

LE CARDIOPATIE CONGENITE

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MALATTIE EREDITARIE**Sindrome di Ellis-Von-Creveld****Sindrome di Holt-Oram****Sindrome di Kartagener****Sindrome di Noonan****Sindrome di Leopard****Distrofie muscolari****CC ASSOCIATA****Pervietà del SIA****Pervietà del SIA****Destrocardia****Stenosi polmonare****Stenosi polmonare****Cardiomiopatia dilatativa****MALATTIE DEL CONNETTIVO****Sindrome di Marfan****Sindrome di Ehlers-Danlos****Dilatazione aortica****Insufficienza aortica e mitralica****Dilatazioni arteriose****MALATTIE METABOLICHE****Malattia di Pompe****Omocistinuria****Mucopolisaccaridosi****Cardiomiopatia dilatativa****Dilatazione grossi vasi****Insufficienza aortica – CMD****DISORDINI CROMOSOMICI****Sindrome di Down****Sindrome di Turner****Pervietà settali – TF – CAV****Coartazione aortica**

Principali fattori eziologici alla base delle cardiopatie congenite

Fattori genetici primitivi

geni con carattere di dominanza
geni recessivi

Infezioni materne

rosolia
toxoplasmosi

Farmaci teratogeni

anfetamine
idantoinici
talidomide
litio
warfarin
ormoni sessuali

Condizioni materne

abuso di alcool
diabete
LES
fenilchetonuria

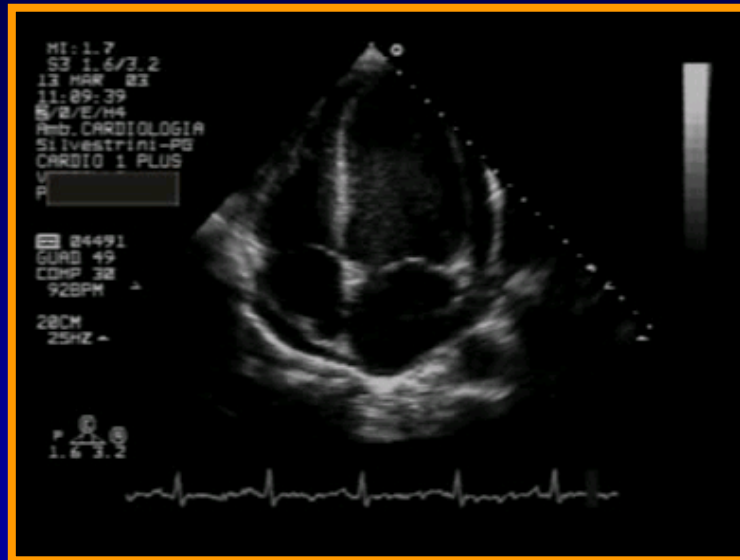
Condizioni ambientali

ipossia da altitudini
radiazioni ionizzanti

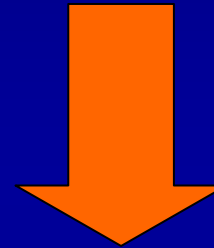
Interazioni genetico-ambientali

Frequenza percentuale delle principali cardiopatie congenite

DIV	25 - 40 %
DIA	10 - 15 %
Persistenza DA	5 - 15 %
Stenosi polmonare	7 - 8 %
Coartazione aortica	5 - 13 %
Tetralogia di Fallot	5 - 10 %
Stenosi aortica	3 - 6 %
Trasposizione corretta GV	2 - 7 %
Tronco arterioso comune	1 - 4 %
Atresia della tricuspid	1 - 2 %



REGOLA



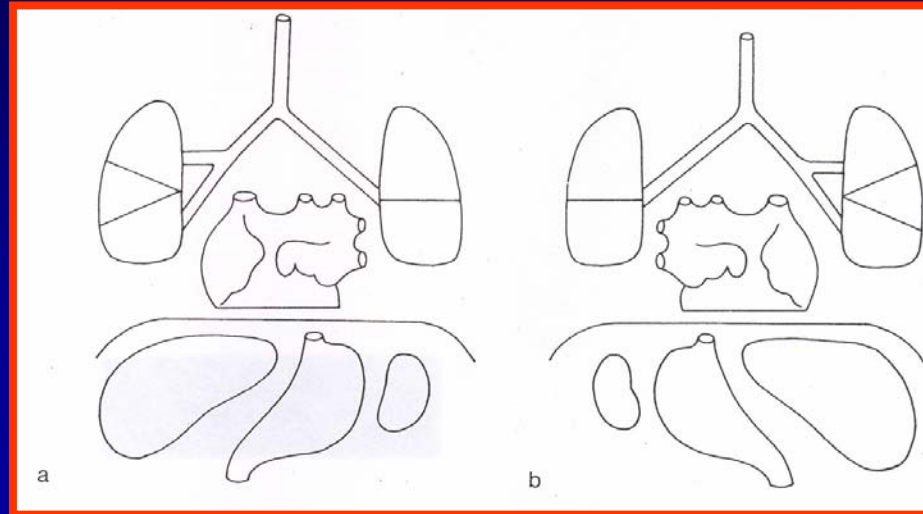
Situs addominale = Situs toracico = Situs atriale

Auricola dx : larga, tozza, a larga base di impianto

Auricola sx : stretta, lunga, a dito di guanto

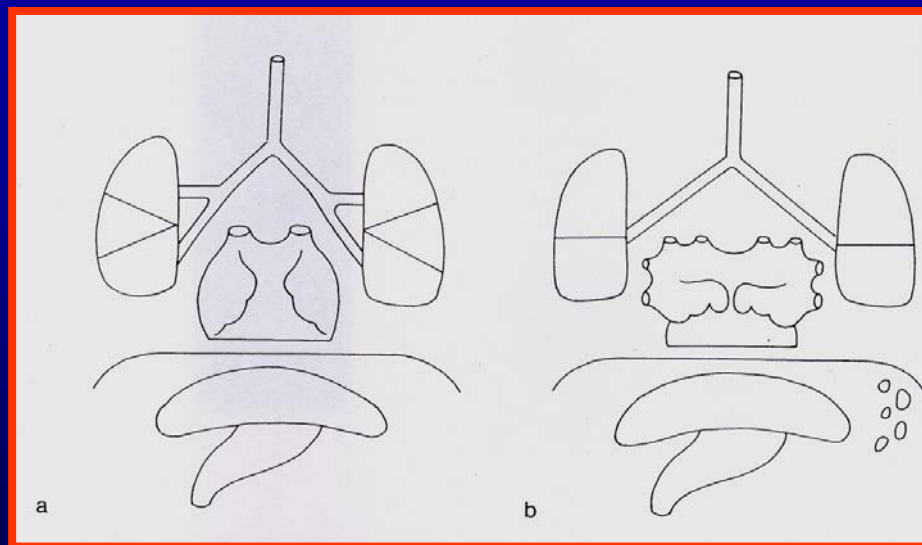
SITUS VISCERO-ATRIALE

Situs solitus



Situs inversus

Isomerismo dx



Isomerismo sx

GIUNZIONE ATRIO-VENTRICOLARE:

tipo di connessione

modo di connessione

2 atri – 2 ventricoli

- **Concordante**
- **Discordante**
- **Ambigua** **D-loop**
 L-loop

2 atri – 1 ventricolo principale

- **Doppia entrata in VS**
- **Doppia entrata in VD**
- **Assente connessione sx**
- **Assente connessione dx**

- **2 valvole** (1 può essere imperforata)

- **Valvola comune**

- **Valvola “overriding”** (anello)

- **Valvola “straddling”** (apparato tensore)

GIUNZIONE VENTRICOLO-ARTERIOSA :

Tipo di connessione

- **Concordante**
- **Discordante**
- **Doppia uscita** **da ventricolo dx**
 da ventricolo sx
- **Uscita singola** **truncale**
 aortica
 polmonare

Modo di connessione

- **2 valvole**
(1 può essere imperforata)
- **1 valvola comune**
(solo nel tipo: singola uscita
truncale)

