L'anatomie des cardiopathies congénitales : un apprentissage sans fin?

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Cardiac anatomy : a very old science…

- 500 BC: Alcméon, Crotone : the first to practice dissections, difference between arteries and veins
- First anatomic description of the heart : Philistion, Sicilia, 300-350 BC

Cardiac anatomy : congenital heart defects

- The first CHD described = 1671 : Stenson
  → 1888 : Fallot
- Malpositions of the great arteries
  - 1797: Baillie: « a very singular malformation of the heart » → 1814 : Farré : transposition of the great vessels
  - 1875 : von Rokitansky : congenitally corrected transposition
  - 1888 : Vierordt : partial transposition (DORV = 1957, Taussig-Bing 1949)
  - 1967 : DOLV, anatomically corrected malposition of the great arteries

The modern era : the pioneers

1964

Stella et Richard Van Praagh
Boston, MA, USA

1971

Robert H. Anderson
London, GB

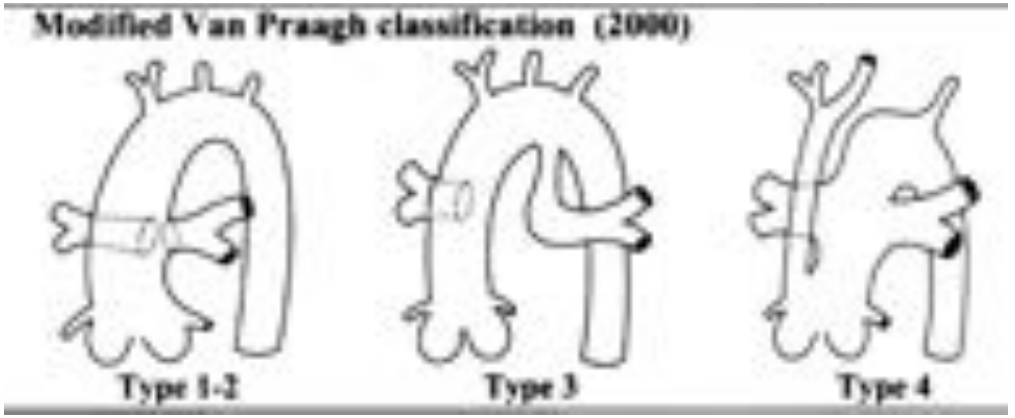
Anatomy of CHD : are we still learning ?

- Yes, of course
- Constantly !
- New cases, new malformations never described before
- Classifications
- « Revisiting » already known malformations
  - Link with cardiac development and genetics
  - Improve the description of the phenotypes

Can we still find « new » CHD?

Can we still find « new » CHD ?

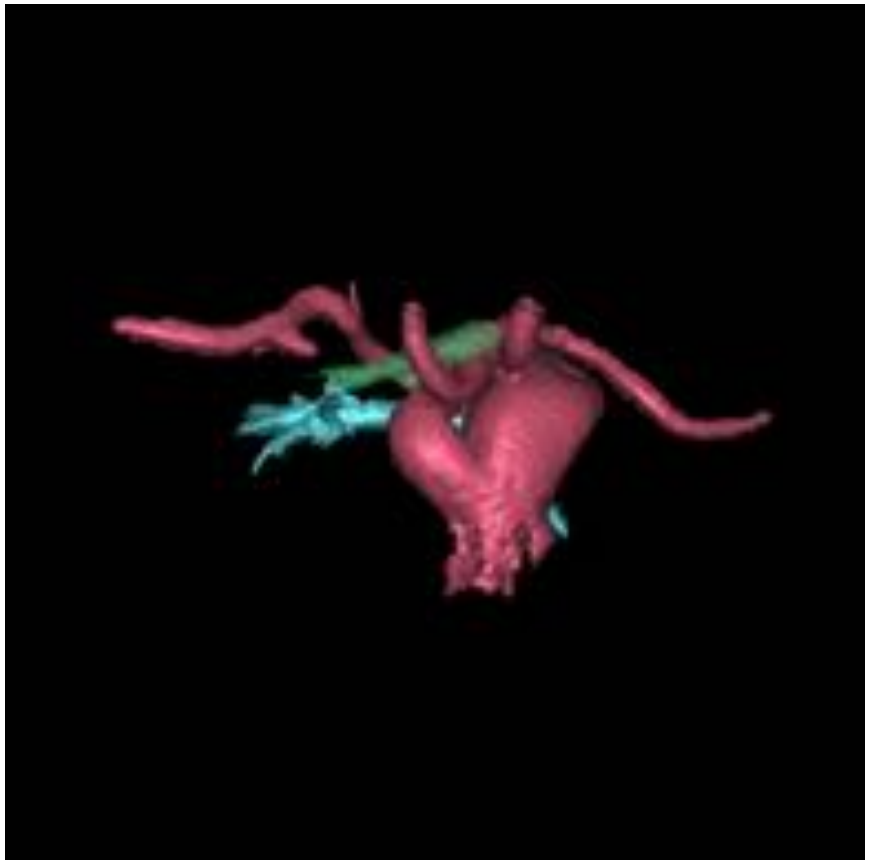
- First pregnancy
- Antenatal diagnosis of common arterial trunk (2nd trimester)
- Truncal valve quadricuspid, stenotic
- Coarctation? IAA?
- ??????

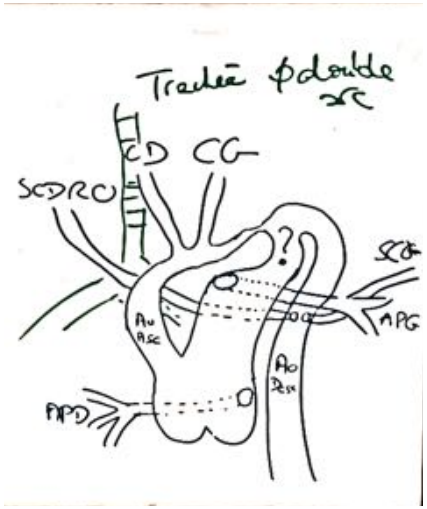


Jacobs ML. Ann Thorac Surg 2000
Van Praagh R. Eur J Cardioth Surg 1987

Can we still find « new » CHD ?

Common arterial trunk « 3/4 »



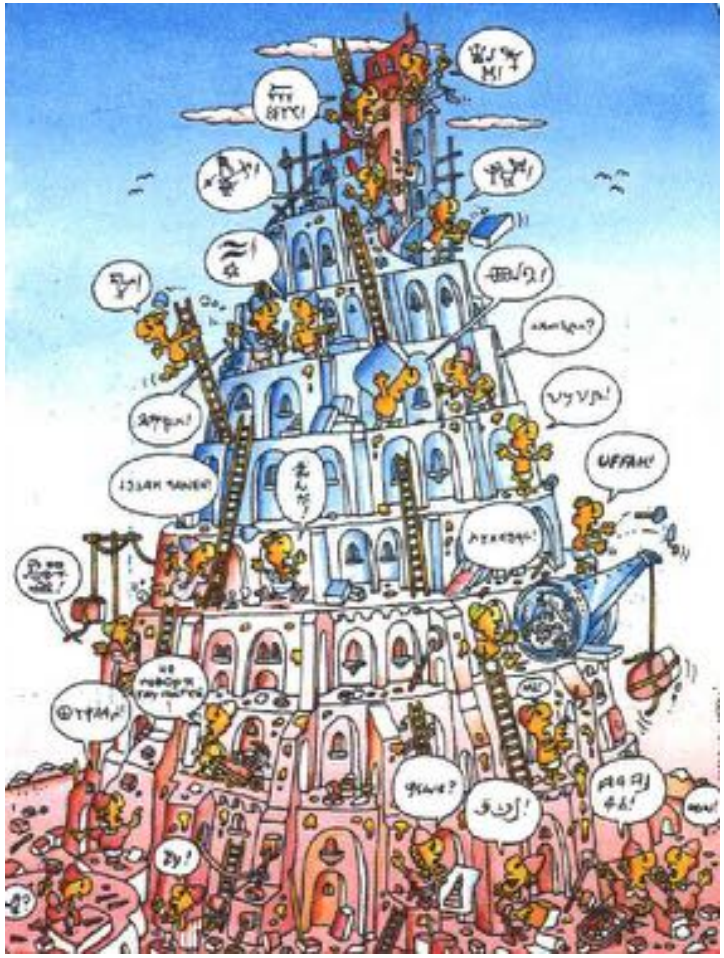


Courtesy F. Raimondi

Classifications
What for?

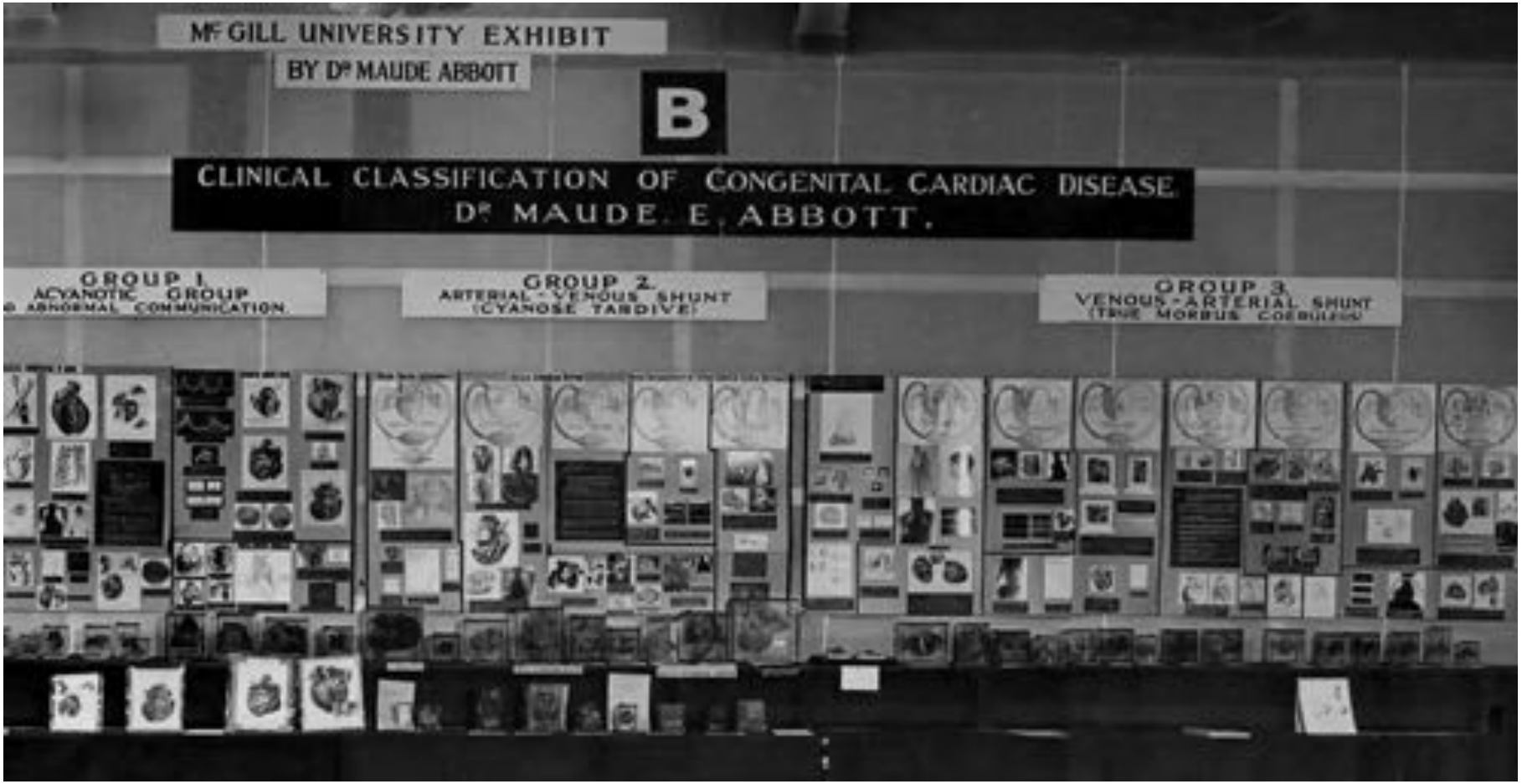
« Mal nommer un objet c'est ajouter au malheur de ce monde » Albert Camus

