



CARDIOPATÍAS CONGÉNITAS



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DRA. ALEXIS STRICKLER – PEDIATRA DOCENTE USS

HOJA DE RUTA

- Introducción.
- Epidemiología.
- Asociaciones Sindromáticas.
- Factores de Riesgo.
- Clasificación:
 - Cianóticas.
 - No Cianóticas.
- Resumen.
- Conclusiones.
- Bibliografía.



INTRODUCCIÓN

- Las **causas de cianosis** son variadas y multisistémicas.
- Dentro de las causas cardíacas, un porcentaje esta relacionado a **Cardiopatías Congénitas (CC)**.
- Malformaciones cardíacas se originan en las **primeras 8 a 10** semanas de gestación.

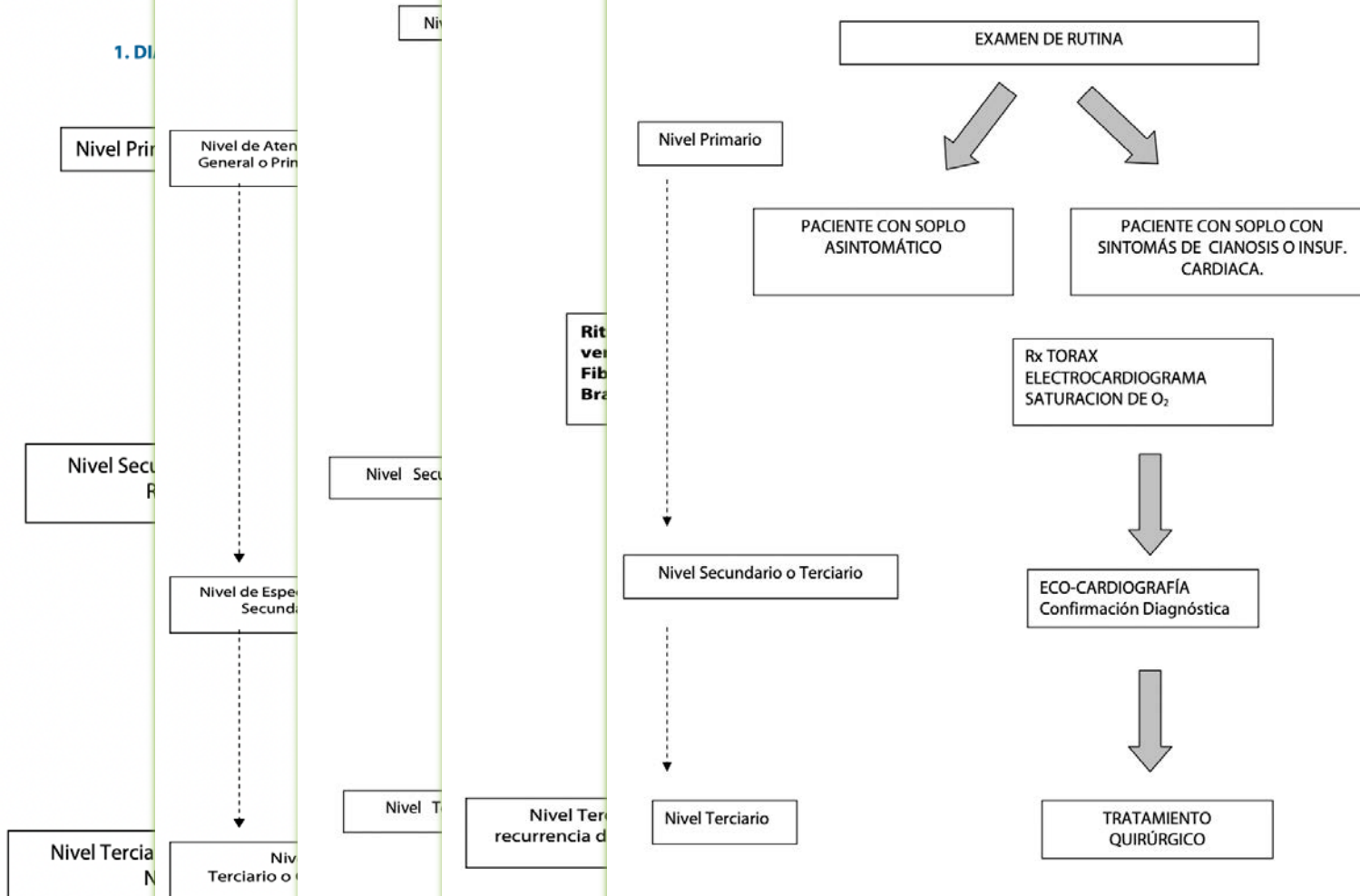


Causes of central cyanosis in the neonate

Disease category	Primary underlying mechanism
Airway obstruction	
Choanal atresia	Hypoventilation
Laryngotracheomalacia	
Macroglossia	
Micrognathia or retrognathia (eg, Pierre-Robin syndrome)	
Cardiac	
Congenital cyanotic heart disease	Right-to-left shunting
Heart failure/pulmonary edema	Impaired alveolar-arterial diffusion and V/Q mismatch
Hematologic	
Hemoglobinopathies (eg, methemoglobinemia)	Impaired oxygen saturation
Polycythemia	Elevated hemoglobin resulting in low oxygen saturation
Metabolic	
Severe hypoglycemia	Hypoventilation due to decreased or absent respiratory effort secondary to lethargy, seizures, and/or apnea
Inborn errors of metabolism	
Neurologic	
Central nervous system depression	
Apnea of prematurity	Hypoventilation
Infection (eg, meningitis, encephalitis)	
Intraventricular hemorrhage	
Maternal sedation	
Seizure	
Neuromuscular disorder	
Neonatal myasthenia gravis	Hypoventilation
Phrenic nerve injury	
Spinal muscular atrophy type 1 (Wernig-Hoffman disease)	
Pulmonary	
Parenchymal disease	
Atelectasis	V/Q mismatch
Alveolar capillary dysplasia	
Lobar emphysema	
Pneumonia	
Pulmonary hypoplasia	
Pulmonary hemorrhage	
Respiratory distress syndrome (Hyaline membrane disease)	
Transient tachypnea of the newborn	
Pulmonary fibrosis	Impaired alveolar-arterial diffusion
Pulmonary edema	Impaired alveolar-arterial diffusion and V/Q mismatch
Nonparenchymal disease	
Pleural effusion	V/Q mismatch
Pneumothorax	
Other	
Persistent pulmonary hypertension of the newborn	Right-to-left shunting

2.4. CARDIOPATÍA CONGÉNITA CON ARRITMIAS

SOPLOS



EPIDEMIOLOGÍA CC:

- Representan el **10% de las malformaciones congénitas**.
- Según American Heart Association (AHA), 9 de cada 1.000 RN en USA tienen una cardiopatía congénita **(0,5-1%): defectos de nacimiento más comunes**.
- **Reincidencia:** probabilidad de tener 2° hijo con CC aumenta en un 3%, pero tienen un 97% de tener un hijo sano.
- Se asocia en un **44% a otras malformaciones:** Digestivo, Esquelético, SNC o Renal.
- **Lesiones cianóticas** comprenden aproximadamente **un tercio** de las formas potencialmente mortales de cardiopatía congénita.