♥ Situs

solitus
inversus
ambiguus

♥ Posizione del cuore

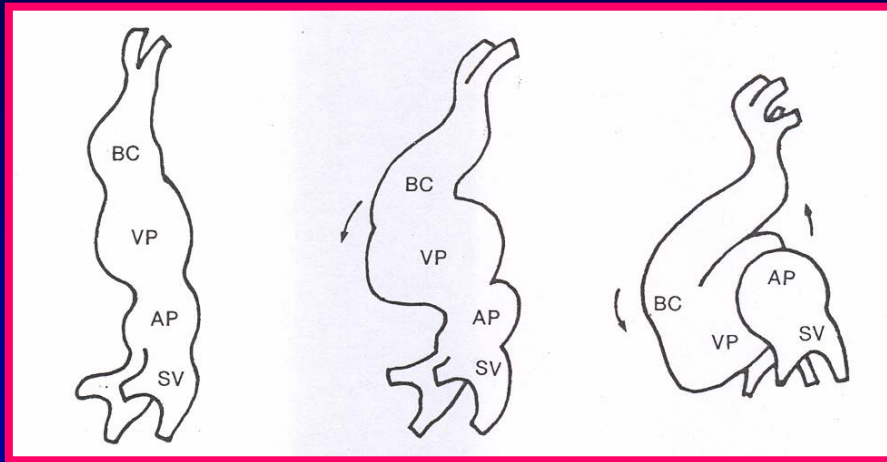
levocardia
destrocardia
mesocardia

♥ Connessioni

veno-atriali
atrio-ventricolari
ventricolo-arteriose

♥ Relazioni

dai ventricoli
dalle grandi arterie

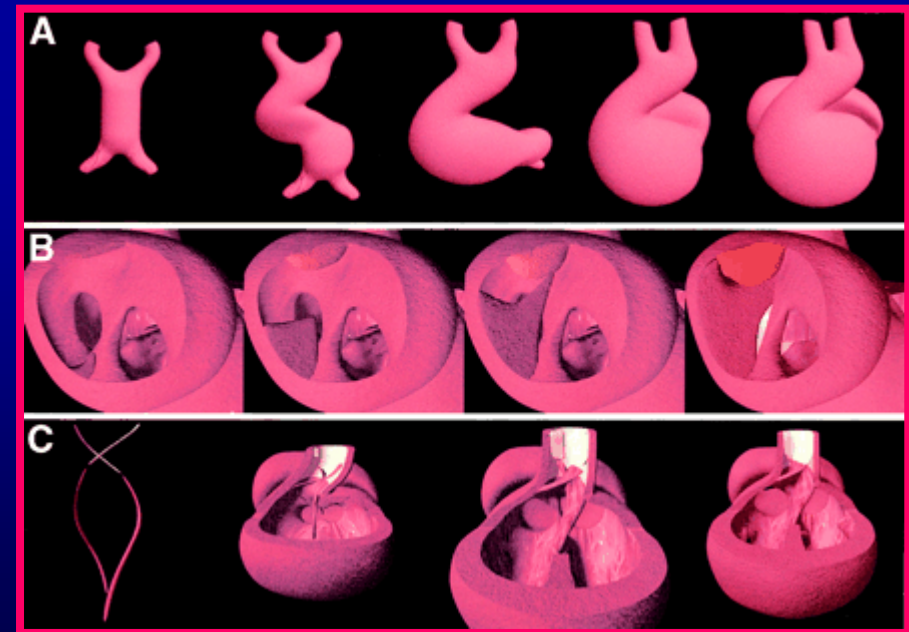


BC: bulbus cordis

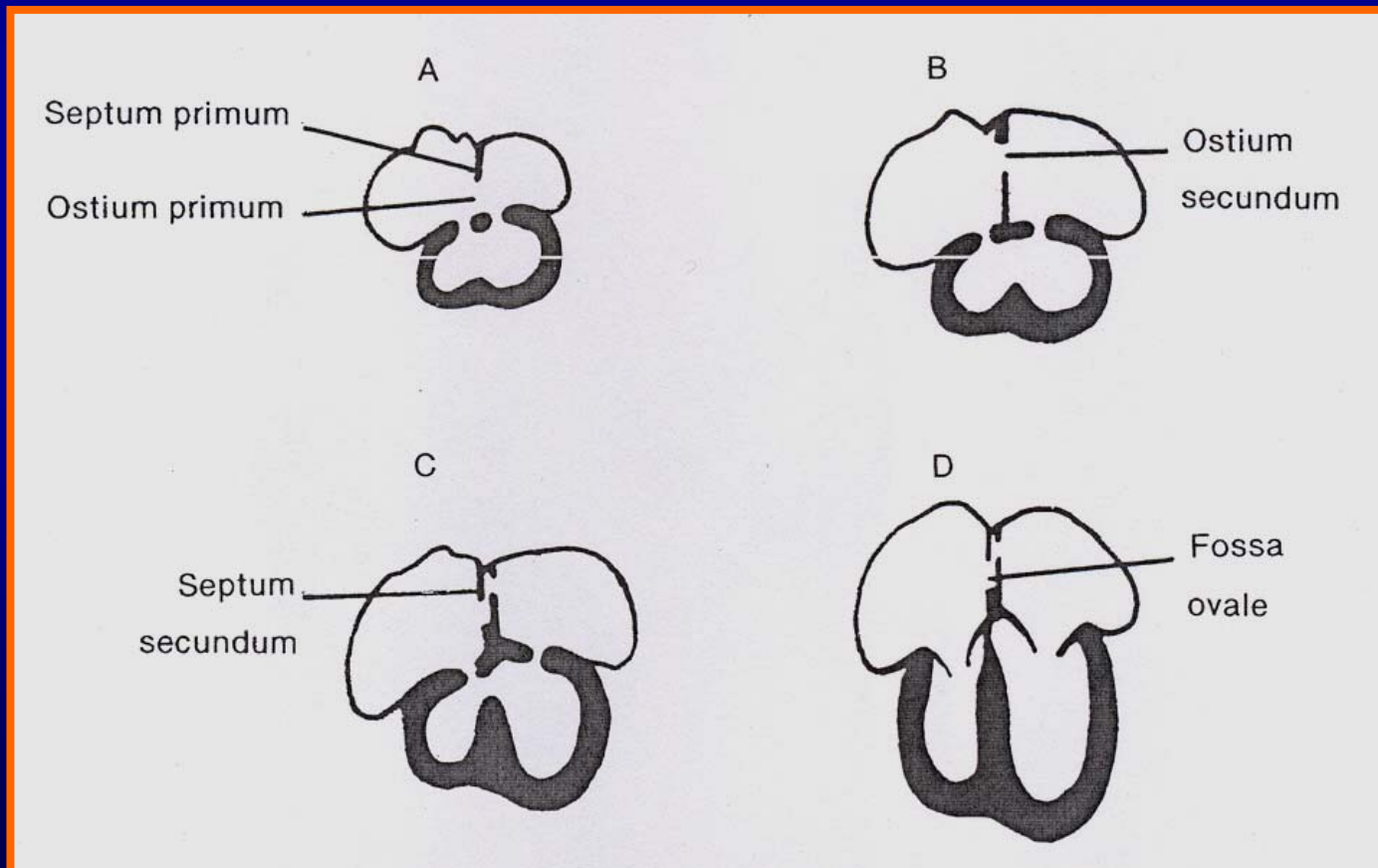
VP: ventricolo primitivo

AP: atrio primitivo

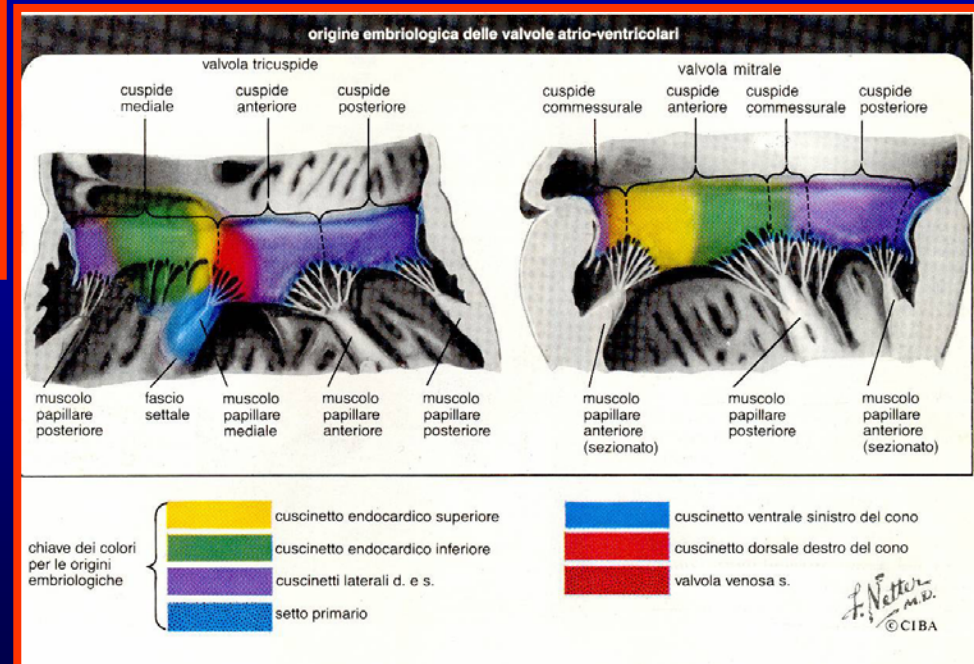
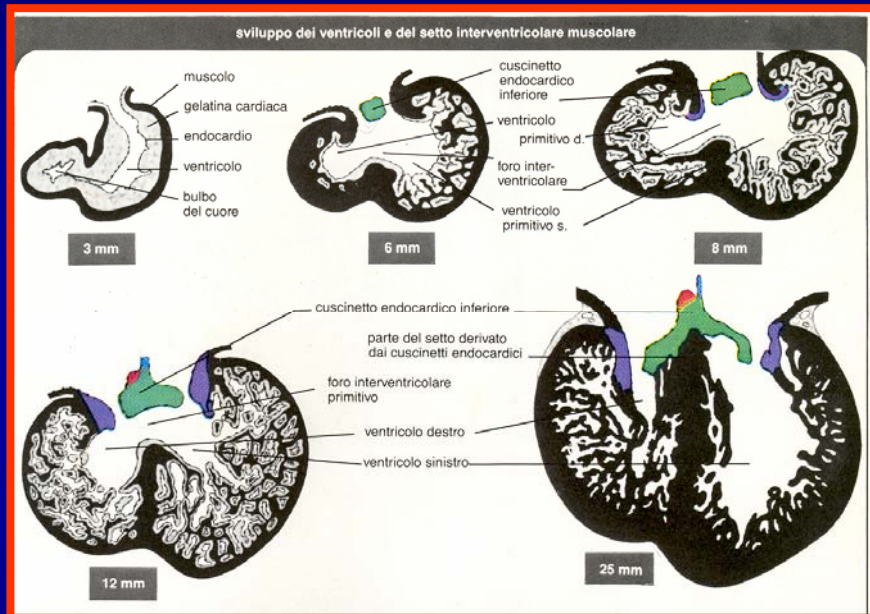
SV: seno venoso