Classifications : the end of the Babel's tower?



- Classifications : absolute necessity, in order to speak the same language
- Multimodality imaging
- Clinicians, imagers, and surgeons

First classification of CHD : Maude Abbott

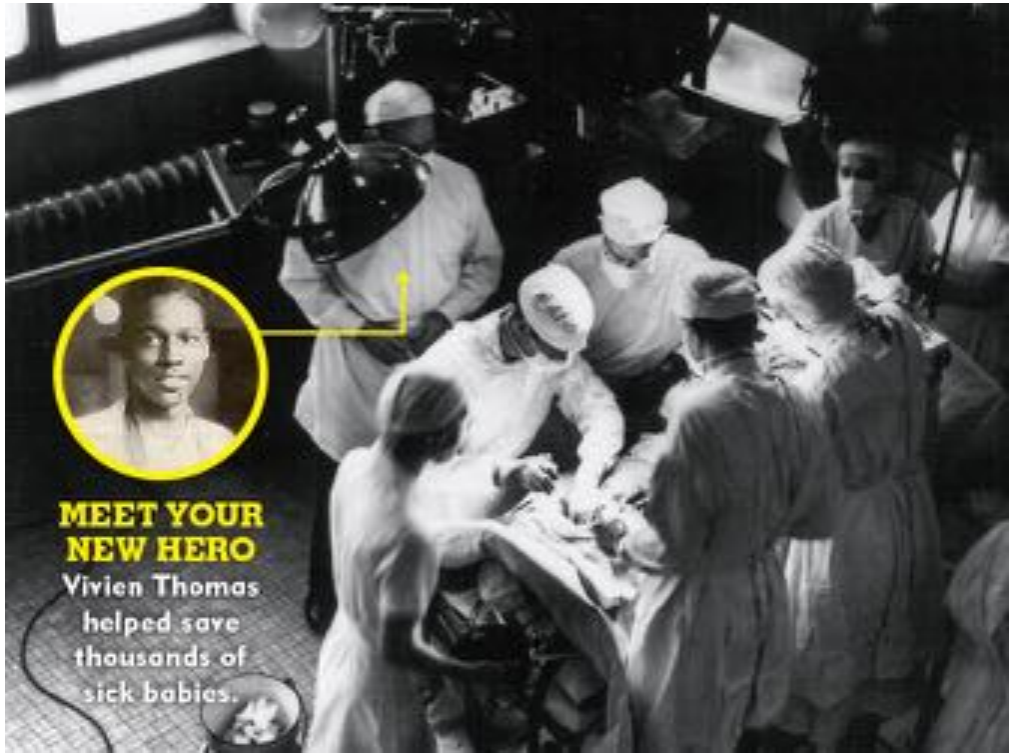


Centenary Meeting of the British Medical Association in London, England, 1932

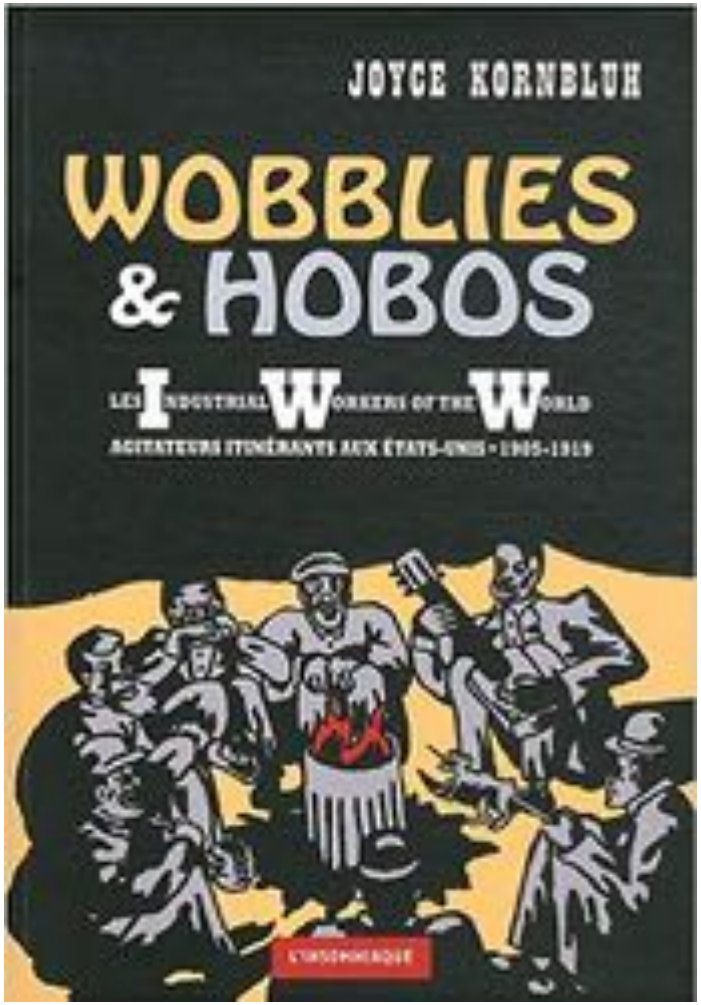
Atlas of congenital cardiac disease Maude Abbott, 1936

Atlas of congenital cardiac disease Maude Abbott, 1936

- 92 cases of persistence of the « shunt between aorta and PA »
- 1938: Gross = ligature of persistent arterial duct
- 1945 : first Blalock-Taussig-Thomas shunt



Anatomic and Clinical Classification of Congenital Heart Defects (ACC-CHD) based on the IPCCC



EPICARD Study

« *HoBo Classification* »

***Ho**uyel L, Khoshnood B, Anderson RH, Lelong N, Thieulin AC, Goffinet F, **Bo**nnet D; the EPICARD Study group. **Orphanet J Rare Dis**. **2011** Oct 3;6(1):64*

*Khoshnood B, Lelong N, Houyel L, Thieulin AC, Jouannic JM, Magnier S, Delezoide AL, Magny JF, Rambaud C, Bonnet D, Goffinet F; on behalf of the EPICARD Study Group. **Heart 2012**;98;1667-73.*

Anatomic and Clinical Classification of Congenital Heart Defects (ACC-CHD) based on the IPCCC

- *10 main categories*
  1. **Heterotaxy,** including isomerism
  2. Anomalies of the **venous returns**
  3. Anomalies of the **atria** and interatrial communications
  4. Anomalies of the **atrioventricular junction** and valves
  5. Complex anomalies of the atrioventricular connections
  6. **Functionally univentricular hearts**
  7. **Ventricular septal defects**
  8. Anomalies of the **ventricular outflow tracts** (VA connections)
  9. Anomalies of the extrapericardial **arterial trunks**
  10. Congenital anomalies of the **coronary arteries**
- *23 subcategories*
- *IPCCC codes*

ICD (International classification of diseases, WHO) and Congenital Heart Defects

- ICD-9 (1975) : 29 items
- ICD-10 (1989) : 73 items
- ICD-11 (2018) : 324 items, with corresponding IPCCC code, definitions, synonyms and commentaries

International Society for Nomenclature of Paediatric and Congenital Heart Disease

Franklin RCG et al. Nomenclature for congenital and paediatric cardiac disease: The IPCCC and the ICD-11. Cardiol Young ***2017****;27:1872-1938.*

Why do we need classifications ?

- To establish an universal language for people dealing with CHD all over the world
- ICD-11 : translation in progress

Béland MJ et al. Can J Cardiol 2018

- Databases
- Coding not only for billing, but also for scientific purposes
- To improve the precision of diagnostic and better identify the phenotypes

Revisiting the anatomy of CHD
Do we have still something to learn?

- Heterotaxy
- Ventricular septal defects
- Congenitally corrected TGA (double discordance)

Revisiting the anatomy of CHD

Heterotaxy

Heterotaxy.. or isomerism?