ASOCIACIONES SINDROMÁTICAS CARDIOPATÍAS CONGÉNITAS



Selected genetic disorders associated with cardiovascular malformations

Syndrome	Cardiovascular anomaly
Genetic associations	
VATER/VACTERL association	Wide-ranging (including VSD, TOF, TGA, and others)
Chromosomal syndromes	
Down syndrome (trisomy 21)	AV canal defect, VSD, ASD, TOF, PDA, pulmonary hypertension
Edwards syndrome (trisomy 18)	ASD, VSD, PDA, PS, AS, CoA, TOF, HLHS, pulmonary hypertension
Patau syndrome (trisomy 13)	ASD, VSD, TOF, PS, AS, CoA, HLHS, DORV, pulmonary hypertension
Turner syndrome	Aortic valve abnormalities, CoA, systemic and pulmonary venous abnormalities, VSD, HLHS, ASD, aortic dilation/rupture
Chromosomal deletion/microdeletion syndromes	
DiGeorge syndrome (22q11 deletion syndrome)	TOF, IAA and other aortic arch anomalies, truncus arteriosus, VSD
Williams syndrome	Pulmonary and aortic valve defects, ASD, VSD, coronary ostial stenosis, branch pulmonary artery stenosis, arteriopathy
Single-gene syndromes – Autosomal dominant inheritance pattern	
Alagille syndrome	Peripheral pulmonary artery stenosis, ASD, VSD, TOF, CoA
Albright hereditary osteodystrophy	Cardiomyopathy
Cardiofaciocutaneous syndrome	PS, ASD, VSD, HCM
CHARGE syndrome	VSD, ASD, TOF, DORV, PDA, PS, pulmonary vein anomalies
Costello syndrome	HCM, PS, ASD, VSD, atrial arrhythmias
Ehlers-Danlos syndrome (vascular type)	Rupture of large vessels
Holt-Oram syndrome	ASD, VSD, left-sided lesions, conotruncal defects, AV block
Leopard syndrome	PS, prolonged PR interval, ASD, VSD, HCM
Marfan syndrome	Aortic root dilation/aneurysmal dissection, AI, MVP
Myotonic dystrophy	Cardiomyopathy
Neurofibromatosis	CoA, renal artery stenosis
Osler-Weber-Rendu disease	Multiple telangiectasis, pulmonary AVM
Treacher Collins syndrome	ASD, VSD, PDA
Tuberous sclerosis	Myocardial rhabdomyoma, Wolff-Parkinson-White syndrome
Noonan syndrome	PS, ASD, AS, subaortic stenosis, HCM
Single-gene syndromes – Autosomal recessive inheritance pattern	
Carpenter syndrome	PDA
Cutis laxa	Pulmonary hypertension
Ellis-van Creveld syndrome	ASD, AVSD
Friedreich ataxia	Cardiomyopathy
MPS type IH (Hurler syndrome)	AI, MR, premature CAD, cardiomyopathy
MPS type IS (Scheie syndrome)	Aortic valve disease
MPS type IV (Morquio syndrome)	Aortic valve disease
MPS type VI (Maroteaux-Lamy syndrome)	Aortic valve disease
Pompe disease (GSD type 2)	Cardiomyopathy
Pseudoxanthoma elasticum	Premature CAD, MVP
Smith-Lemli-Opitz syndrome	VSD, PDA, HLHS
Thrombocytopenia absent radii syndrome	ASD, TOF
Single-gene syndromes – X-linked inheritance pattern	
MPS II (Hunter syndrome)	Valvular disease, premature CAD, cardiomyopathy
Duchenne muscular dystrophy	Cardiomyopathy
Emery-Dreifuss muscular dystrophy	Cardiomyopathy
X-linked heterotaxy	Heterotaxy
Incontinentia pigmenti	PDA, hypertension
Genetic mutations associated with nonsyndromic congenital heart disease	
NKX2-5 mutation	ASD, VSD, TOF, HLHS
GATA4 mutation	ASD, VSD, TOF

CHD: congenital heart disease; OR: odds ratio; RVOTO: right ventricular outflow tract obstruction; VSD: ventricular septal defect; ASD: atrial septal defect; PDA: patent ductus arteriosus; RR: relative risk; PKU: phenylketonuria; CoA: coarctation of the aorta; HLHS: hypoplastic left heart syndrome; AVSD: atrioventricular septal defect; LVOTO: left ventricular outflow tract obstruction; ACE: angiotensin-converting enzyme; NSAID: nonsteroidal antiinflammatory drug; D-TGA: D-transposition of the great arteries.