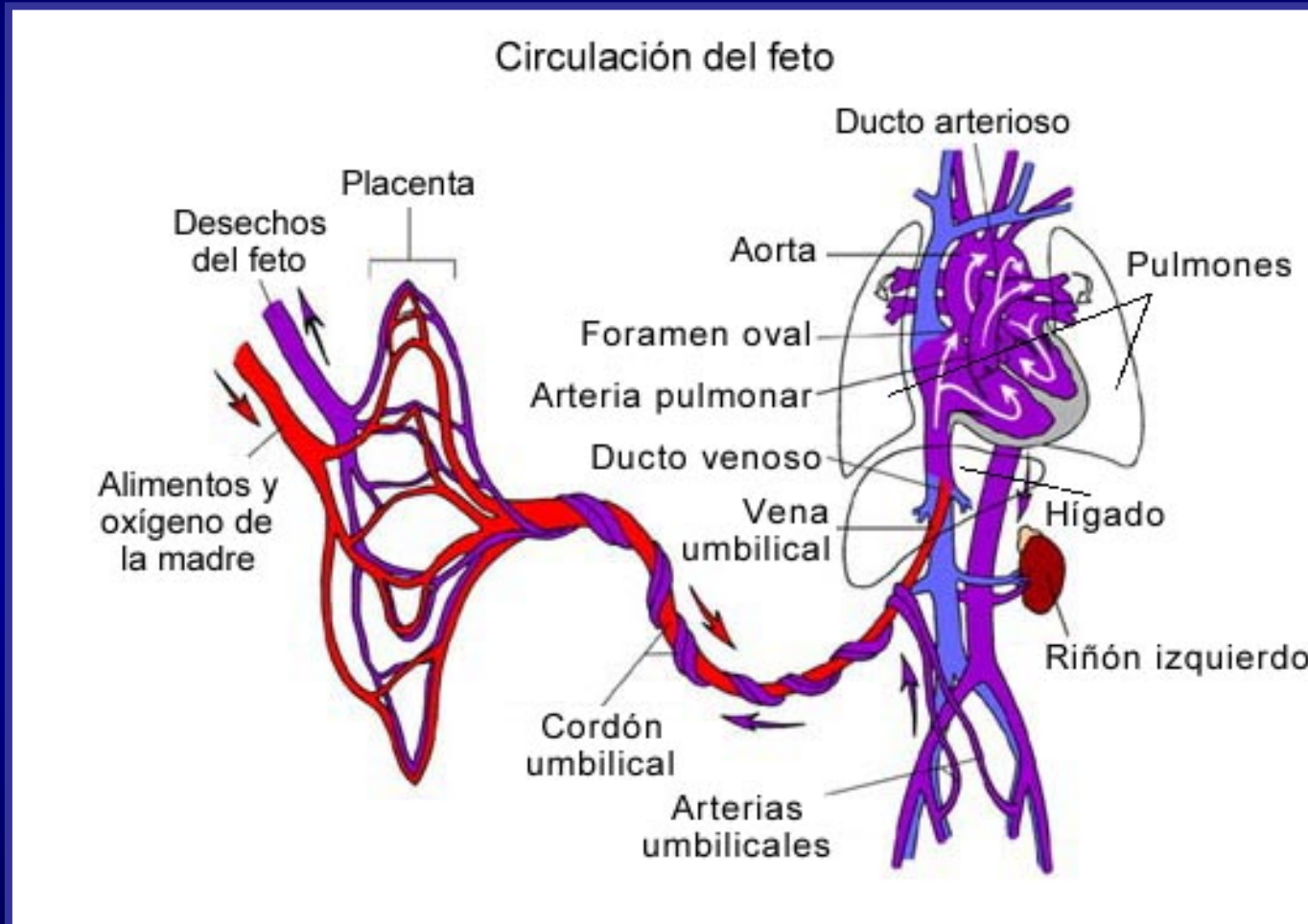
SVILUPPO DEL SETTO INTERATRIALE



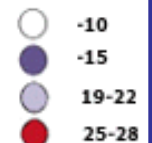
SVILUPPO DEL SETTO INTERVENTRICOLARE E DELLE VALVOLE ATRIO-VENTRICOLARI



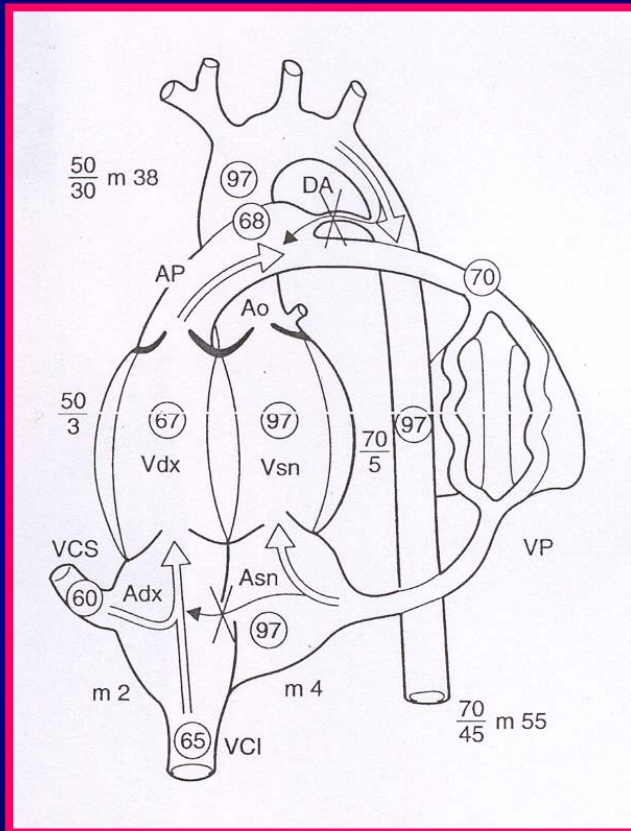
CIRCOLAZIONE FETALE



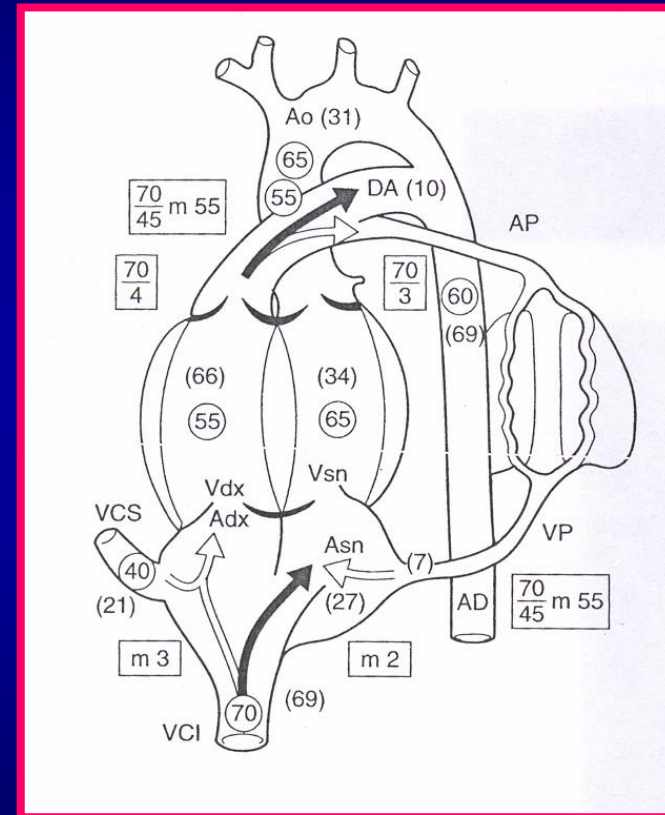
PO₂ mmHg



CIRCOLAZIONE FETALE E POST-NATALE



Limitata capacità di rendimento
Lavoro del miocardio fetale alla max capacità
PC fortemente dipendente dalla FC
(H₂O₂, mm, immaturità simpatico e metab mit)



Distacco placenta: ↑ RVS ↑ PA
Atti respiratori: ↓ P polmonari
↑ P AS → ↑ AS → chiusura FOP
↑ pO₂ polmonare (vasodilat polm ↑ bradichinine e PG)

IP PERSISTENTE NEL NEONATO

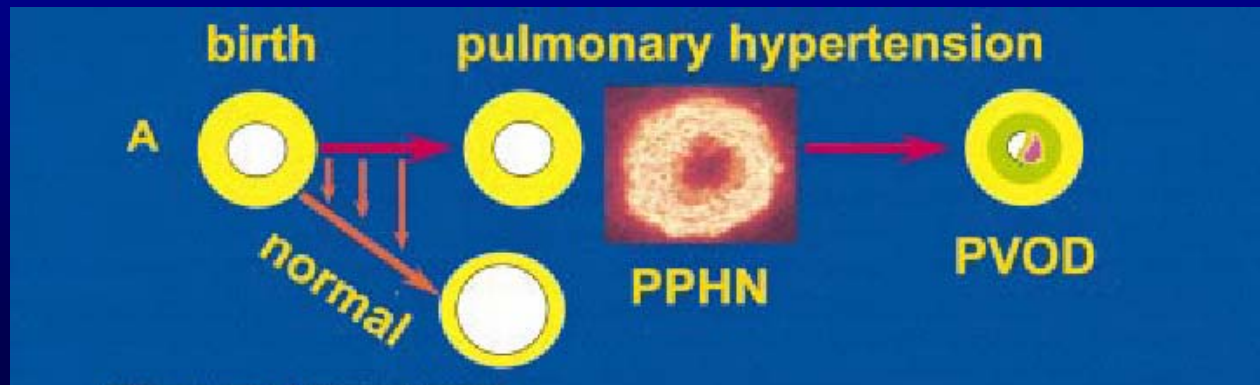
