *z* score	5.37 ± 4.3 (n = 6)	5.18 ± 0.23 (n = 2)	0.96
LPA *z* score	6.22 ± 5.2 (n = 7)	5.93 ± 5.18 (n = 2)	0.95
RPA *z* score	9.30 ± 6.05 (n = 6)	4.8 ± 6.42 (n = 2)	0.41
1-min Apgar score	8 (5–9) (n = 6)	3.5 (1–6) (n = 2)	0.14
5-min Apgar score	8.5 (8–9) (n = 6)	4.5 (1–8) (n = 2)	0.14
Palliation during initial hospitalization	4/8	0/2	0.20
Intubation before initial surgical palliation	6/8	1/2	0.49
Supplemental oxygen before palliation	6/8	1/2	0.49
Days of intubation before initial palliation	4 (1–28) (n = 6)	21 (n = 1)	0.22
ICU days before initial palliation	13.5 (3–110) (n = 8)	28.0 (21–35) (n = 2)	0.27
Total hospitalization days	14.5 (6–110) (n = 8)	28.0 (21–35) (n = 2)	0.35
Genetic syndrome	2/8	0/2	0.43
Tracheostomy	2/8	1/2	0.49
Severe LV dysfunction	0/8	1/2	0.035
Severe RV dysfunction	1/8	1/2	0.24

APV = absent pulmonary valve; ICU = intensive care unit; LPA = left pulmonary artery; LV = left ventricle; MPA = main pulmonary artery; RPA = right pulmonary artery; RV = right ventricle; TOF = tetralogy of Fallot.

Prenatal Role of the Ductus Arteriosus in Absent Pulmonary Valve Syndrome

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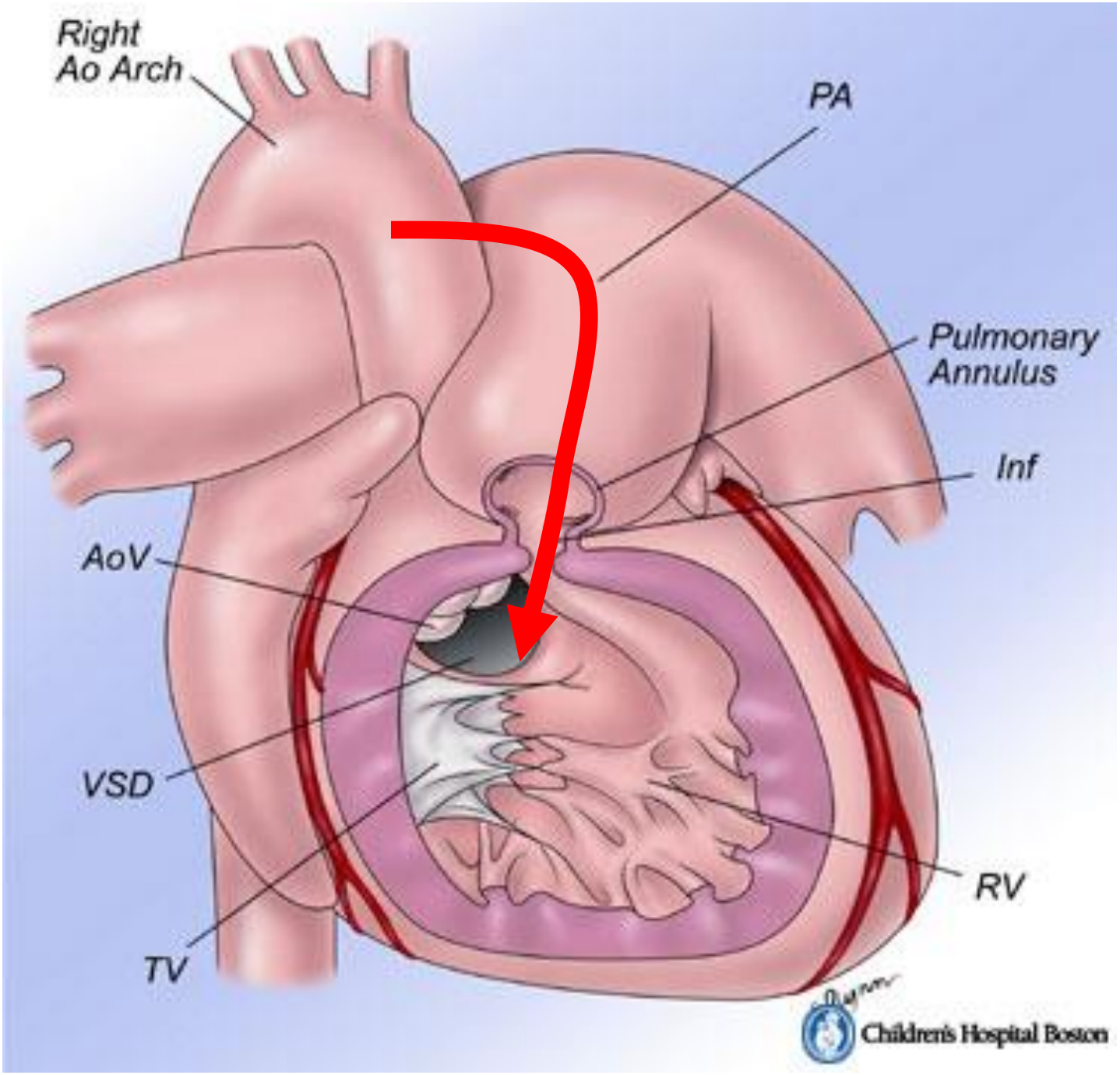
Absent pulmonary valve with ventricular septal defect is associated with ductal agenesis and markedly dilated main and branch pulmonary arteries. The less common variant with intact ventricular septum generally exhibits a patent ductus and smaller branch pulmonary arteries, and may be associated with tricuspid atresia. We identified 7 patients with the prenatal diagnosis of absent pulmonary valve, 5 with ventricular septal defect (Group 1) and 2 with an intact ventricular septum (Group 2). Imaging, color Doppler, and pulsed-Doppler recordings were analyzed. The branch and main pulmonary arteries were measured and expressed as a ratio with the descending aorta. Pulmonary regurgitation time (PRT) and diastolic acceleration time (DAT) were derived, and DAT/PRT was calculated to characterize diastolic pulmonary flow. Group 1 patients all had a large ventricular septal defect, normal biventricular size and function, and dilated main and branch pulmonary arteries. Group 2 patients had dilated main but smaller branch pulmonary arteries, moderate right ventricular dilation with severe dysfunction, and limited or absent tricuspid inflow. Group 1 demonstrated shorter acceleration time and earlier peak velocity, resulting in a smaller DAT/RT ratio. We speculate that free communication between the fetal aorta and the ventricles may limit atrial inflow and elevate diastolic pressure, affecting cardiac output, ventricular function, and atrioventricular valve development. With an intact ventricular septum, these physiologic and anatomic repercussions are limited to the right ventricle, but with a ventricular septal defect, both ventricles would experience similar consequences and cardiac performance could be critically impaired. (ECHOCARDIOGRAPHY, Volume 19, August 2002)