FACTORES DE RIESGO

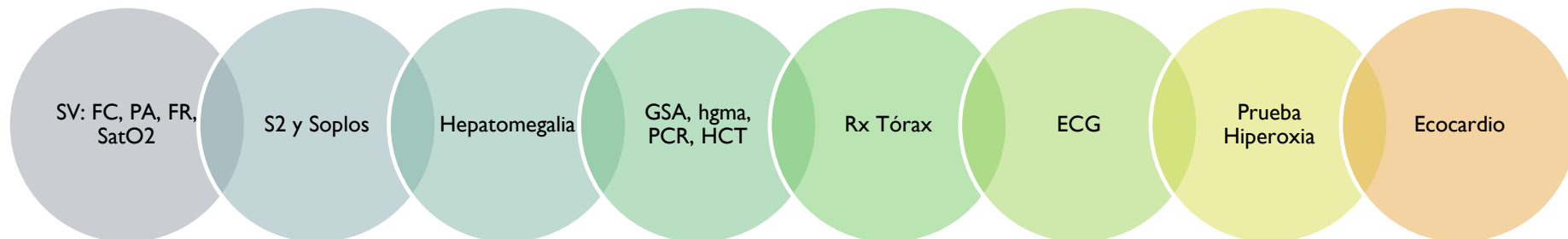


Factors associated with an increased risk of congenital heart disease

	Estimated risk*	Associated CHD lesions
Prematurity (gestational age <37 weeks)^[1]	OR 2.4 (95% CI 2.2-2.7)	Various
Multifetal pregnancy^[2]	OR 4.53 (95% CI 4.28-4.8)	RVOTO lesions, VSD, ASD
In utero infection		
Rubella ^[3]	Cardiac defects occur in 10 to 20% of infants with congenital rubella	PDA and peripheral pulmonary artery stenosis
Maternal influenza or flu-like illness ^[4]	OR 2.04 (95% CI 1.27-3.27)	RVOTO lesions
Maternal factors^[2]		
Preeclampsia ^[5]	RR 1.57 (95% CI 1.48-1.67)	Septal defects
PKU ^[6]	Cardiac defects occur in 15% of infants with poorly controlled maternal PKU	CoA and HLHS
Diabetes mellitus	OR 4.65 (95% CI 4.13-5.24)	Heterotaxy syndrome, conotruncal defects, AVSD, LVOTO and RVOTO lesions
Hypertension	OR 1.81 (95% CI 1.61-2.03)	RVOTO, VSD, ASD
Obesity	OR 1.48 (95% CI 1.32-1.65)	Various
Thyroid disorders	OR 1.45 (95% CI 1.26-1.67)	Various
Systemic connective tissue disorders	OR 3.01 (95% CI 2.23-4.06)	Heterotaxy syndrome
Epilepsy and mood disorders [¶]	OR 1.41 (95% CI 1.16-1.72)	Various
Age ≥40 years	OR 1.48 (95% CI 1.39-1.58)	AVSD
Alcohol or substance use	OR 1.88 (95% CI 1.74-2.04)	RVOTO, VSD, ASD
First-trimester cigarette smoking ^[7]	OR 1.9 (95% CI 1.04-3.45) OR 1.32 (95% CI 1.06-1.65) OR 1.36 (95% CI 1.04-1.78)	Truncus arteriosus RVOTO Secundum ASD
Maternal medications during pregnancy ^[6,8] : Increased risk of CHD has been reported with thalidomide, ACE inhibitor, retinoic acid, NSAIDs, phenytoin, and lithium	Estimates vary	Various NSAIDs are associated with D-TGA, AVSD, VSD, bicuspid aortic valve Lithium has been reported to be associated with Ebstein anomaly ⁴
Assisted reproductive technology^[9]	RR 1.64 (95% CI 1.30-2.07)	Various
Family history of CHD^[10]		
First-degree relative with any CHD	RR 3.21 (95% CI 2.96-3.49)	Various
Second-degree relative with any CHD	RR 1.78 (95% CI 1.09-2.91)	Various
First-degree relative with heterotaxy	RR 79.1 (95% CI 32.9-190.0)	Heterotaxy
First-degree relative with RVOTO	RR 48.6 (95% CI 27.5-85.6)	RVOTO
First-degree relative with AVSD	RR 24.3 (95% CI 12.2-48.7)	AVSD
First-degree relative with LVOTO	RR 12.9 (95% CI 7.48-22.20)	LVOTO
First-degree relative with conotruncal defect	RR 11.7 (95% CI 8.01-17.00)	Conotruncal defect
First-degree relative with isolated ASD	RR 7.07 (95% CI 4.51-11.10)	ASD
First-degree relative with isolated VSD	RR 3.41 (95% CI 2.20-5.29)	VSD

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ENFRENTAMIENTO INICIAL



- Importancia: Anamnesis, Ex. Físico → búsqueda orientada a **Sepsis, patología Pulmonar y Cardíaca.**

- **CC no cianótica:**

- Dificultades alimentación.
- Cambios de color sutiles en el color (palidez).
- Irritabilidad excesiva e inexplicable.
- Sudoración que aumenta con alimentación y sueño.
- Mal incremento ponderal.
- Disminución de actividad e hipersomnia.
- RDSM.

- **CC Cianótica:** Tratamiento inmediato:

- Oxígeno.
- Volumen y DVA: Dobutamina + Dopamina.
- ATB post HCT: Ampicilina + Gentamicina.
- Corrección Ácido Base, glicemia.
- PGEI

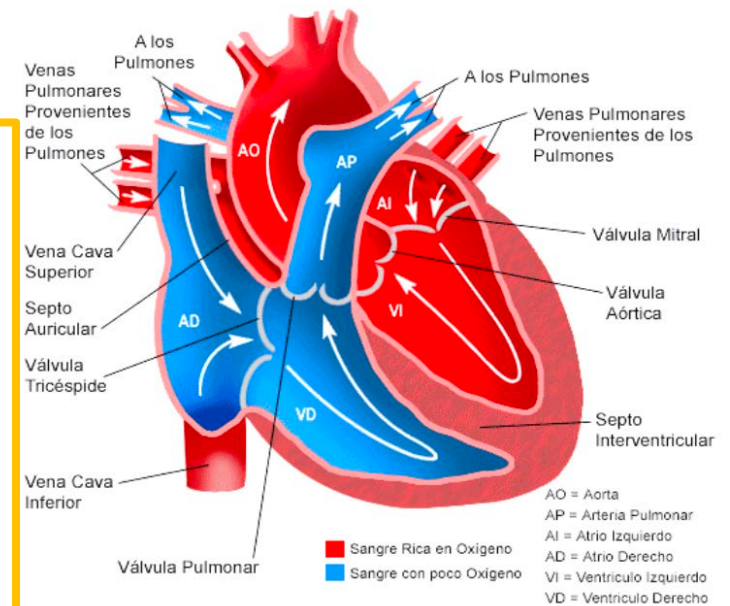
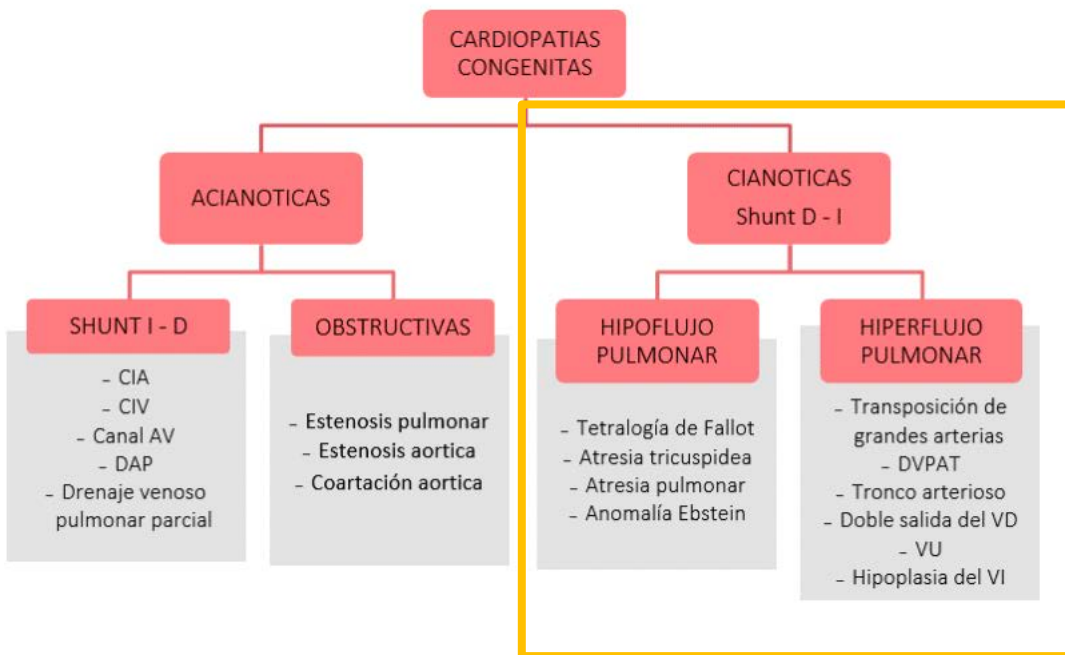
Typical physical examination, chest radiography, and electrocardiography findings in some forms of cyanotic heart disease