Alterato adattamento del circolo polmonare alla vita extrauterina

Incidenza 2-9%

Mortalità fino al 20%

Varianti:

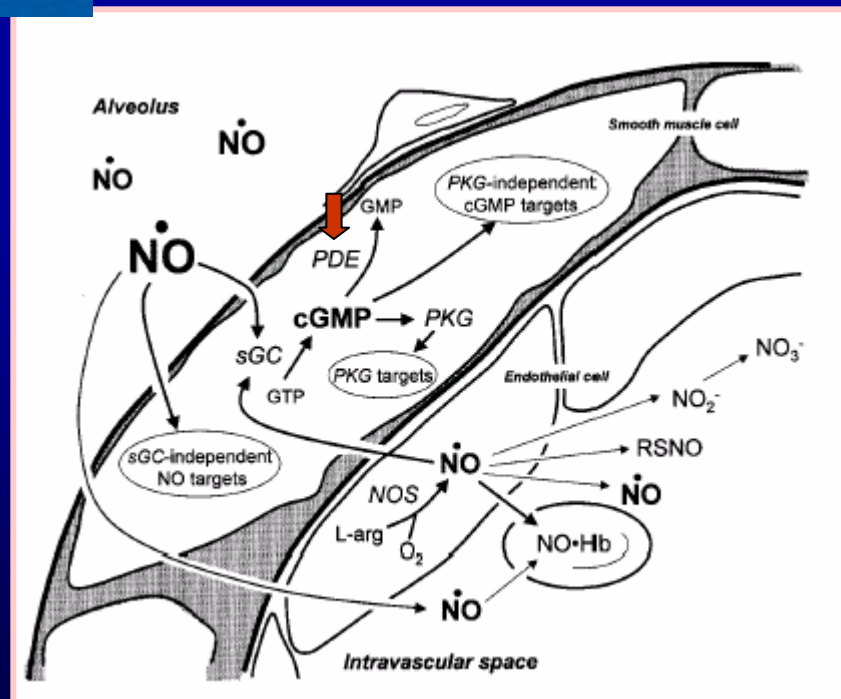
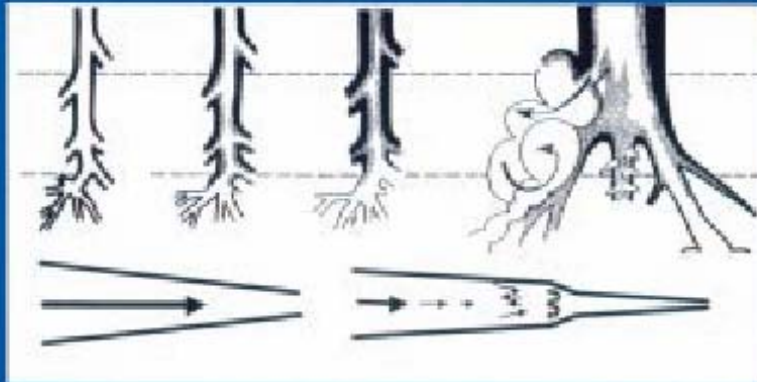
- ☹ incompleto sviluppo del letto vascolare polmonare
- ☹ anomalo sviluppo del letto vascolare polmonare **ipossia fetale cr.**
chiusura precoce di FOP , DA
- ☹ mal adattamento del letto vascolare **sofferenza fetale**
asfissia perinatale
s. aspirazione di meconio
m. infettiva polmonare



Evolution of pulmonary vascular disease and changing flow patterns



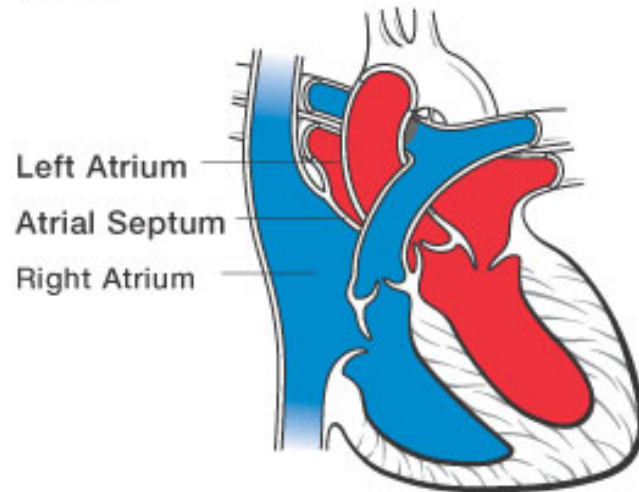
Normal



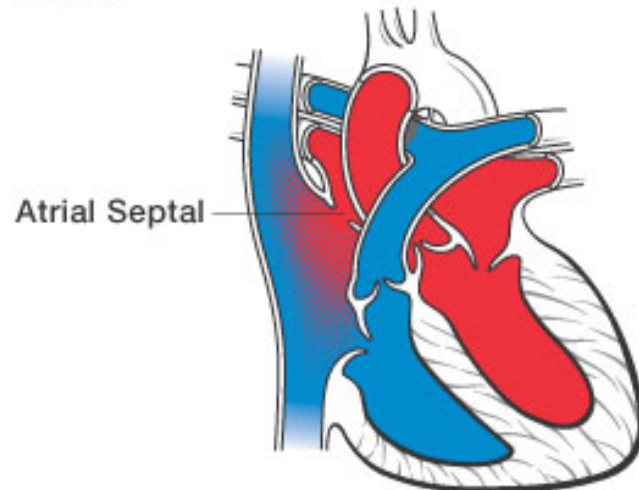
DIA

Atrial Septal Defect (ASD)