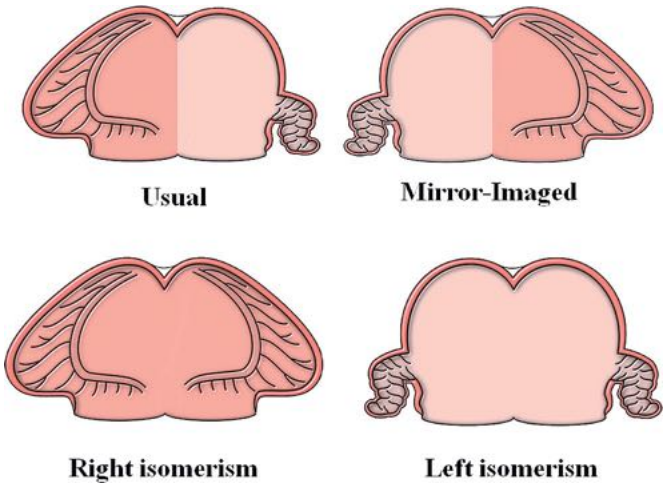
- Laterality defect
- Random organisation of the intrathoracic and intraabdominal organs

Van Praagh R, Van Praagh S. Am J Cardiol 1990

- Isomerism : implies an idea of symmetry and a not so random organisation

Uemura H et al. Ann Thorac Surg 1995
Tremblay C et al. Cardiol Young 2017

- Isomerism of the pectinate muscles : not always present in heterotaxy

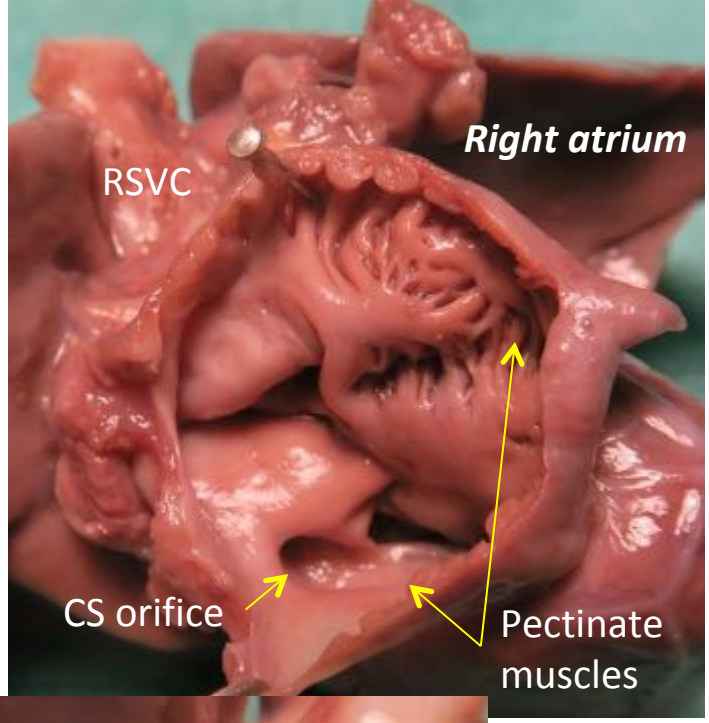


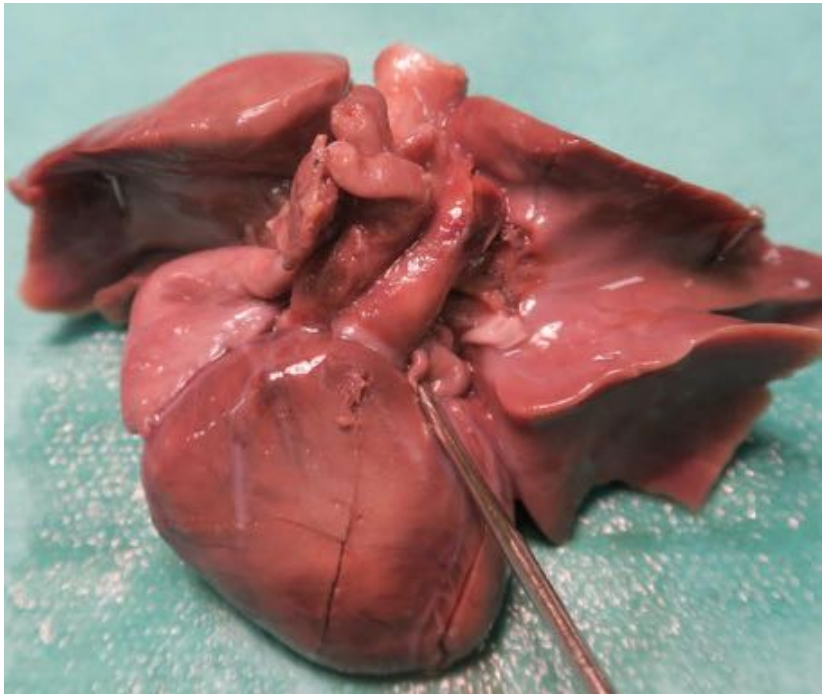
Anderson RH

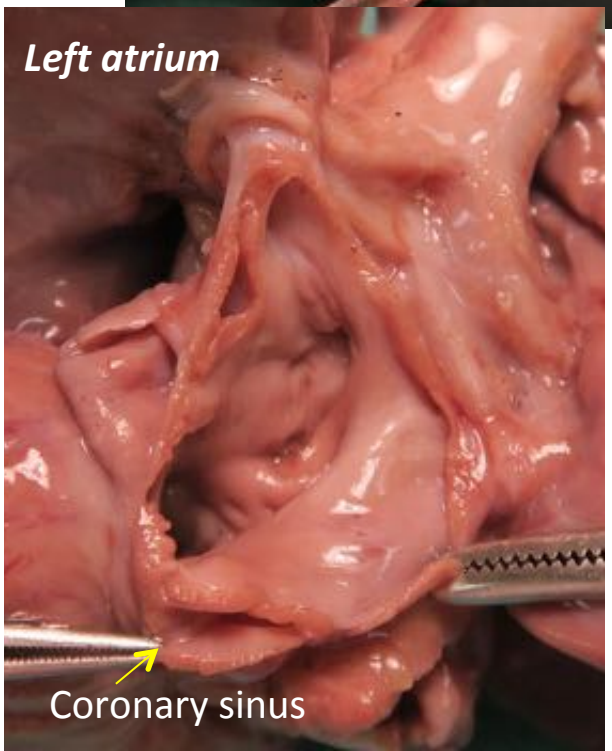
Heterotaxy.. or isomerism?

Fetal heart, 32 SA
Bronchopulmonary left isomerism, midline liver, intestinal malrotation, bilobed spleen

Interruption of the IVC with azygos return, LSVC to coronary sinus, normal PVs
Complete AV canal, DORV {S,D,D}, LV hypoplasia, coarctation







Heterotaxy: can (should) we classify?

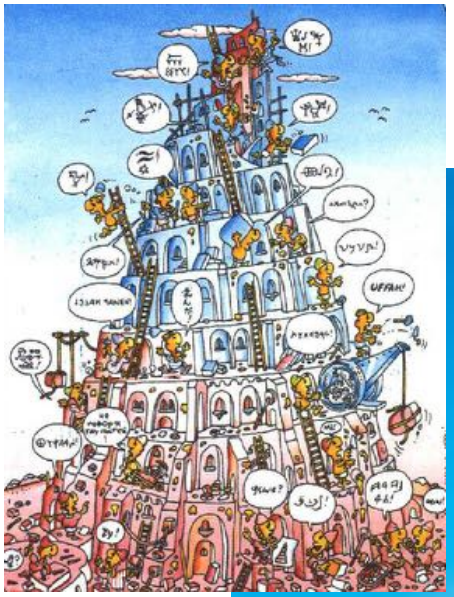
- Aim : to establish developmental and genetic links
- Historically : right isomerism = asplenia,
  left isomerism = polysplenia
- But: the spleen is abnormal only in 60% of heterotaxy patients *(Lin, Am J Med Genet A 2014)*
- Bronchial anatomy: better correlation, but discordance in 21% to 25% of patients *(Loomba, Cardiol Young 2016) (Yim, Circ CV imaging 2018)*
- Pectinate muscles can be analysed only at autopsy…..
- More and more exceptions, challenging all classifications
- Each patient is unique, and the arrangement of organs is often a mix of the two categories « right-sidedness » et « left-sidedness »

Heterotaxy? Or isomerism?

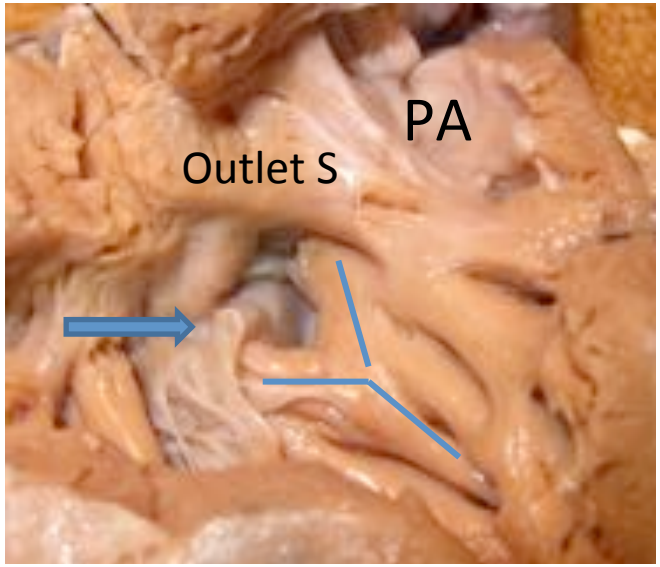
- Establish the diagnosis of heterotaxy
  - Abnormal symmetry of certain viscera and veins, and/or situs discordance between various organ systems and between the various segments of the heart *(Van Praagh S, 2006)*
  - At least 3/8 characteristic CHD or extracardiac abnormalities *(Lin 2014)*
- Look for bronchial isomerism
- Then : describe and be analytic ++++

1	**Characteristic CHD** **- TAPVR, PAPVR** **- Atrial SI or SA, common atrium** **- AVSD** **- Ventricular hypoplasia or malposition** **- VA alignment abnormalities (DORV, DOLV, TGA, CAT, TOF)** **- LVOTO or RVOTO**
2	Biliary atresia
3	Abdominal situs abnormality
4	Spleen abnormality
5	Isomerism of bronchi
6	Isomerism of the lungs
7	Similar morphology of the atrial appendages
8	2 of the following : - Systemic venous anomalies - Intestinal malrotation - Absent gallbladder

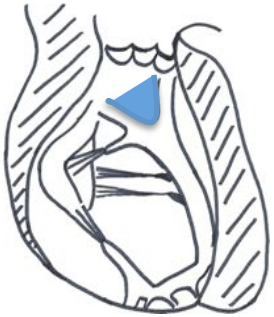
Revisiting the anatomy of CHD
The VSDs

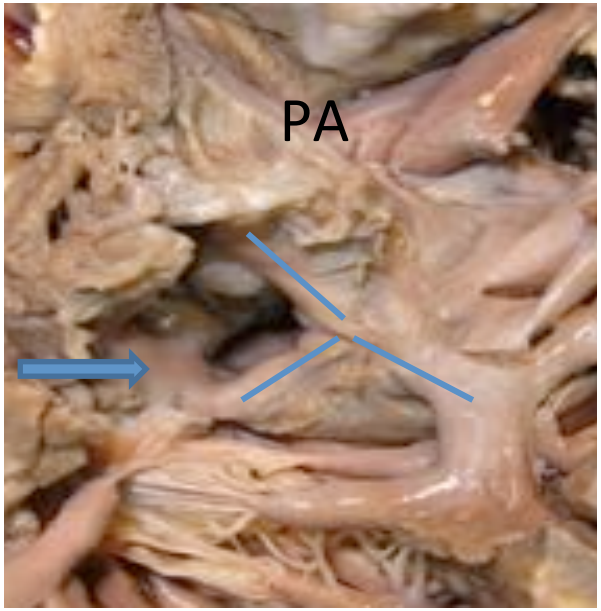


International Society for Nomenclature of Paediatric and Congenital Heart Disease