N: +30+180

Diagnosis	Physical examination		Chest radiography			Electrocardiogram		Incidence (per 100,000 live births)*
	S2	Murmur	Heart size	PBF	Percent RAA	QRS axis	Hypertrophy	
D-TGA	Single	None	↑	↑	4	90 to 150	nml	21
TOF	Single	Systolic	Boot	↓	20	90 to 150	nml	20 to 26
HLHS	Single	+/- systolic	↑	Venous congestion		90 to 150	↓ LV forces	16
PA-IVS	Single	Systolic	↑↑	↓		30 to 90	LVH, RAE	7
Pulmonary stenosis	Single	Systolic	↑	↓		30 to 90	RVH, RAE	7
TAPVC	Split	Systolic	↑, normal*	↑, venous congestion*		90 to 150	RAE	6
Tricuspid atresia	Single	+/- systolic	↑	↓		-30 to -90	LVH, RAE	6
Truncus arteriosus	Single	Systolic +/- diastolic	↑	↑	30	90 to 150	nml	3
Ebstein	Split	Systolic	↑↑	↓		90 to 150	RAE	1

S2: second heart sound; PBF: pulmonary blood flow; RAA: right aortic arch; D-TGA: D-transposition of the great arteries; TOF: tetralogy of Fallot; HLHS: hypoplastic left heart syndrome; PA-IVS: pulmonary atresia intact ventricular septum; TAPVC: total anomalous pulmonary venous connection; nml: normal for neonate (right ventricular predominance); LV: left ventricular; LVH: left ventricular hypertrophy; RAE: right atrial enlargement; RVH: right ventricular hypertrophy.

CLASIFICACIÓN



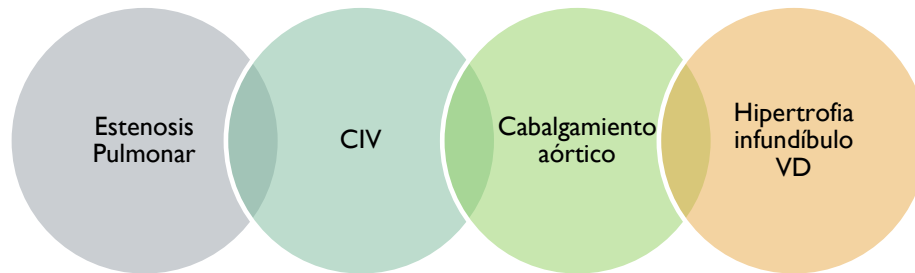
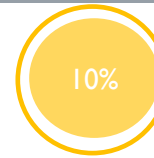


CARDIOPATÍAS CONGÉNITAS

I. CIANÓTICAS

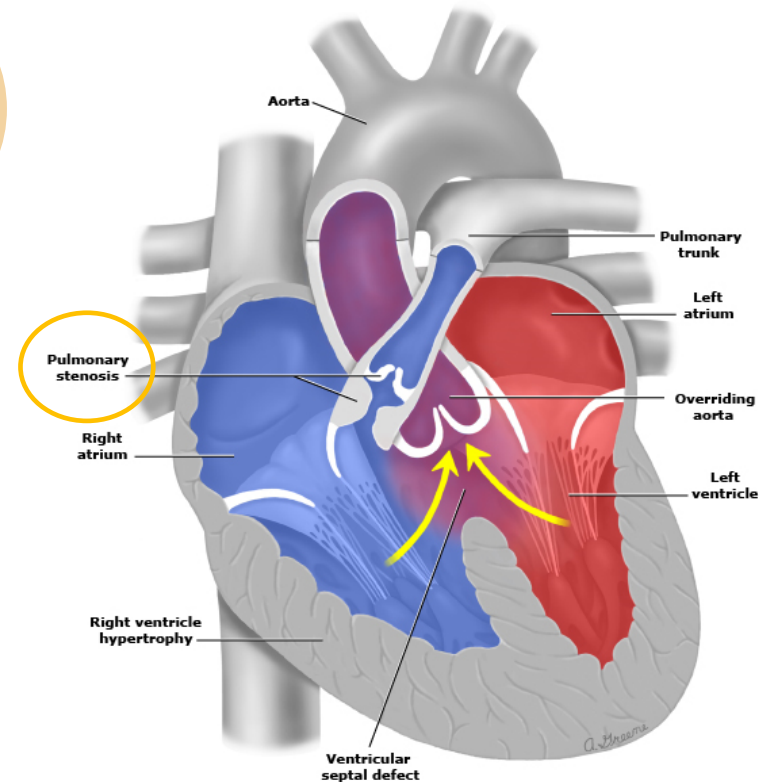
I.I. CCC: HIPOFLUJO PULMONAR

A) TETRALOGÍA FALLOT



- **Estenosis pulmonar leve:** Shunt I-D → FALLOT ROSADO (no cianóticos: estenosis pulmonar suficiente para invertir shunt).
- **Estenosis pulmonar severa:** Shunt D-I → FALLOT CIANÓTICO (estenosis pulmonar grave con hipoflujo pulmonar que invierte el shunt).

Anatomy of tetralogy of Fallot



Tetralogy of Fallot is characterized by a large VSD, an aorta that overrides the left and right ventricles, obstruction of the right ventricular outflow tract, and right ventricular hypertrophy. As a result of the substantial obstruction of the right ventricular outflow tract, there is right-to-left shunting through the VSD, resulting in cyanosis.