Normal



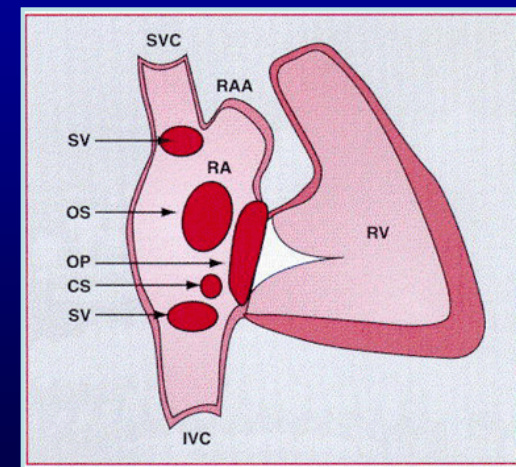
Defect



**Rappresenta il 10-15% delle CC
e 1/3 delle CC nell'adulto**

**75% tipo ostium secundum
15% tipo ostium primum
10% tipo seno venoso**

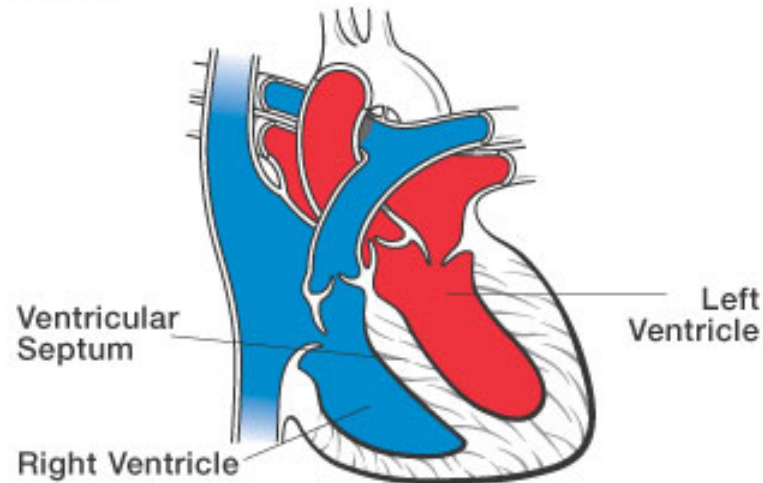
**$\leq 0,5$ cm piccolo shunt
 ≥ 2 cm ampio shunt**



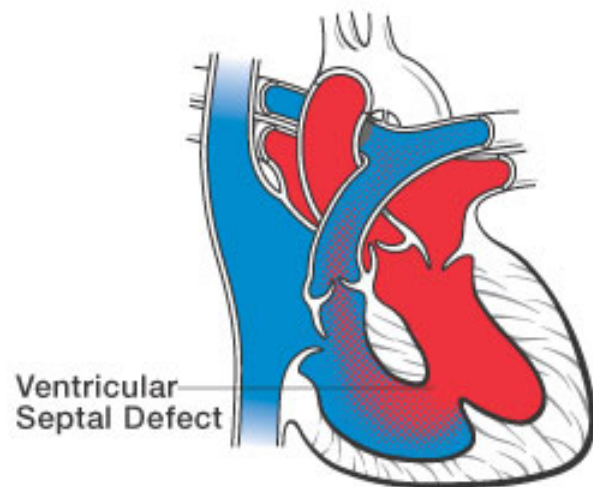
DIV

Ventricular Septal Defect (VSD)

Normal



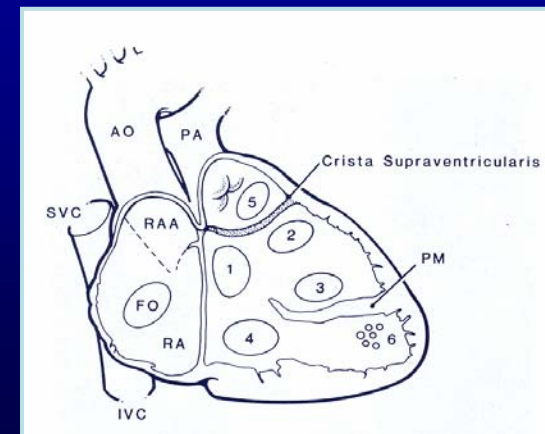
Defect



**E' il tipo più comune di CC con una
incidenza compresa fra 25 – 40 %**

**70% DIV membranoso
20% DIV setto muscolare
5% DIV sotto aortico
5% DIV giunzione Mi/Tri**

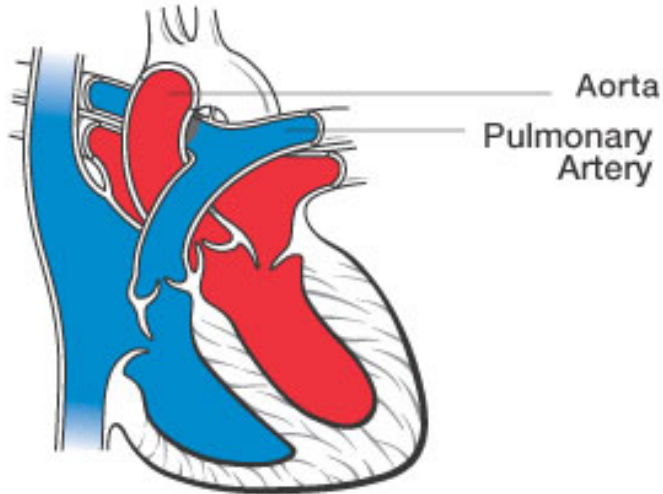
Nel 20-40% chiude spontaneamente



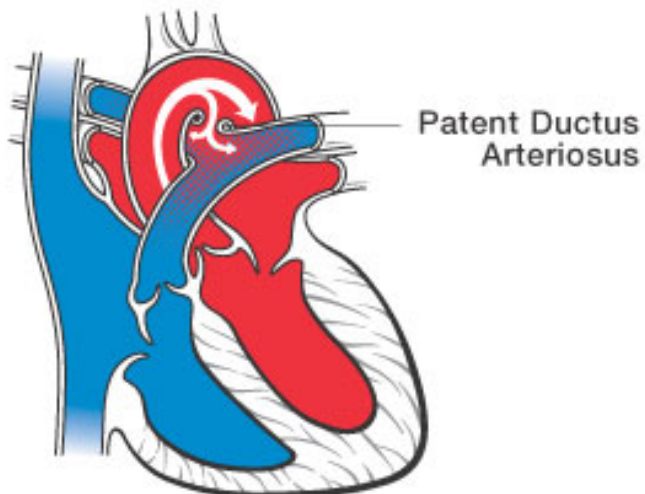
DA

Patent Ductus Arteriosus (PDA)