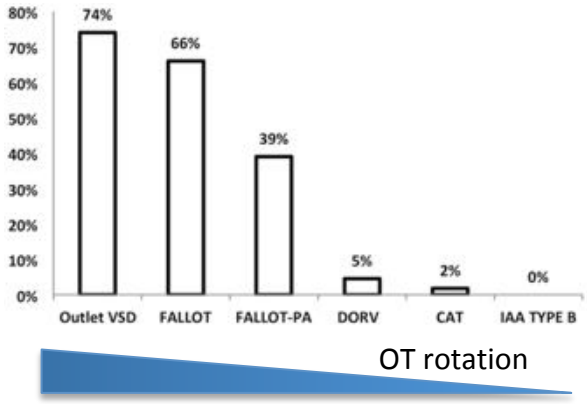


Outlet VSD
Anatomic characteristics



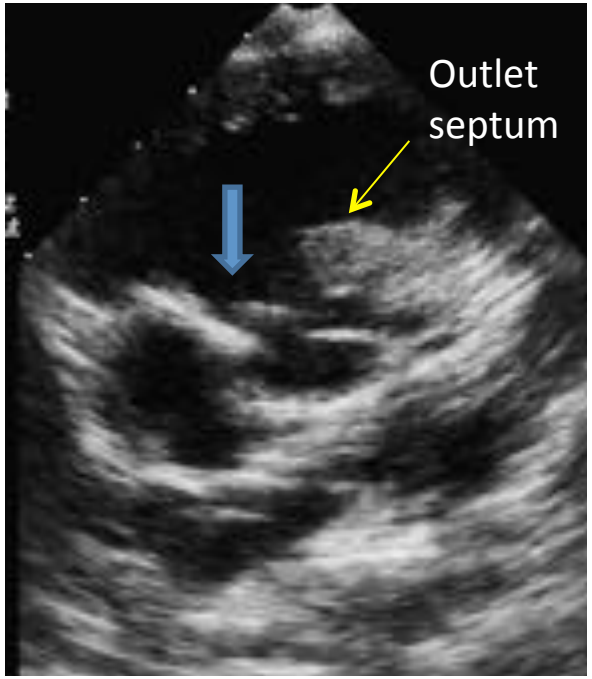


- All outlet VSDs (except some juxta-arterial VSDs) are located between the two limbs of the Y of the septal band
- Lack of fusion between the outlet septum and the ventricular septum
- All cardiac neural crest defects share the same VSD
- Borders : the postero-inferior rim can be fibrous (« outlet pm ») or muscular (outlet muscular)

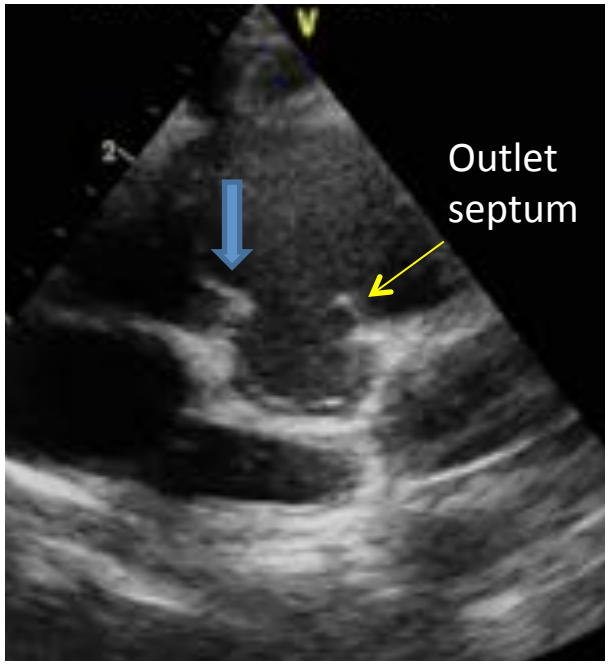


Mostefa-Kara et al, JTCS 2015

Outlet VSD with anteriorly malaligned outlet septum:
same geography, different borders



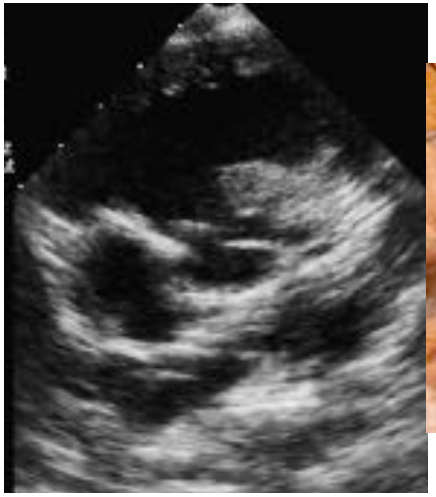
Fibrous (« outlet perimembranous »)

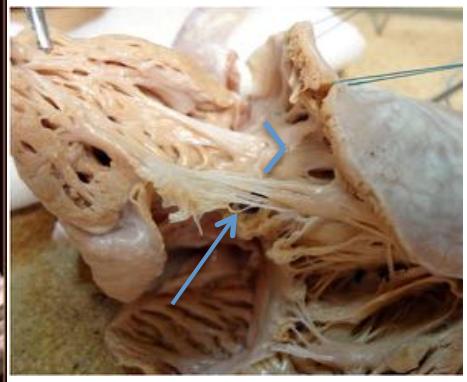


Muscular (« outlet muscular »)

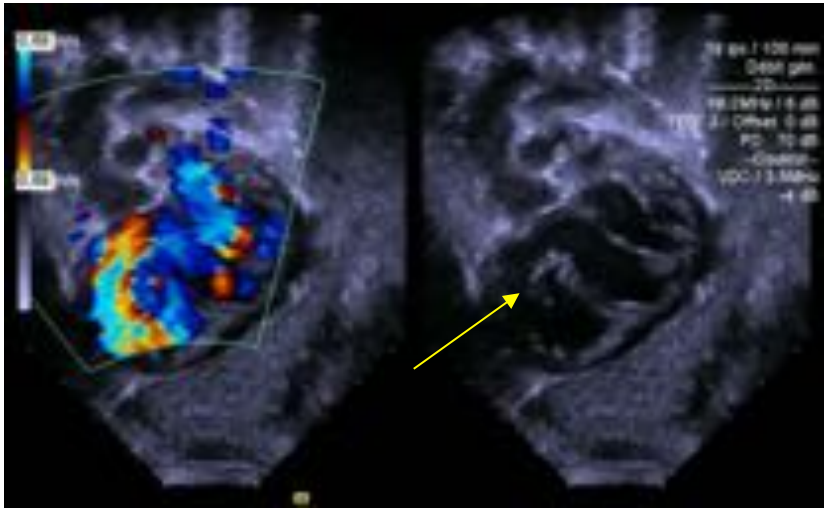
Courtesy D. Bonnet

Outlet VSD versus central perimembranous VSD:
same borders, different geography



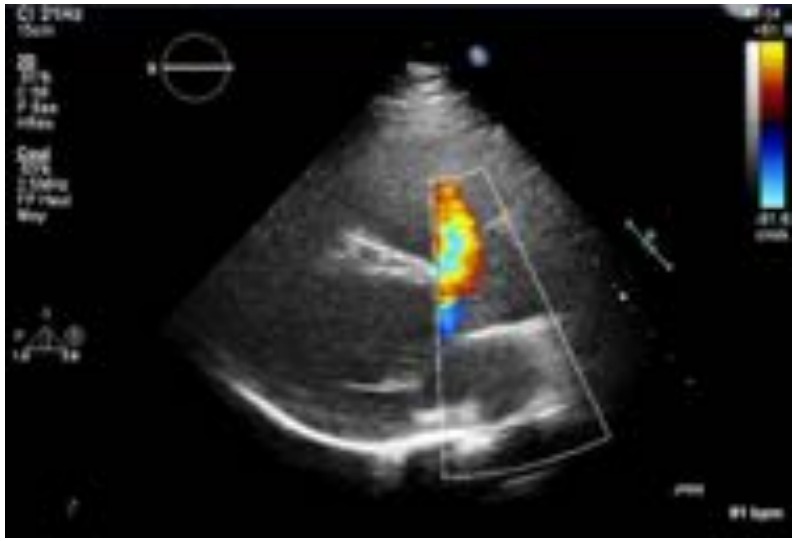


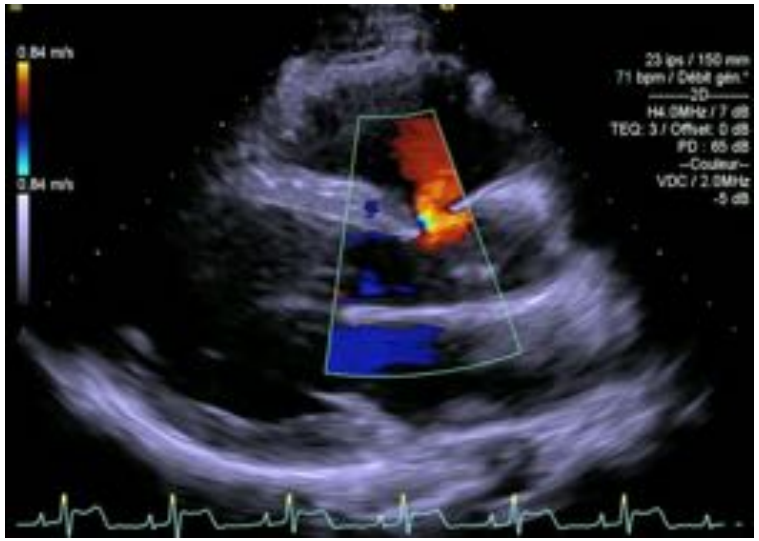
Outlet VSD with anteriorly malaligned outlet septum : ABOVE the upper septal attachments of the TV



Central pm VSD : BELOW the upper septal attachments of the TV

Outlet VSD versus central perimembranous VSD:
same borders, different geography





Courtesy X. Iriart

- Antenatal diagnosis
- Outlet VSD = cardiac neural crest defects (microdeletion 22q1.1)
- Central pm VSD = trisomies

Classification and nomenclature of VSDs

- ISNPCHD → ICD-11
- Classification in 4 main categories, based on *geography*
  - Central perimembranous
  - Inlet
  - Trabecular muscular
  - Outlet

- In each category (outlet VSDs) : subclassification according to *borders*
  - Perimembranous (fibrous continuity)
  - Muscular
- Aim : harmonize and unify the different approaches between clinicians, imagers, surgeons, and anatomists

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REPORT

Classification of Ventricular Septal Defects for the Eleventh Iteration of the International Classification of Diseases—Striving for Consensus: A Report From the International Society for Nomenclature of Paediatric and Congenital Heart Disease