VSD: ventricular septal defect.

I.I. CCC: HIPOFLUJO PULMONAR

A) TETRALOGÍA FALLOT

Clínica

- Al nacer → cianosis variable, según obstrucción TSVD.
- R2 aumentado y Soplo sistólico eyectivo PEI por TSVD.

Radiografía

- Vascularización pulmonar disminuida.
- Silueta cardíaca tamaño normal pero segmento cóncavo AP con el ápex del corazón inclinado hacia arriba → “en bota”.

ECG

- HVD, eje +120-150 → en un RN esto podría pasar como algo normal.

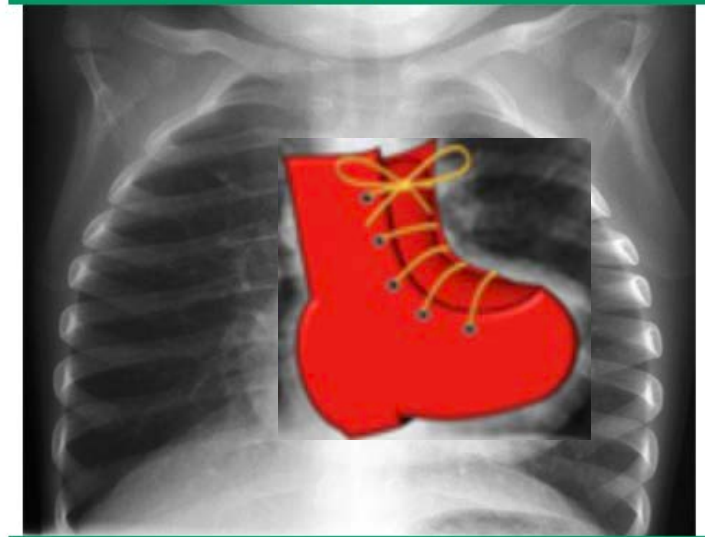
Tratamiento médico

- Oxigenación 100%, sedación con morfina 0.1mg/kg, tratar acidosis con Ketamina, vasoconstrictores, propranolol.

Tratamiento quirúrgico

- Cirugía convencional (3-4 meses): cierre CIV con parche, ensanchamiento TSVD hipertrofico con resección del tejido muscular infundibular.

Chest radiograph of tetralogy of Fallot



This chest radiograph demonstrates some of the classic findings of tetralogy of Fallot. The lungs are hyperinflated, and there is overall decreased pulmonary vascularity. There is a right aortic arch. The cardiac apex is upturned due to right ventricular enlargement.

I.I. CCC: HIPOFLUJO PULMONAR

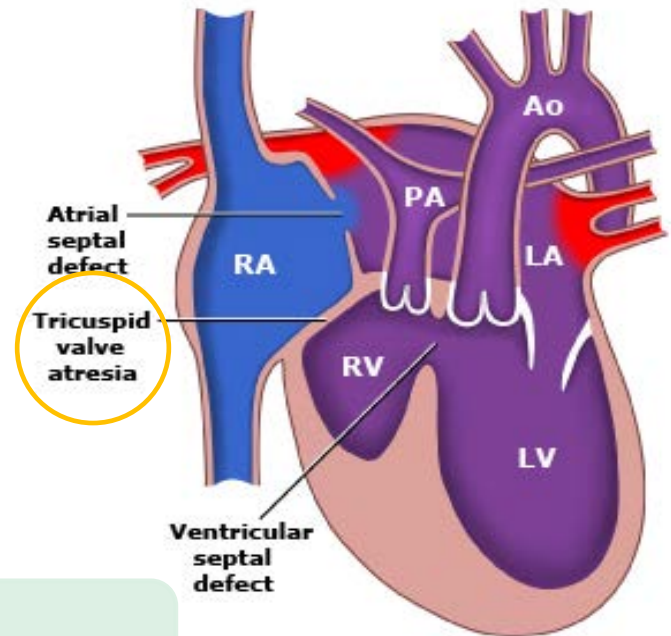
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B) ATRESIA TRICUSPÍDEA



FISIOPATOLOGÍA:

- Válvula tricúspide atrésica → ausencia flujo VD → sin desarrollo tronco A. Pulmonar → VD y AP hipoplásicos.
- 30%. Asociación a TGA.
- Defectos asociados para supervivencia: CIA, CIV o DAP dependiente.
- **Flujos:** Retorno venoso sistémico va desde AD a AI (hipertrofia AD) y de la aorta a la pulmonar por el ductus.



Tratamiento médico

- RN PGE2: mantener ductus abierto.

Tratamiento quirúrgico

- Septostomía auricular balón Rashkind: si CIA chica.
- Cx transitoria Glenn: anastomosis VCS a AP D°.
- Cx definitiva "Fontan": Glenn+ anastomosis VCI a AP.

I.I. CCC: HIPOFLUJO PULMONAR

C) ATRESIA PULMONAR

<1%

- Válvula Pulmonar Atrésica → CIA y Ductus dependiente.

Pulmonary atresia with intact ventricular septum (PA/IVS)

Clínica

- Signos de ICC agudos o subagudos según defectos asociados CIA/ductus.

Radiografía

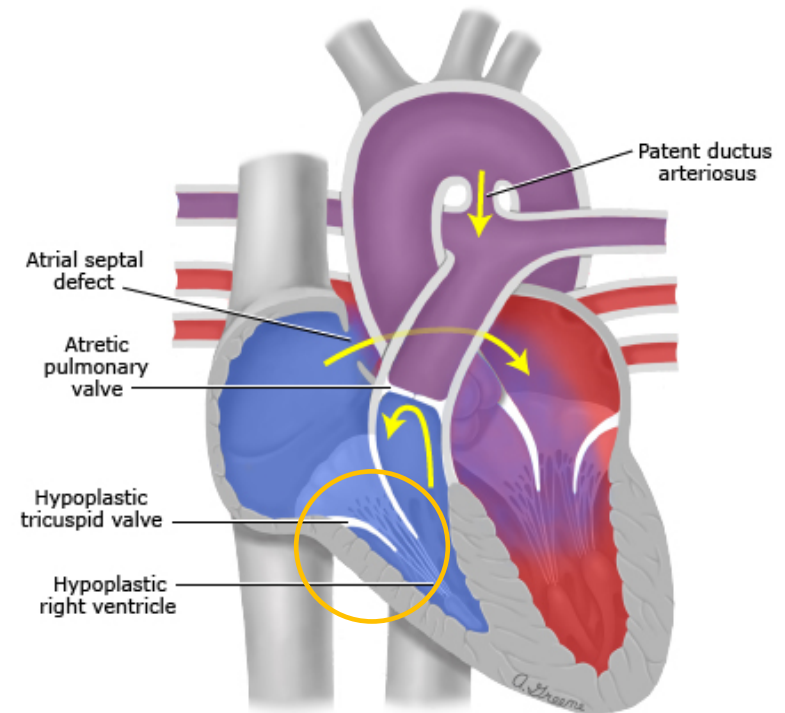
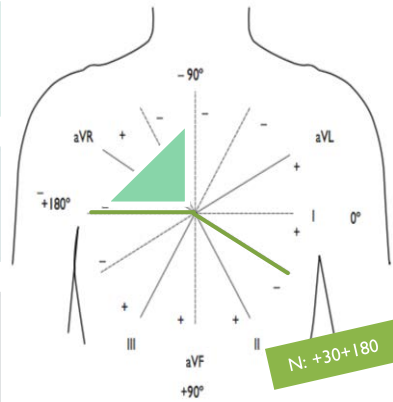
- Cardiomegalia.