Normal



Defect



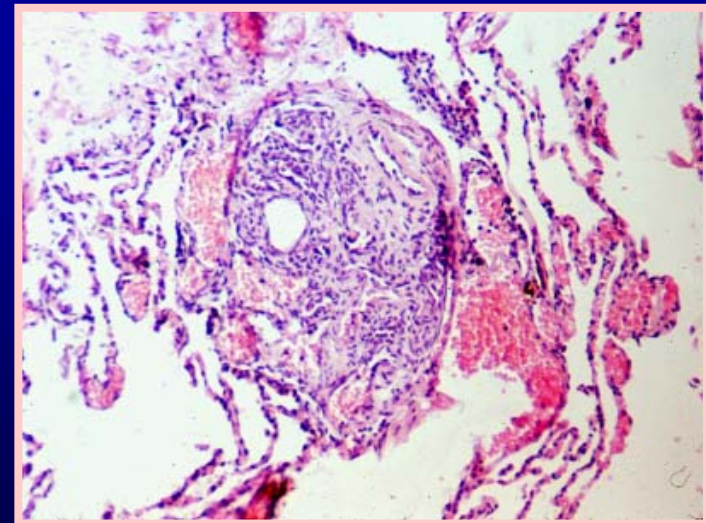
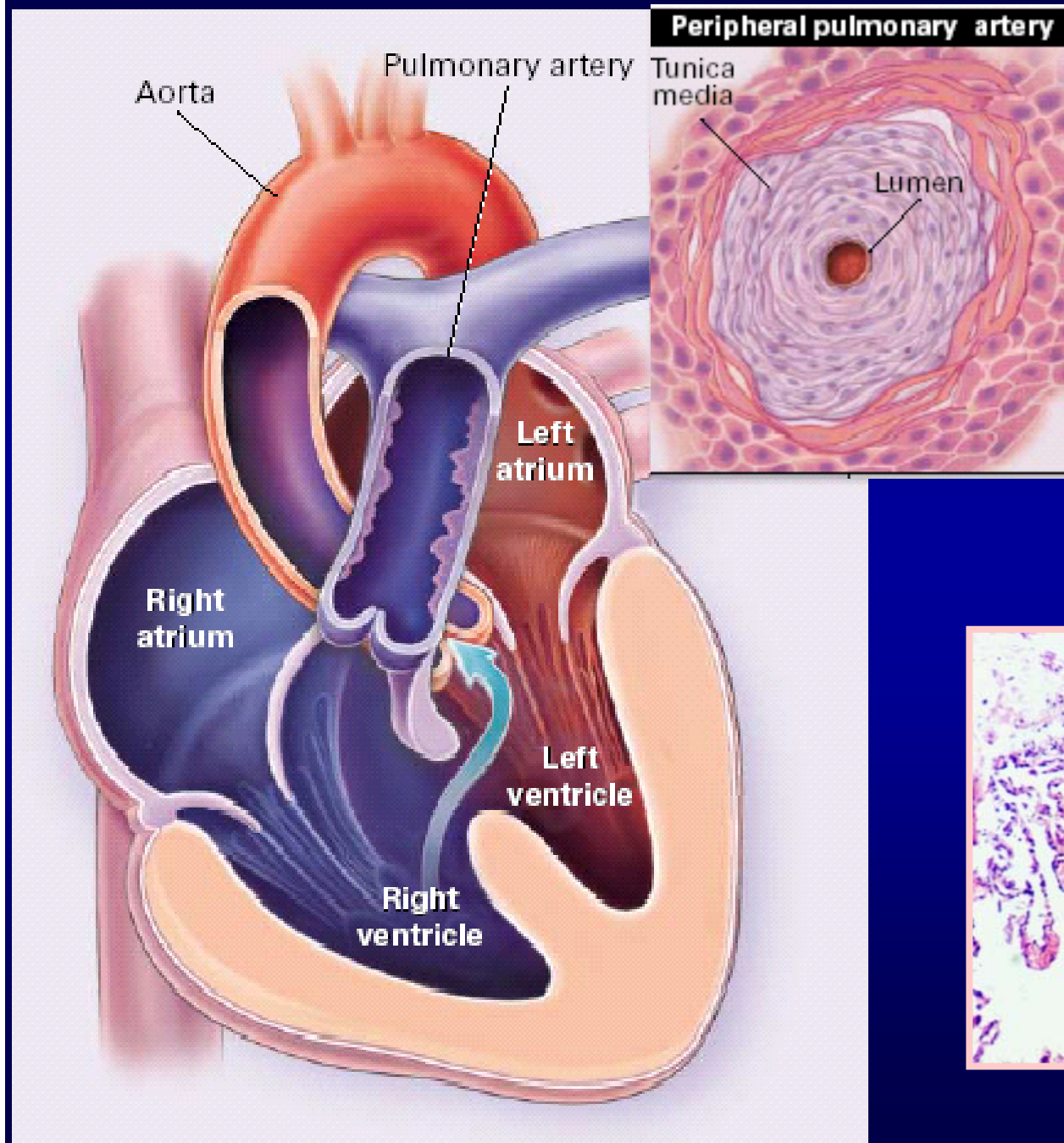
Incidenza del 10 %

Maggiore frequenza:
ipossiemia perinatale
nati prematuri
nati da madri con rosolia

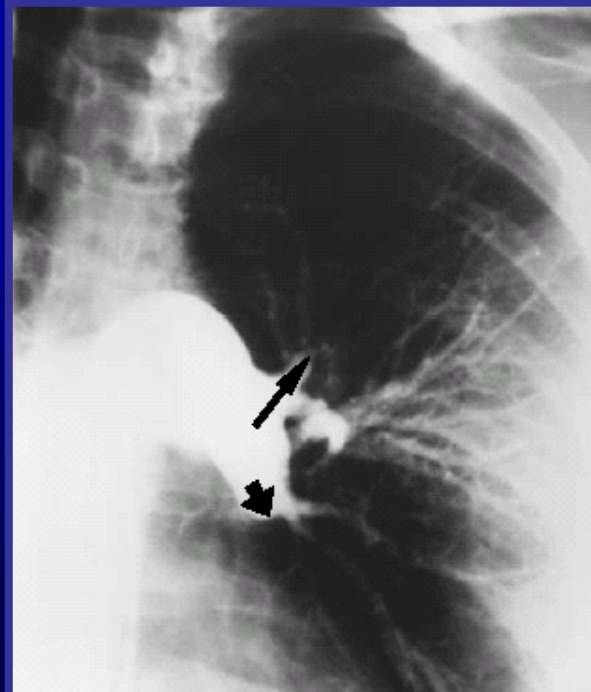
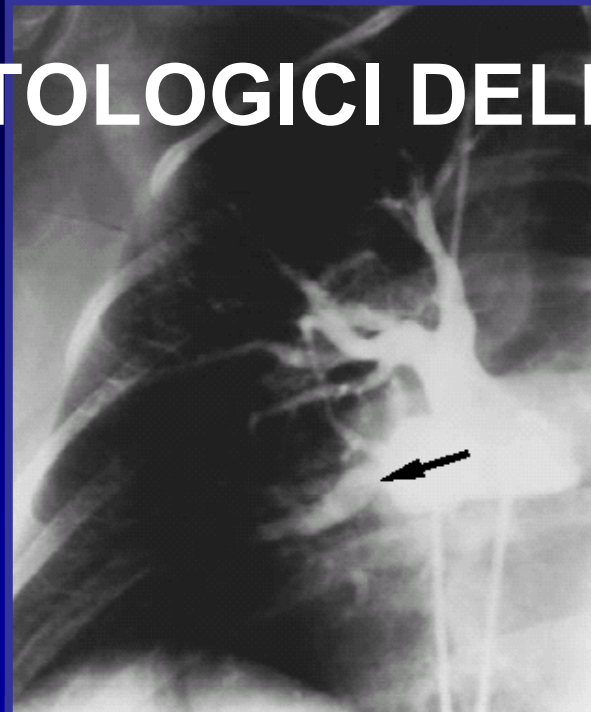
Piccole dimensioni
asintomatico
rischio endocarditi

Ampie dimensioni
Scompenso cardiaco
Ipertensione polmonare
Endoarteriti
Mortalità elevata (40-60 aa)