[illegible]

Lee Lopez, MD, Lucile Houyel, MD, Steven D. Colan, MD, Robert H. Anderson, MD, PhD (Hon), Marie J. Béland, MDCM, Vera D. Aiello, MD, PhD, Frederique Ruiffard, MD, MS, Meryl S. Cohen, MD, Jeffrey P. Jacobs, MD, Hiromi Kurosawa, MD, Stephen P. Sanders, MD, Henry L. Walters, III, MD, Paul M. Weinberg, MD, Jeffrey R. Boris, MD, Andrew C. Cook, PhD, Adrian Crucean, MD, PhD, Allen D. Everett, MD, J. William Gaynor, MD, Jorge Giroud, MD, Kristine J. Guleserian, MD, Marina L. Hughes, DPhil, FRACP, Amy L. Juraszek, MD, Otto N. Krogmann, MD, Bohdan J. Maruszewski, MD, PhD, James D. St. Louis, MD, Stephen P. Sualar, MD, PhD, Diane E. Spicer, BS, PA, Shubhika Srivastava, MBBS, Giovanni Stellin, MD, Christo I. Tchervenkov, MD, Lianyi Wang, MD, and Rodney C. G. Franklin, MD

[illegible]

Classification of VSD for ICD-11 (Lopez L et al. Ann Thorac Surg 2018;106:1578-89)

- **Central perimembranous VSD (07.10.01)**

- **Inlet VSD without a common atrioventricular junction (07.14.05)***
  - Inlet VSD without atrioventricular septal malalignment without a common AV junction and with perimembranous extension (07.10.02)
  - Inlet VSD with atrioventricular septal malalignment and without a common AV junction (07.14.06)
  - Inlet muscular VSD (07.11.02)

- **Trabecular muscular VSD (07.11.01)**
  - Trabecular muscular VSD: Midseptal (07.11.04)
  - Trabecular muscular VSD: Apical (07.11.03)
  - Trabecular muscular VSD: Postero-inferior (07.11.12)
  - Trabecular muscular VSD: Anterosuperior (07.11.07)
  - Trabecular muscular VSD: Multiple ("Swiss cheese" septum) (07.11.05)

* The interventricular communication associated with a common AV junction (VSD component of an AV septal or AV canal defect) should be considered in the common AV junction section for coding purposes (AV septal defect: ventricular component, 06.06.04).

- **Outlet VSD (07.12.00)**
  - Outlet VSD **without malalignment** (07.12.09)
    - Outlet muscular VSD without malalignment (07.11.06)
    - Doubly committed juxta-arterial VSD without malalignment (07.12.01)

      Doubly committed juxta-arterial VSD without malalignment and with muscular postero-inferior rim (07.12.02)

      Doubly committed juxta-arterial VSD without malalignment and with perimembranous extension (07.12.03)

  - Outlet VSD **with anteriorly malaligned outlet septum** (07.10.17)
    - Outlet muscular VSD with anteriorly malaligned outlet septum (07.11.15)
    - Outlet VSD with anteriorly malaligned outlet septum and perimembranous extension (07.10.04)
    - Doubly committed juxta-arterial VSD with anteriorly malaligned fibrous outlet septum (07.12.12)

    Doubly committed juxta-arterial VSD with anteriorly malaligned fibrous outlet septum and muscular postero-inferior rim (07.12.07)

    Doubly committed juxta-arterial VSD with anteriorly malaligned fibrous outlet septum and perimembranous extension (07.12.05)

  - Outlet VSD **with posteriorly malaligned outlet septum** (07.10.18)
    - Outlet muscular VSD with posteriorly malaligned outlet septum (07.11.16)
    - Outlet VSD with posteriorly malaligned outlet septum and perimembranous extension (07.10.19)
    - Doubly committed juxta-arterial VSD with posteriorly malaligned fibrous outlet septum (07.12.13)

    Doubly committed juxta-arterial VSD with posteriorly malaligned fibrous outlet septum and muscular postero-inferior rim (07.12.08)

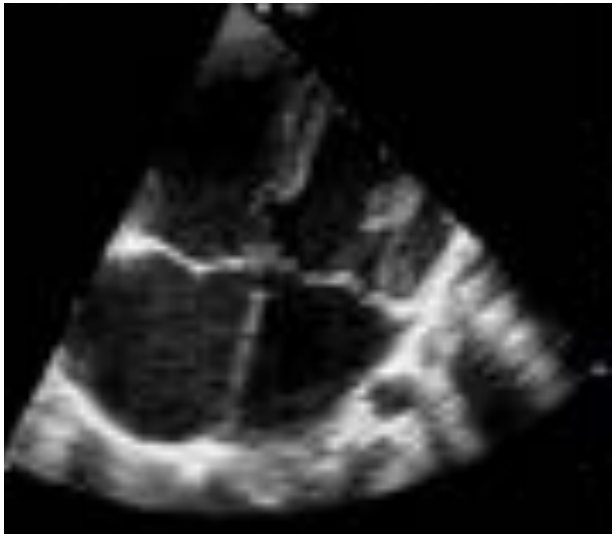
    Doubly committed juxta-arterial VSD with posteriorly malaligned fibrous outlet septum and perimembranous extension (07.12.06)

Central perimembranous VSD: Parasternal long-axis and short-axis view

Courtesy X. Iriart

Inlet VSD : the 4-chamber view

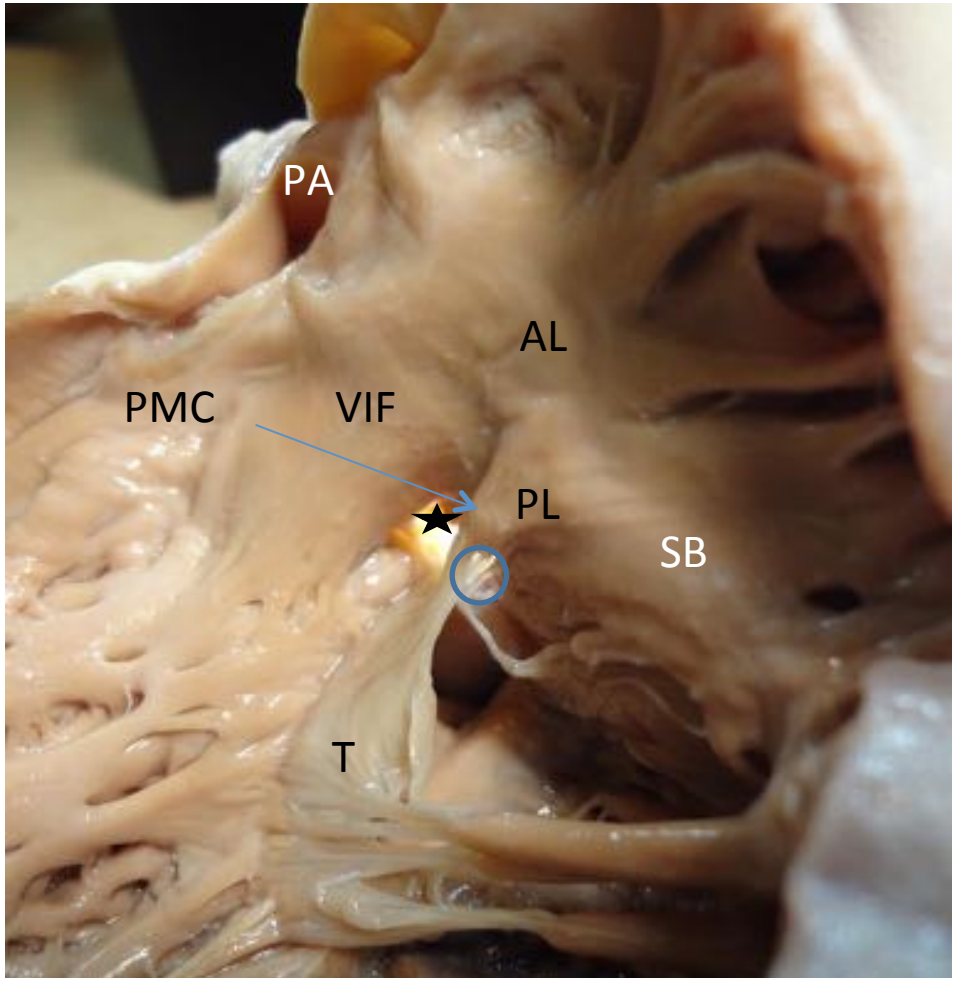
Central perimembranous VSD
with inlet extension



Inlet VSD
Common AV junction

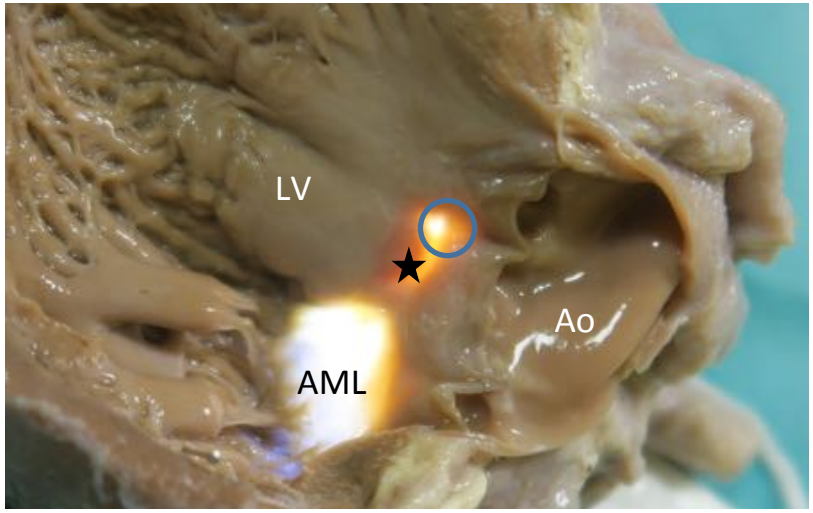
Inlet VSD
Malalignement AS/VS
Straddling TV