fetal, absent pulmonary valve, tetralogy of Fallot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Prenatal diagnosis of absent pulmonary valve syndrome from first trimester onwards: novel insights into pathophysiology, associated conditions and outcome

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KEYWORDS: absent pulmonary valve syndrome; congenital heart defect; fetus; prenatal diagnosis; tetralogy of Fallot

ABSTRACT

Objective *To assess the spectrum of associated anomalies, intrauterine course and outcome in fetuses with absent pulmonary valve syndrome (APVS).*

Methods *All cases with a prenatal diagnosis of APVS at two centers over a period of 13 years were analyzed retrospectively. APVS was diagnosed in the presence of rudimentary or dysplastic pulmonary valve leaflets with to-and-fro blood flow in the pulmonary trunk on color and pulsed-wave Doppler ultrasound. Data on demographic characteristics, presence of associated conditions, Doppler studies and pregnancy outcome were reviewed.*

eight had hydrops and/or increased nuchal translucency, six were associated with trisomy 13 or 18 and none survived.

Conclusion *APVS diagnosed in the first trimester is significantly associated with TOF, patency of the DA, abnormal Doppler parameters, lethal trisomies and intrauterine mortality. Cases of APVS with isolated TOF and agenesis of the DA have a better outcome than those with additional anomalies, with > 80% survival.*
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INTRODUCTION

Take home messages

- Overall outcomes of prenatally diagnosed APVS are bad, but fetal demise is rare.
- Prenatal risk factors for bad outcome

  A. Fetal hydrops

  B. Large PVA size (high PVA/AVA ratio)

  C. Ventricular dysfunction

  D. APVS with FSV, APVS with IVS

  E. High DAT/PRT of dutal flow reversal

  F. Genetic: CHARGE, Catch-22. fatal trisomies (13, 18)

What would you do for a 24-week GP fetus with severe APVS?

Get the pregnant mother to give birth to the baby!

Tell the mom ‘If you give birth to the baby, we will do our best!’