ECG

- Eje -90-150.

Tratamiento

- PGE2.
- Septotomía CIA.
- Reconstitución VP.



I.I. CCC: HIPOFLUJO PULMONAR

<1%

D) ANOMALÍA EBSTEIN

Desplazamiento
V. Tricúspide
cámara de
entrada VD

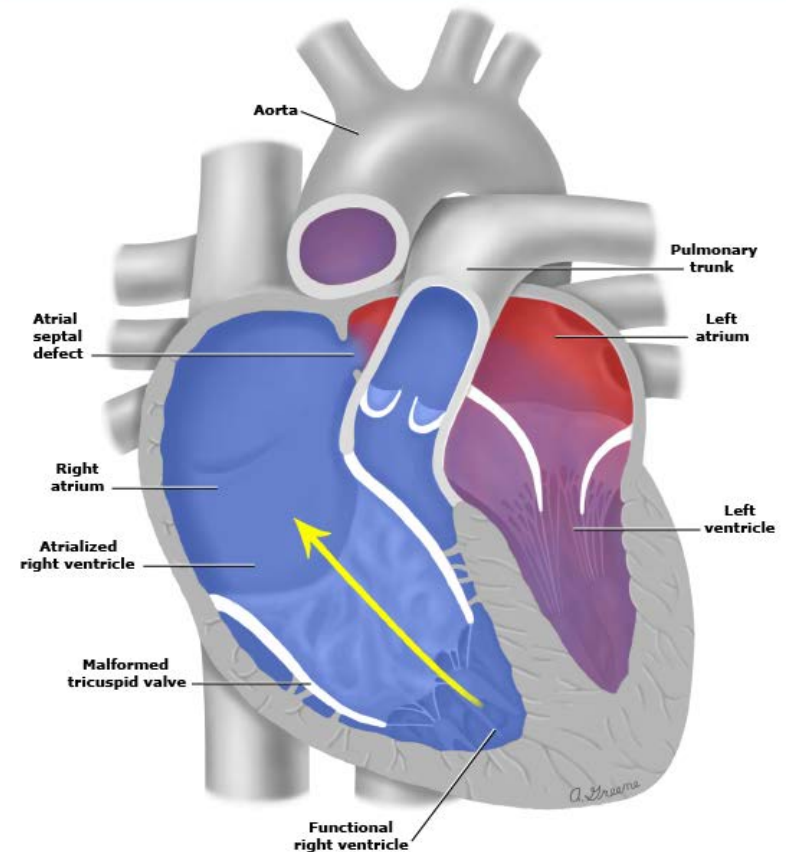
AD dilatada e
hipertrofiada,
con VD
hipoplásico

CIA dependiente

Asociado a
WPW

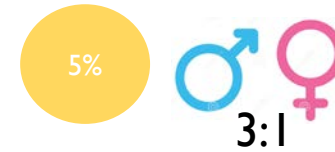
- **Tratamiento médico:** RN gravemente cianóticos
→ PGEI, inótrpos y diuréticos.
- **Tratamiento quirúrgico:**
 - Anuloplastia tricúspide: de elección.
 - Sustitución valvular protésica o biológica + cierre CIA.
 - En VD muy hipoplásico: operación de Fontan y desfuncionalizar VD.
 - En Sd WPW: exéresis de la vía accesoria.

Pathophysiology of Ebstein anomaly



I.2. CCC: HIPERFLUJO PULMONAR

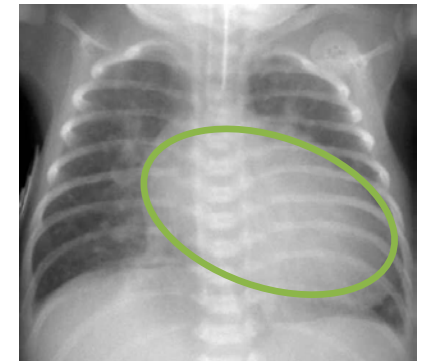
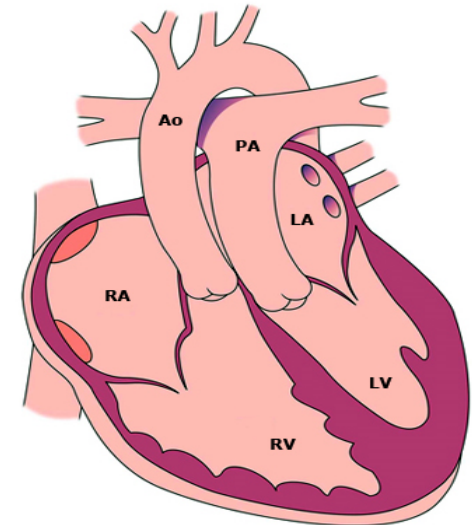
A) TRANSPOSICION GRANDES ARTERIAS (TGA)



FISIOPATOLOGÍA:

- Aorta surge del VD y AP pulmonar del VI.
 - Flujo desde VD a circulación sistémica → vuelve por venas cavas hacia AD.
- Separación total de los dos circuitos unidos por defectos asociados, necesarios para supervivencia (CIV, CIA, DAP).
- Defectos asociados: CIV 40%, Estenosis pulmonar 30 - 35%.

Diagram of the dextro type of transposition of the great arteries



Clínica

- Cianosis moderada a grave, Taquipnea, signos ICC.
- R2 aumentado, sin soplos.

Radiografía

- Corazón forma de huevo + Congestión pulmonar + Mediastino estrecho.