Sindrome di Eisenmenger



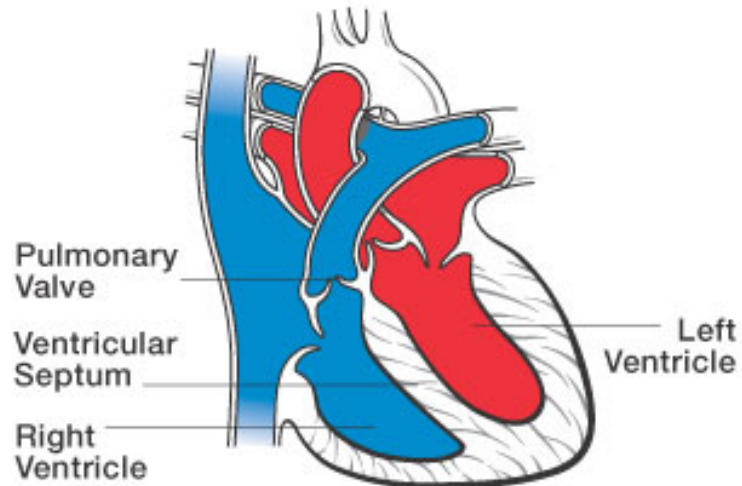
STADI ANATOMO-PATOLOGICI DELL'IP



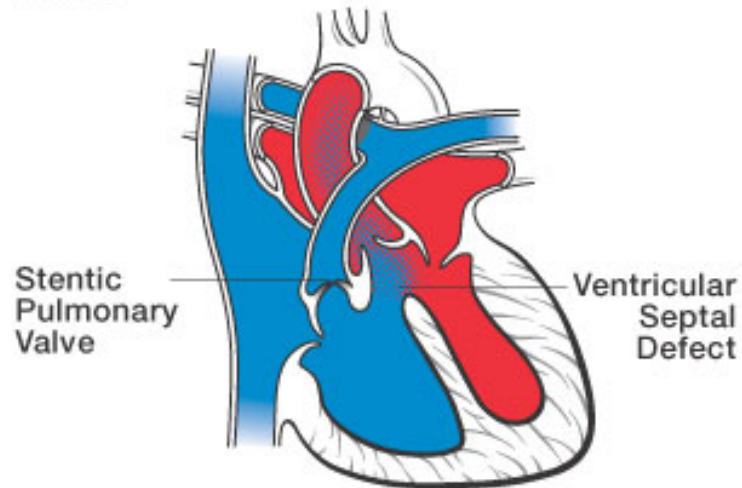
TF

Tetralogy of Fallot

Normal



Defect



Incidenza 5-15%

Anastomosi riparative

WATERSON: side-to-side

POTTS: side-to-side

BLALOCK-TAUSSIG: end-to-side

TGV

Incidenza 5%

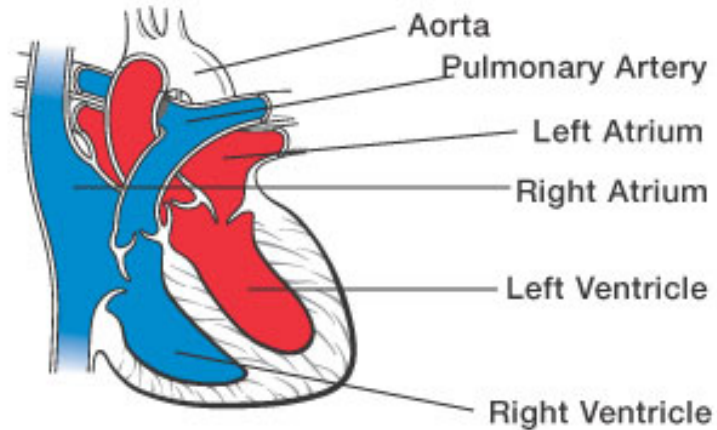
Interventi riparativi

Arterial switch

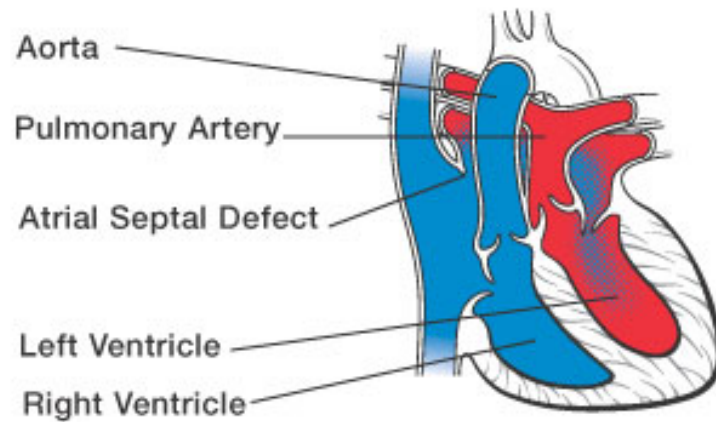
Atrial switch

Transposition of the Great Arteries

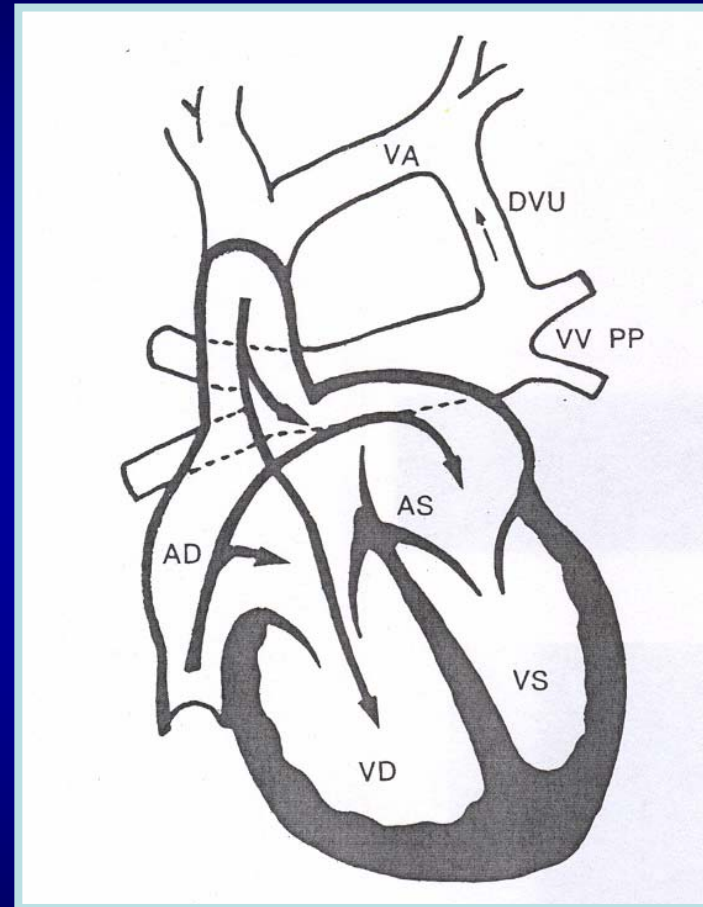
Normal



TGA



RITORNO VENOSO ANOMALO



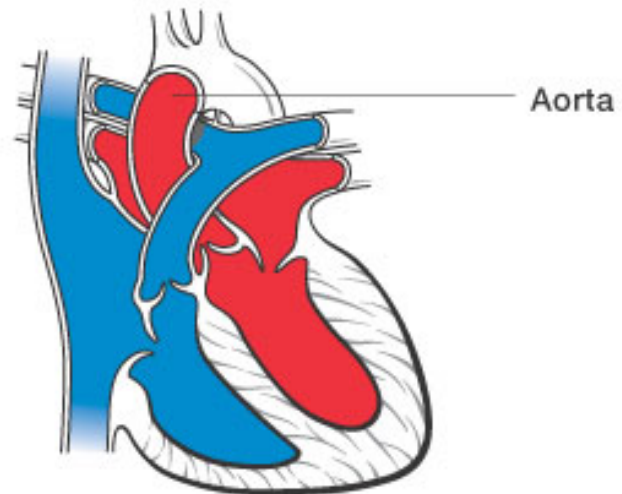
Totale / Parziale

Sopradiaframmatico / Sottodiaframmatico

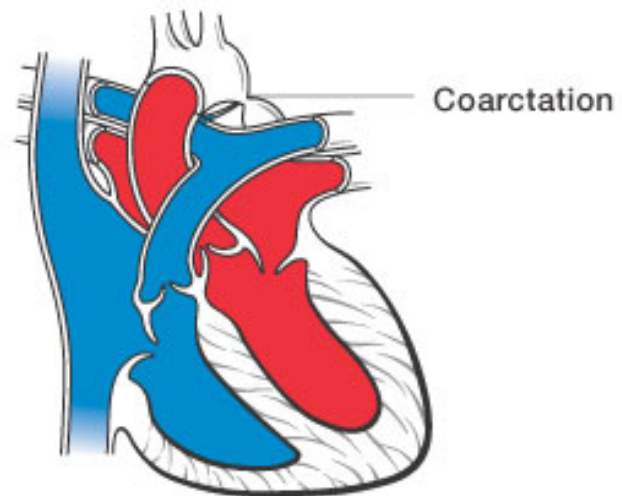