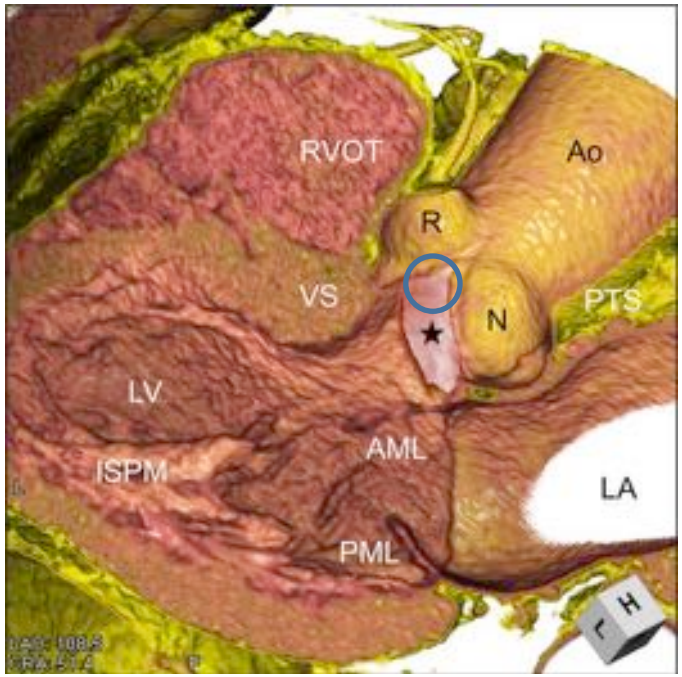
The membranous septum



★ Atrioventricular membranous septum

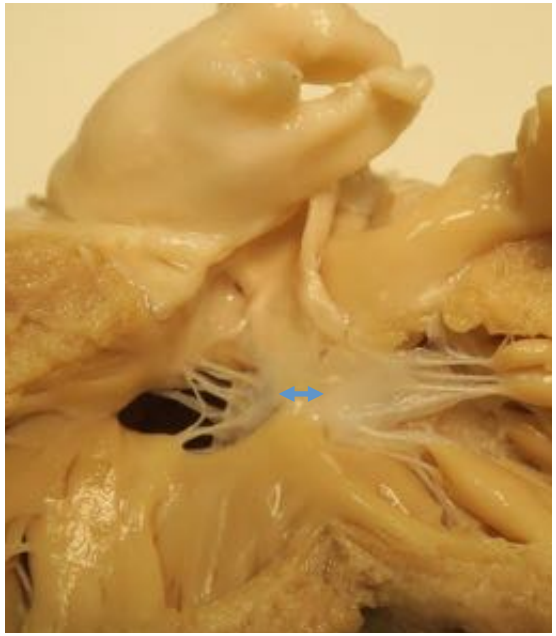
○ Interventricular membranous septum

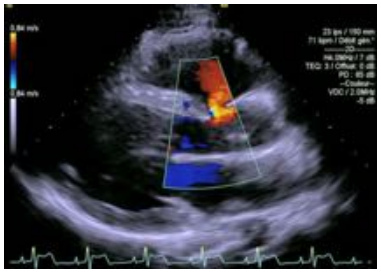


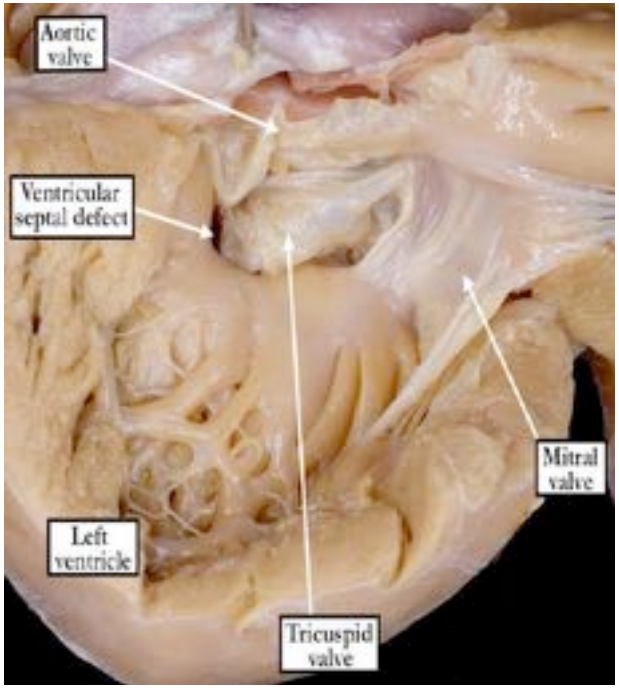


Mori S et al. Clinical Anatomy 2016;29:353–363

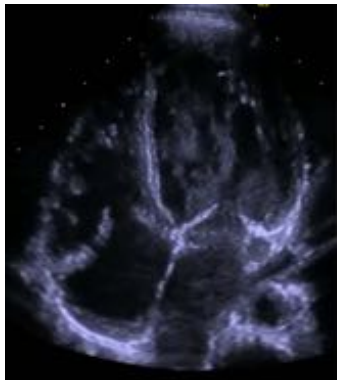
Central/inlet perimembranous VSD



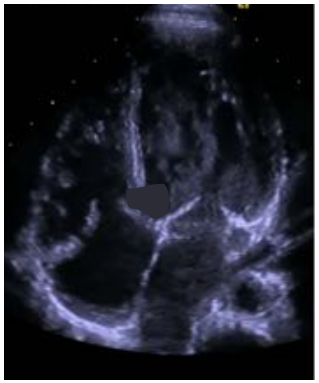




INDIRECT fibrous continuity between M and T valves
= CENTRAL PM



DIRECT fibrous continuity between M and T valves
= CENTRAL PM WITH INLET EXTENSION
(= INLET pm VSD)



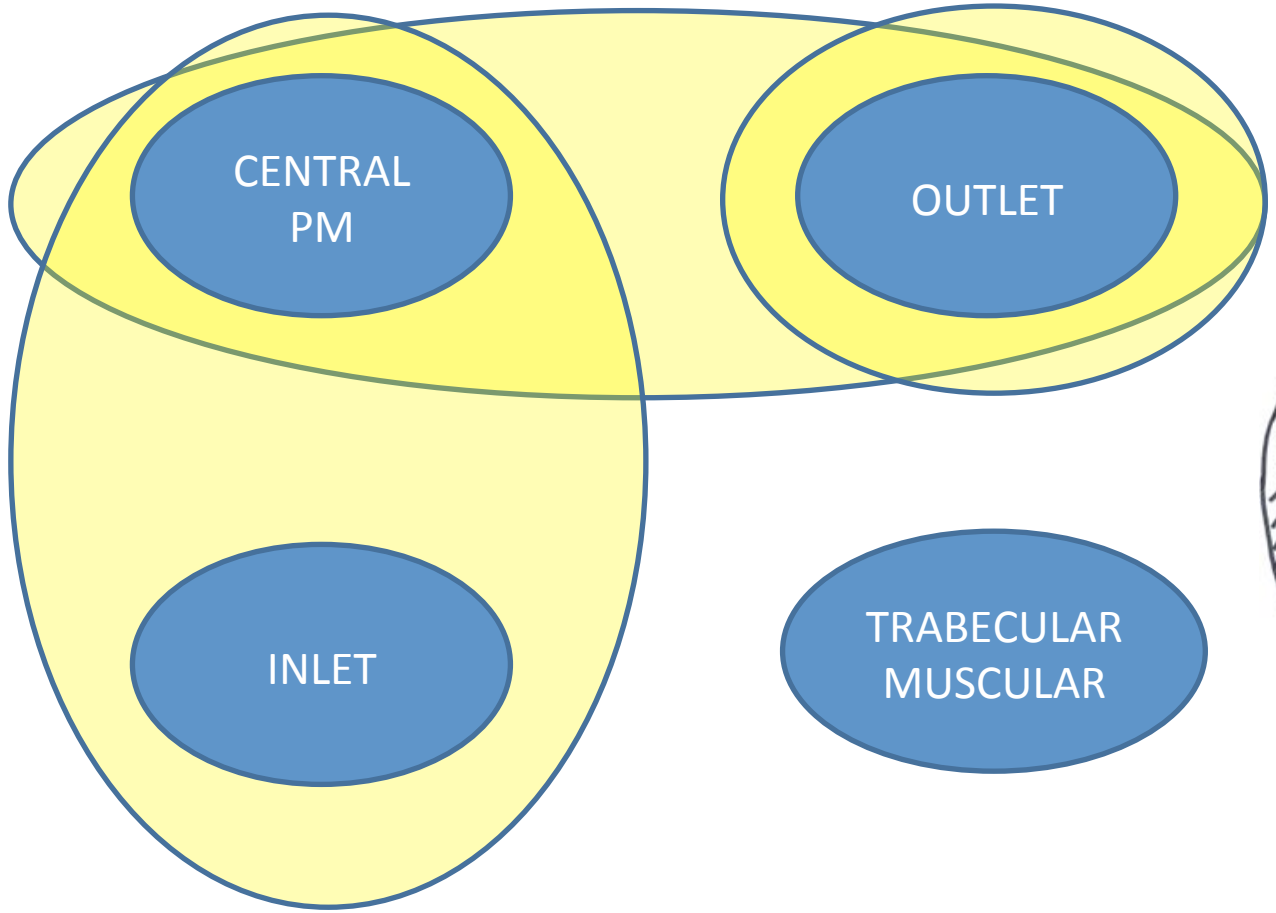
VSD : What did we learn?

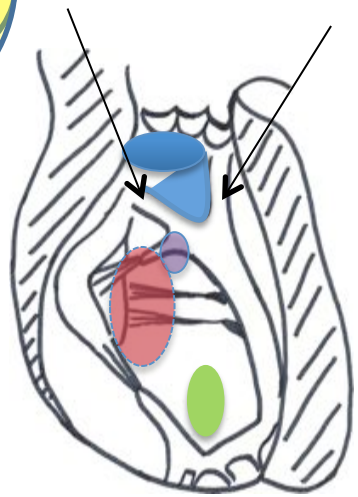
GEOGRAPHY : Etiology, associated lesions, outcome