ECG

- HVD o Hipertrofia biventricular.

Tratamiento médico

- RN PGE2 (mantener ductus) y O2 (disminuir RVP).

Tratamiento quirúrgico

- Rashkind: septostomía para ampliar CIA.
- Switch arterial.

I.2. CCC: HIPERFLUJO PULMONAR

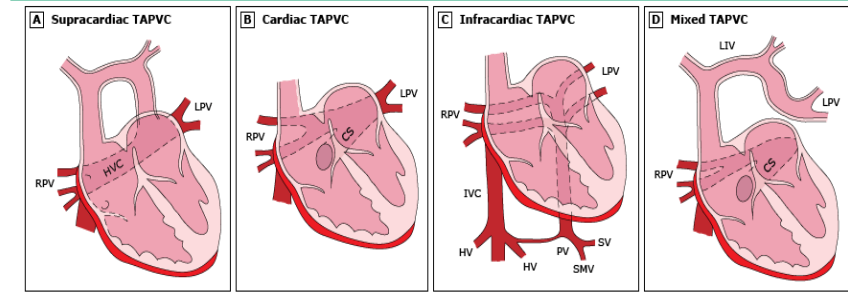
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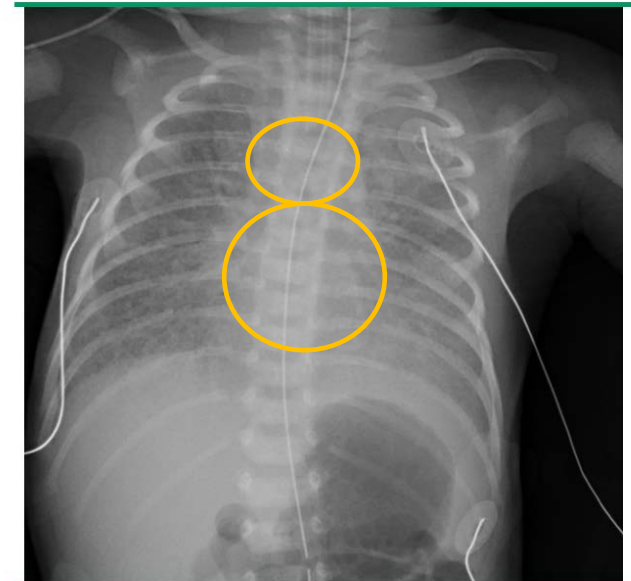
B) DRENAJE VENOSO ANÓMALO PULMONAR TOTAL (DVAPT)

- **Fisiopatología:** Drenaje venoso llega al corazón D° (CVS,VCI,AD) → CIA dependiente.
- **Clasificación:**
 - a) Supracardiaco 50%: VCS.
 - b) Cardíaco 20%: Seno coronario o AD.
 - c) Infracardiaco 20%: V. Porta,V. Hepática o VCI.
 - d) Mixto 10%.

Diagram of common types of totally anomalous pulmonary venous connection



Obstructed total anomalous pulmonary venous connection



Chest radiograph of a newborn infant with obstructed infracardiac total anomalous pulmonary venous connection (TAPVC). The lungs have a ground glass appearance due to pulmonary vascular congestion and diffuse interstitial and alveolar edema.

UpToDate®

I.2. CCC: HIPERFLUJO PULMONAR

C) TRONCO ARTERIOSO

<1%

- **Tronco único da lugar a circulación pulmonar, sistémica coronaria.**
- **Anomalías asociadas:**
 - Siempre CIV de gran tamaño.
 - 30% asociada a arco aórtico derecho.
 - 30% asociado a Sd. de Di George (microdelección crom 21).

Clínica

- Signos de ICC.

Radiografía

- Cardiomegalia.

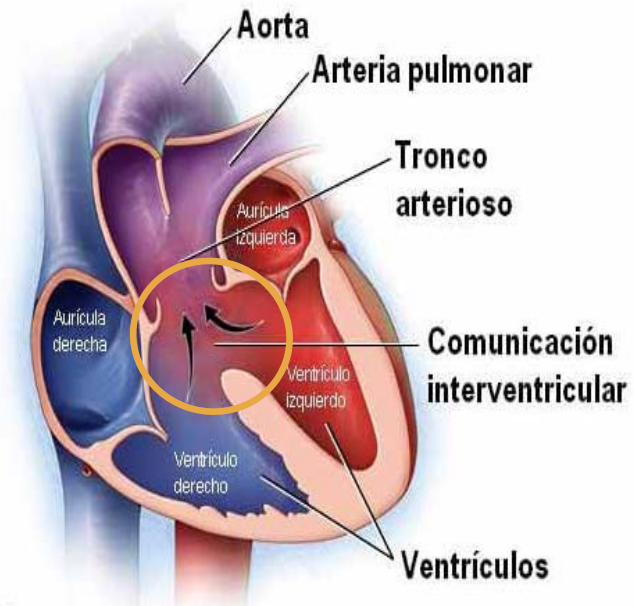
Tratamiento médico

- Medidas anticongestivas con digitálicos y diuréticos.

Tratamiento quirúrgico

- Tronco va a quedar como una aorta, realizando una arteria pulmonar nueva.