Dotto unico / Singole vv polmonari

Coarctation of the Aorta

Normal

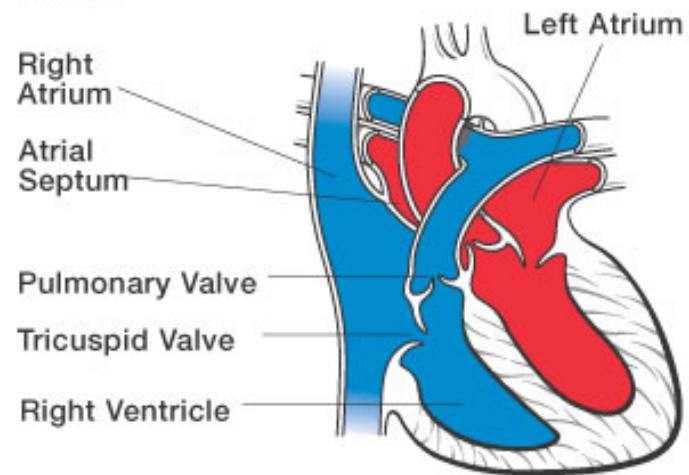


Defect

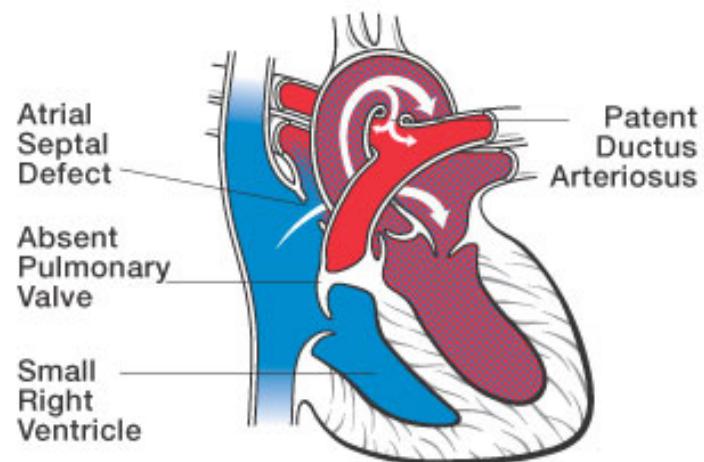


Pulmonary Atresia

Normal



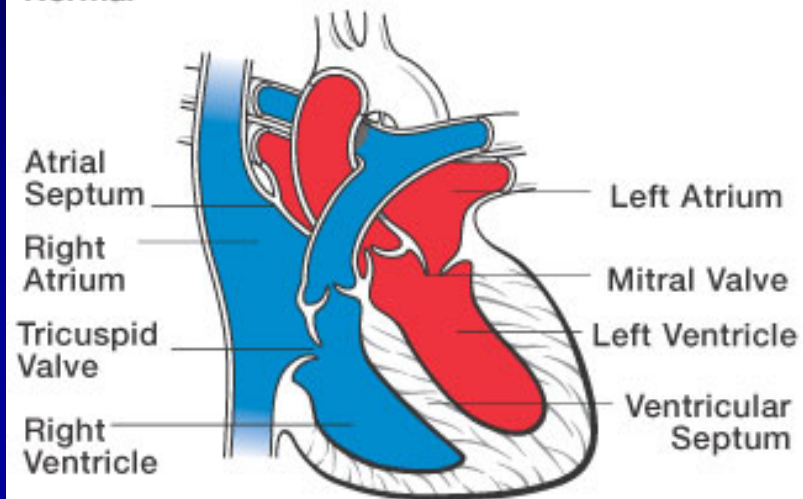
Defect



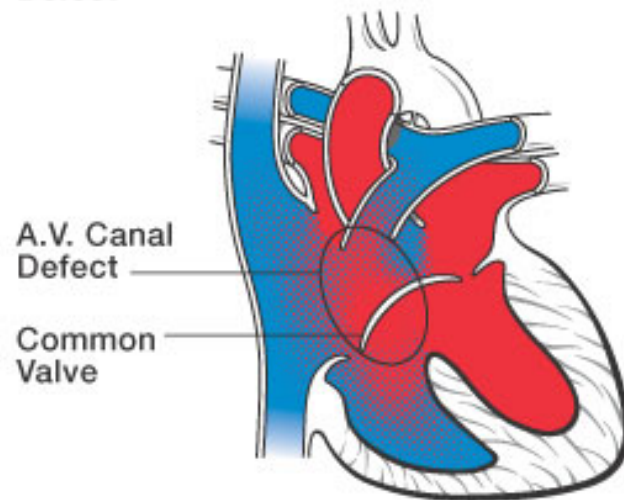
Atrioventricular Canal Defect

(Endocardial Cushion Defect, Atrioventricular Septal Defect)

Normal

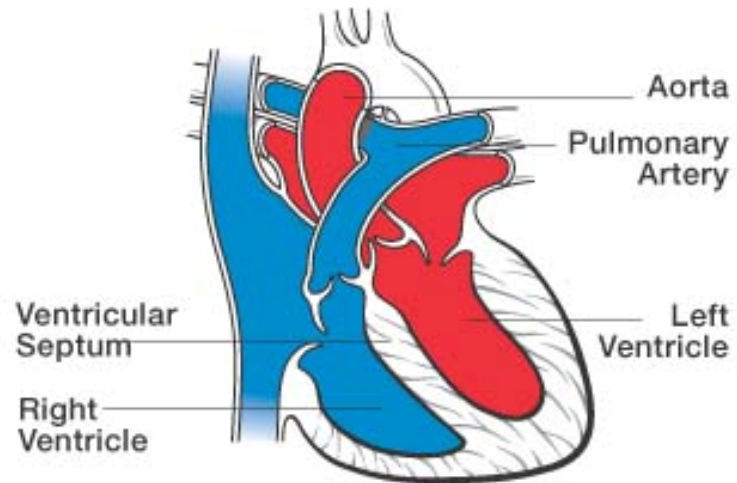


Defect

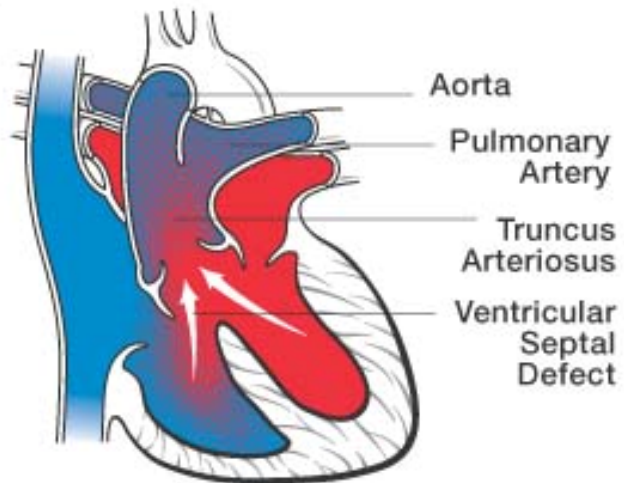


Truncus Arteriosus

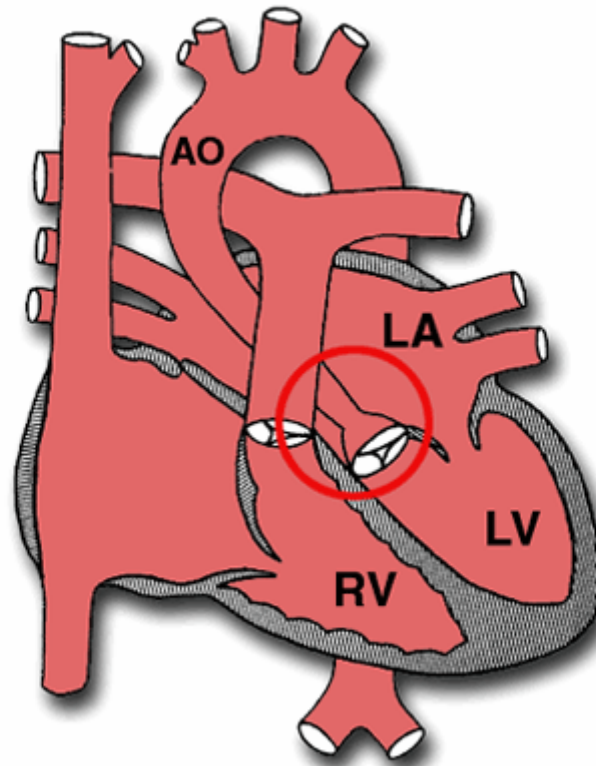
Normal



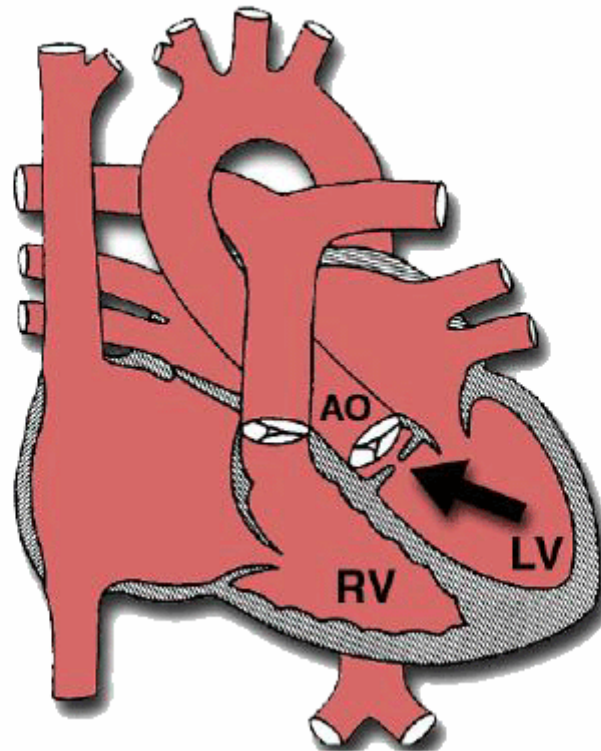
Defect



Supravalvular Aortic Stenosis



Membranous Subaortic Stenosis



Ebstein's Anomaly