OUTLET SEPTUM : Associated lesions, outcome

BORDERS : Conduction tissue

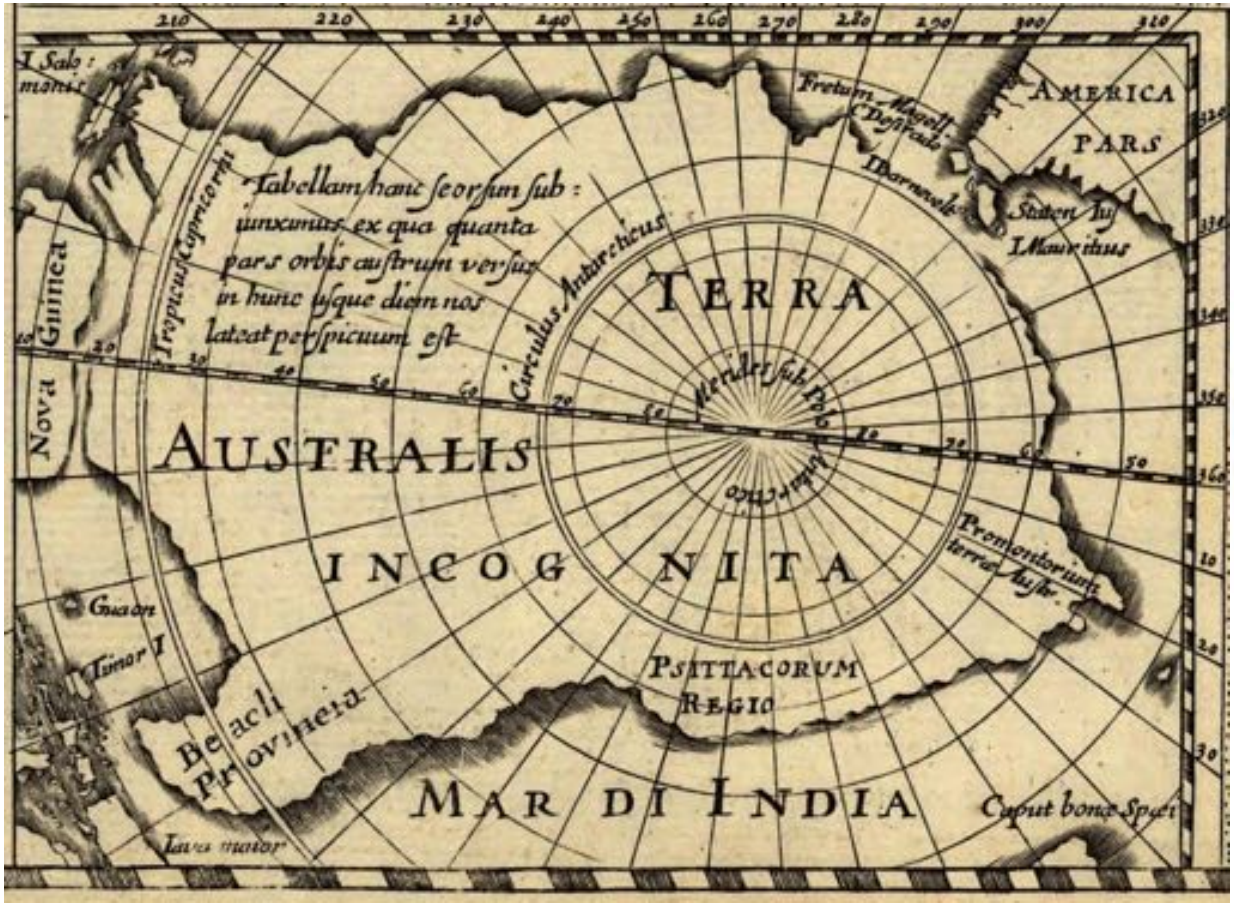
BORDERS : differential diagnosis



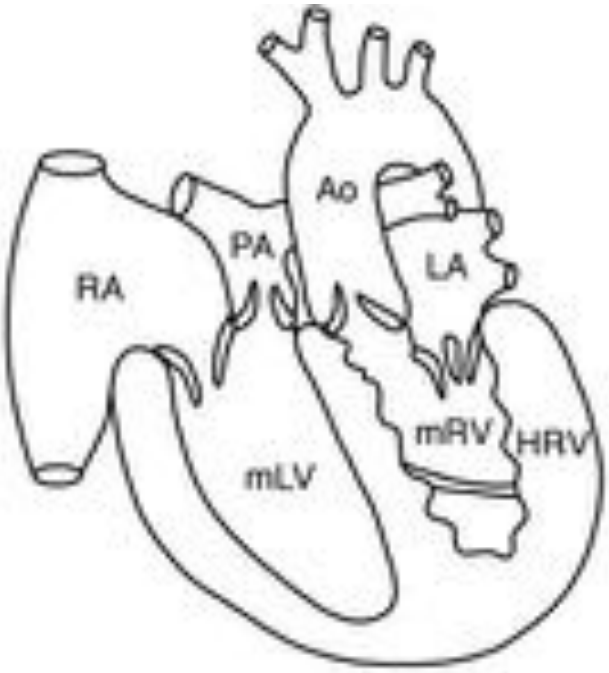


Revisiting the anatomy of CHD

ccTGA (double discordance)



Revisiting the anatomy of CHD
ccTGA (double discordance)



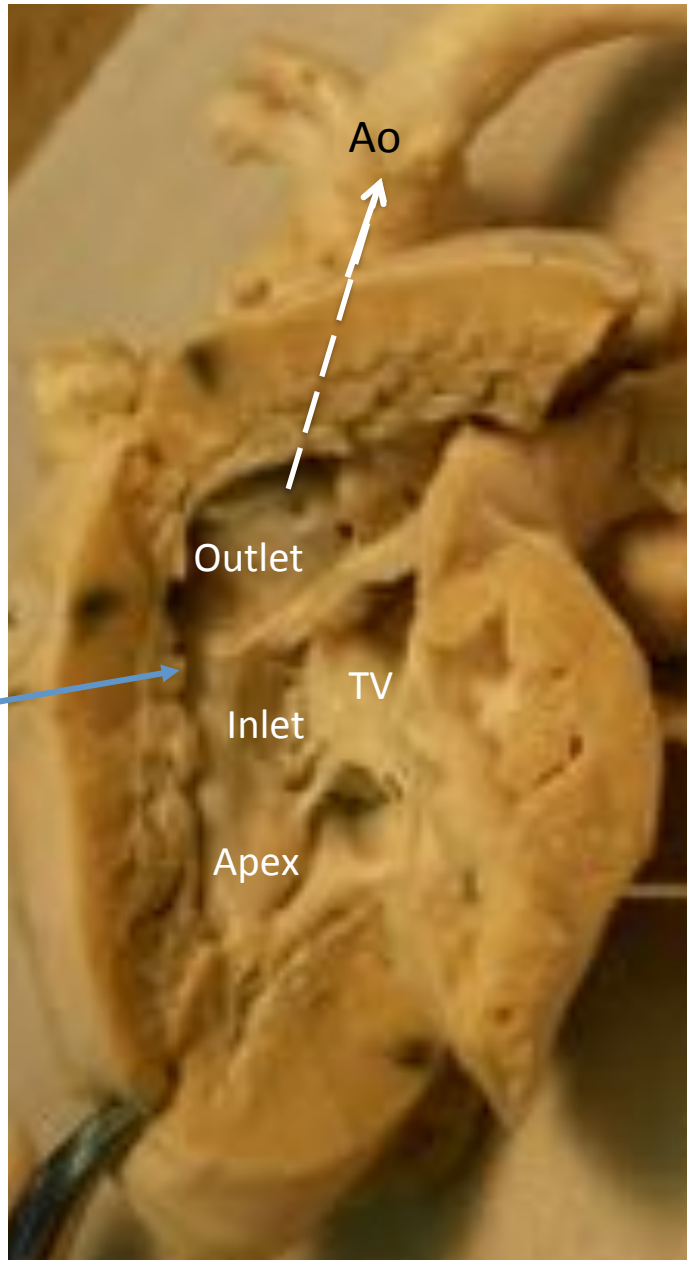
- 0,5% of all CHD
- Laterality defect
- Discordant AV connections
- Discordant VA connections
- S,L,L
- Rarely isolated
- Often associated with VSD, RV hypoplasia, subpulmonary stenosis or pulmonary atresia
- Always associated with abnormal location of the conduction system

ccTGA (double discordance) the VSD enigma

- VSD : 80-85% (anatomic series)
  60-65% (clinical series)
- 1. Where is the VSD?
  - Subpulmonary, outlet pm ? *Hornung et al. Heart 2010*
  - Inlet ? *Kutty et al. Heart 2018*
  - Central pm with inlet extension ? *Allwork et al. Am J Cardiol 1976*
  - Outlet (conoventricular)? *Van Praagh et al. Am Heart J 1998*
- 2. Why is it so difficult to describe ?

The RV in double discordance

- The RV and the tricuspid valve are almost always abnormal
- RV sinus hypoplasia, +/- Ebstein
- Constriction of the junction between inlet and outlet
- The Y of the septal band looks abnormal



Allwork et al. Am J Cardiol 1976
Brida et al. Circulation 2018;137:508-18

Questions

- Are the ventricles in ccTGA just inverted? Or completely different from a normal heart?
- Are they really different or do they just appear different?
- Optical illusion?
- If they are different, is it because of the L-loop? Or of the L-malposition of the vessels?

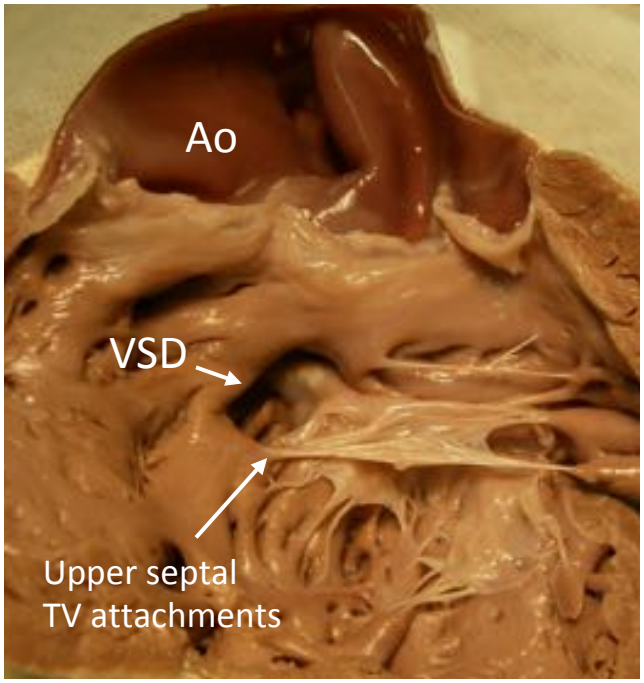
The VSD in ccTGA

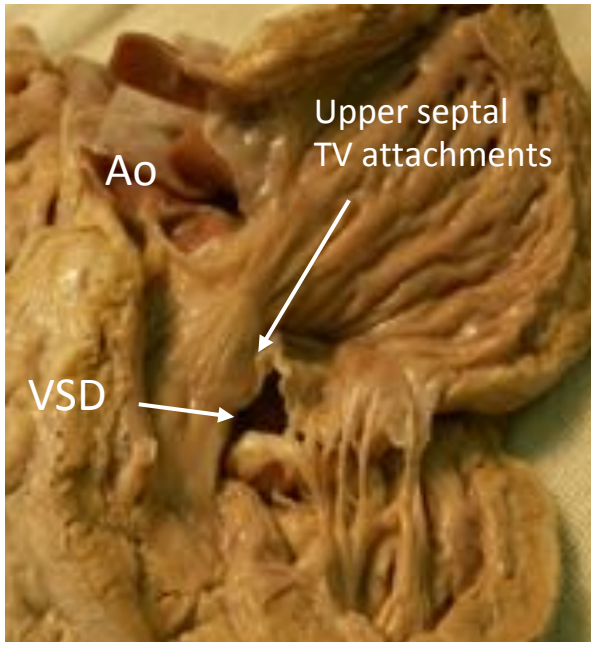
Nicolas Arribard, M2

- 31 ccTGA : VSD = 84%
- Classification : TV upper septal attachments
  - Above : outlet VSD
  - Below : Inlet VSD