Tronco arterioso



I.2. CCC: HIPERFLUJO PULMONAR

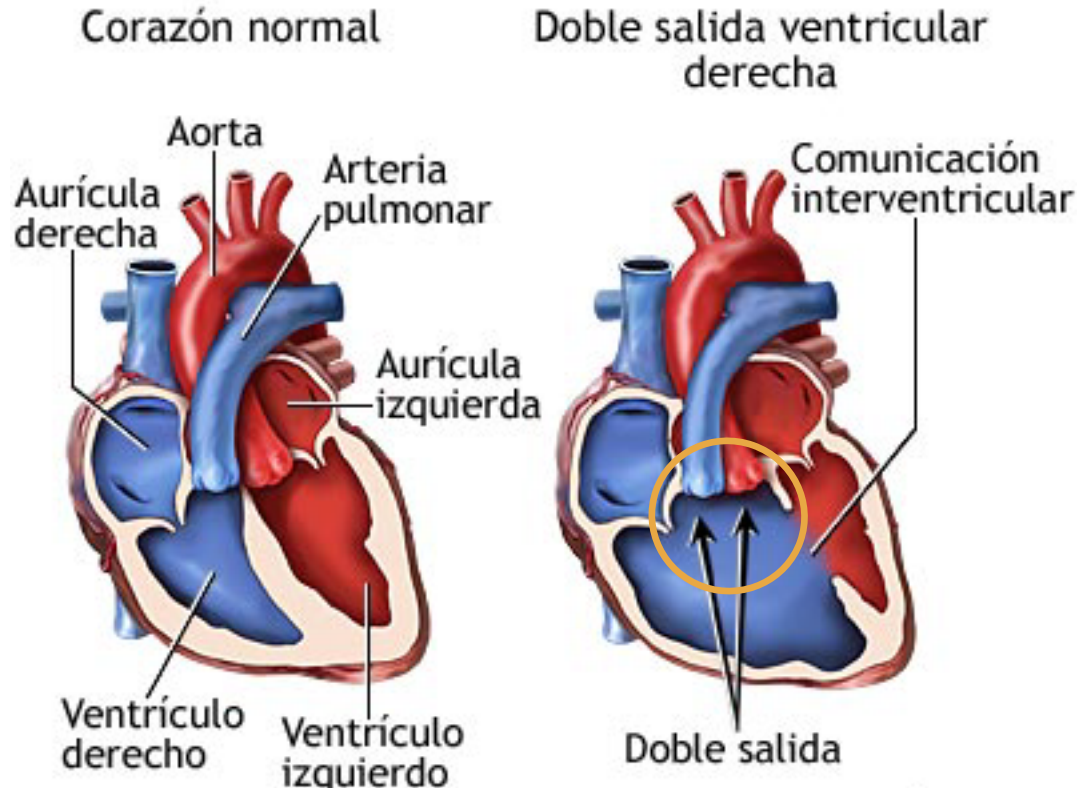
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D) DOBLE SALIDA VD

Aorta y AP se originan del VD.

CIV dependiente: para que exista flujo sistémico.

Generalmente hay un VD más grande y un VI más pequeño.



I.2. CCC: HIPERFLUJO PULMONAR

<1%

E) VENTRÍCULO ÚNICO

- Ambas válvulas AV drenan hacia una única cavidad ventricular.
- No existe tabique interventricular.
- 85% con TGA asociada.
- **Clasificación:**
 - VUD: anatomía predominante VD.
 - VUI: anatomía predominante VI (75% con TGA).
 - VU: indeterminado.

Clínica

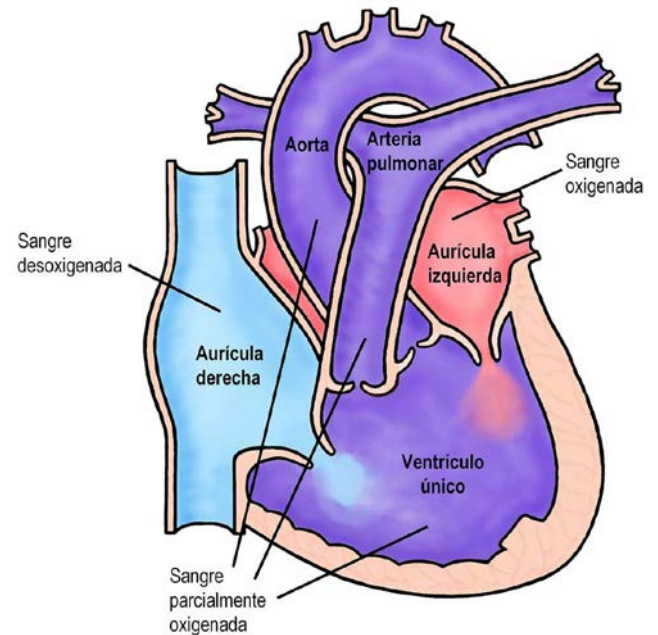
- Signos de ICC.

Radiografía

- Cardiomegalia.

Tratamiento

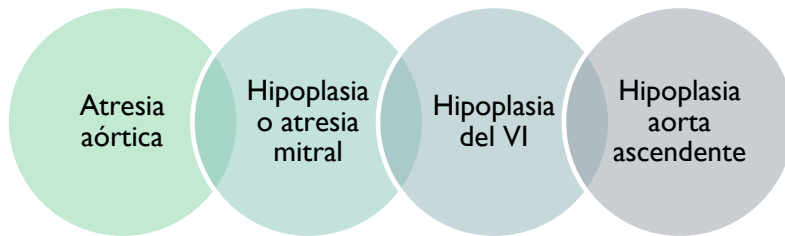
- Medidas anticongestivas con digitálicos y diuréticos, previo a Cx.



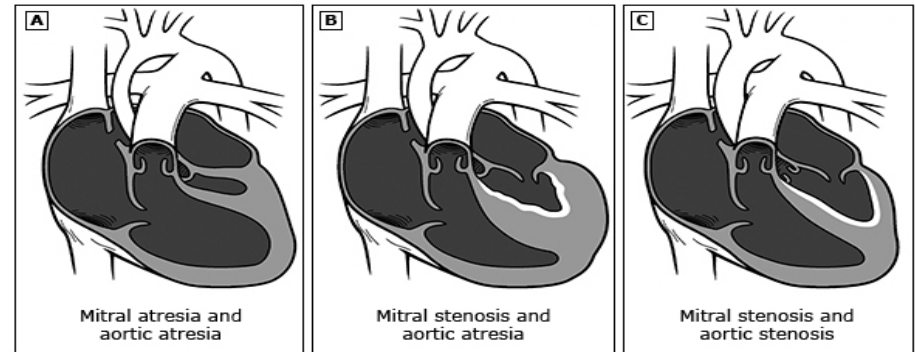
I.2. CCC: HIPERFLUJO PULMONAR

<1%

F) HIPOPLASIA VENTRICULO IZQUIERDO



Spectrum of hypoplastic left heart syndrome



FISIOPATOLOGÍA:

- Circulación Sistémica por AP mediante DAP → HTP e hipertrofia VD.
- DAP y CIA dependiente.
- Urgencia quirúrgica: deben ser operados antes de 15 días de vida.
- Tratamiento quirúrgico en etapas:
 - 1º etapa en la paliación quirúrgica (Norwood: nueva aorta con AP).
 - 2º etapa a los 6 meses (Glenn bidireccional).
 - 3º etapa entre los 2 y 3 años (Fontan).



CARDIOPATÍAS CONGÉNITAS

I. NO CIANÓTICAS

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- Robert L Geggel. “Diagnosis and initial management of cyanotic heart disease in the newborn”. En Uptodate, Agosto de 2021.
- Carolyn A. Altman “ Identification of newborns with critical congenital heart disease”. En uptodate, 15 de Septiembre de 2020.
- Guía Clínica MINSAL: Cardiopatías Congénitas operables en < 15 años 2010.
- <https://www.stanfordchildrens.org/es/topic/default?id=congenitalheartdisease-90-P05455>