Lopez et al. Ann Thorac Surg 2018

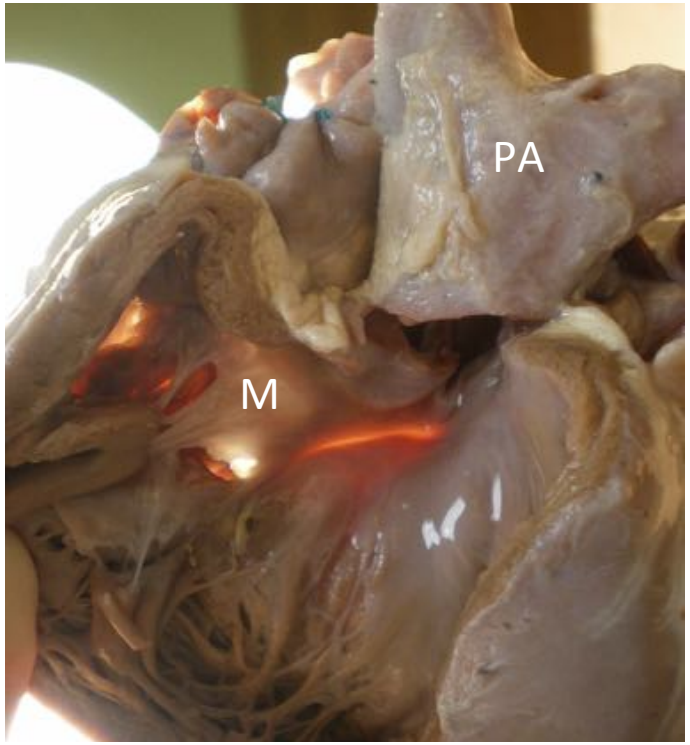
- Results
  - Outlet = 65%
  - Inlet = 23%
  - Muscular = 4%
  - Confluent = 8% (inlet/outlet)



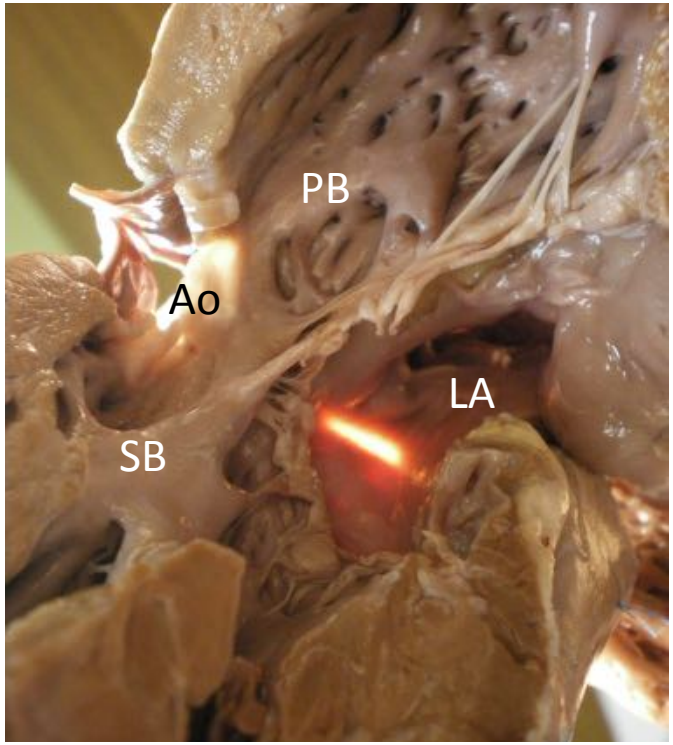


ccTGA: the membranous septum

- Is only an atrioventricular structure (LA/LV)
- Fills the gap between the malaligned A and V septum



Left ventricle, right-sided

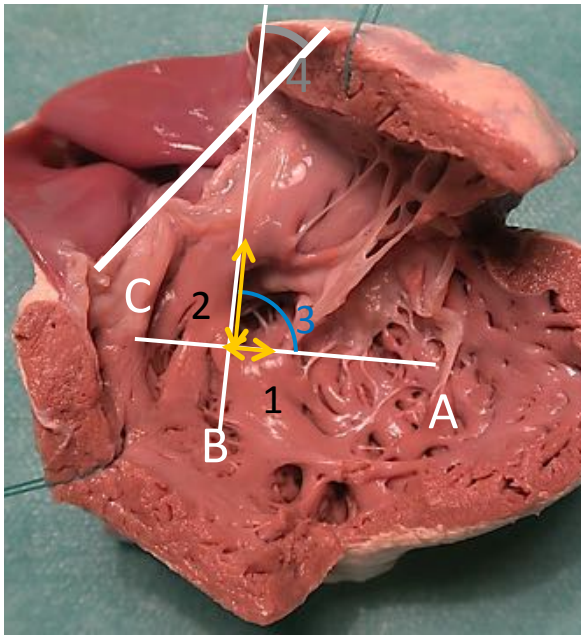


Right ventricle, left-sided

The anatomy of the right ventricle in ccTGA

- 31 ccTGA, 36 TGA, 35 normal hearts
- Anatomy of the septal band : ccTGA is closer to normal heart than TGA !

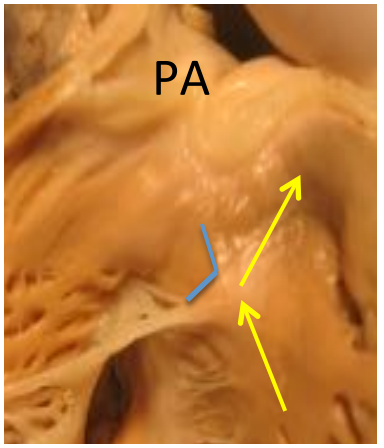
	ccTGA	TGA	Normal heart	p
Angle AL/PL	76°4	90°8 *	76°1	0.01
Angle AL/ arterial valve	70°6	90°6 *	69°1	0.0004
Ratio AL/PL	3.7 *	2.3	1.5	0.0003



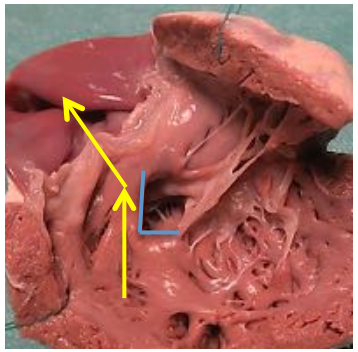
- Posterior limb shorter in ccTGA: illusion of an inlet VSD

The anatomy of the right ventricle in ccTGA

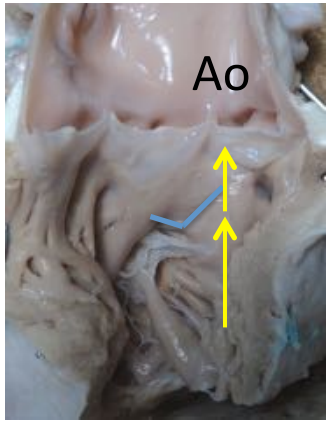
- The goemetry of the outflow tract is similar (but mirror-imaged) in NH and in ccTGA, despite the VA discordance
- In TGA, the outflow tract is straight (no rotation)
- ccTGA is not a TGA!!!
- Could this explain the better longevity of the systemic RV in ccTGA vs TGA post-atrial switch?



NH



ccTGA

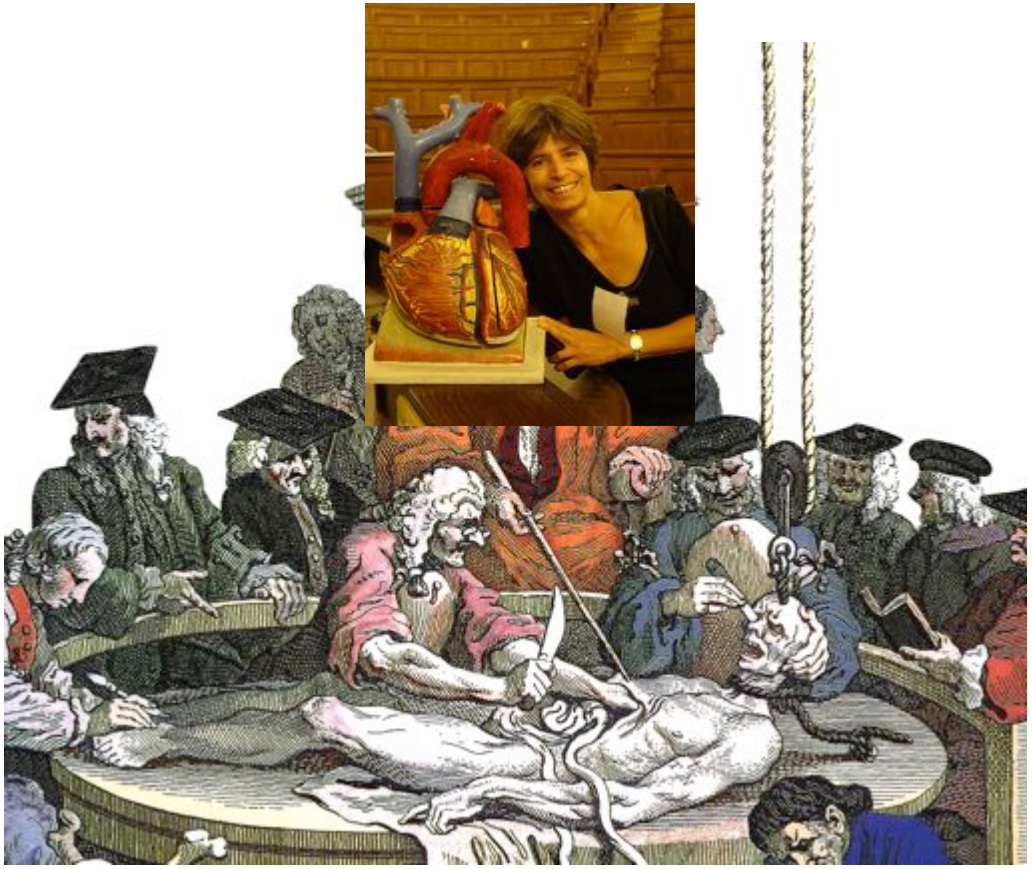


TGA

L'anatomie des cardiopathies congénitales : Imaginer demain

M3C

Cardiac specimens..
A thing of the past?



Courtesy
TWIN MEDICAL