put together by Alex Yartsev: Sorry if i used your images or data and forgot to reference you. Tell me who you are.
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Congenital Heart Disease

History of Presenting Illness

Downs Syndrome will have been diagnosed ANTENATALLY via amniocentesis

- **The kid will present with some sort of genetic SYNDROME**
- **Poor weight gain, like FAILURE TO THRIVE**
- **Slow to feed** (duration for a complete feed - approximately 1 hour).
- **Sweaty and breathless 5-10 minutes from commencing feeding** resulting in "rest" pauses.

Findings on History

Will want to know OBSTETRIC HISTORY:
What did the mother do?
SMOKING / ALCOHOL / DRUGS?
Mother's AGE (most important risk factor)

RISK FACTORS

- Maternal Diabetes Mellitus
- Family history of congenital heart disease
- Maternal history: 5-10% CHD risk
- Sibling history: 2-3% CHD risk
- Indomethacin exposure
- Rubella exposure in first trimester (PDA)
  Residence at high altitude (PDA)

Differential Diagnoses

- **Neonatal Sepsis**
- **Pneumonia**
- **Inborn Errors of Metabolism**
- **Structural heart disease**
- **Myocarditis**
- **Dilated Cardiomyopathy**
- **Supraventricular Tachycardia**
- **Hypoglycemia**
- **Neurologic and Hematologic causes** (much less common)

Findings on Examination

A. Skin Color (cyanosis)

B. Signs of **Respiratory distress**
   1. Grunting
   2. Tachypnea

C. **Difficult feeding** precedes Congestive Heart Failure
   1. Term infant parameters
      a. Prolonged feeding longer than 40 minutes
      b. Less than 2 ounces per feeding
   2. Distress signs provoked by feeding
      a. Tachypnea
      b. Diaphoresis
      c. **Subcostal retraction**

D. Precordial examination
   1. S3 gallup rhythm
   2. Cardiac Murmur
      a. See Pediatric Murmur evaluation
      b. Often the least important of exam

E. Femoral and Brachial Pulse
   1. Compare both brachial pulses for symmetry
   2. Compare one brachial and one femoral pulse
   3. Femoral Pulses diminish with PDA closure
   4. Brachial pulses absent in left sided obstruction

F. Hepatomegaly

G. Concurrent Congenital defects

H. Oxygen Saturation in upper and lower extremities
   1. Pulmonary cause related cyanosis
      a. Supplemental Oxygen 100% increase O2 Sat >95%
   2. Cyanotic Congenital Heart Disease causes
      a. Supplemental Oxygen 100% increases O2 Sat <85%

I. Blood Pressure in all 4 extremities

J. Failure to Thrive
   1. Height and Head Circumference may be normal
   2. Weight falls behind

~Down Syndrome: Cardinal features of the Newborn~

- *hypotonia*
- *excessive skin folds at the back of the neck*
- *maxillary (malar) underdevelopment (hypoplasia)*
- *in curving of the little finger (clinodactyly)*
- *hypoplasia of the middle phalanx of the 5th finger - a short middle segment or a single interphalangeal crease.*
- *wide gap between the first or second toes*

*Bilateral **Single transverse palmar crease** "simian crease"*

Features seen in the Down child:

- Delayed psychomotor development
- Intellectual disability
- Prominence of the tongue (due to a small mouth)
- Persistent epicanthic folds
- Flattening of the back of the head (brachycephaly)
- Short stature
- Brushfield spots (speckling around the rim of the iris) except in subjects with brown irides
- Joint hypermobility
- Atlanto-occipital instability

Tests and Investigations

Arterial Blood Gases

- ***To figure out if it is CYANOTIC or ACYANOTIC***

Chest X-ray

- Cardiomegaly (of all or any of the chambers)
- Increased pulmonary vascular markings

Chest MRI

- Exact nature of the defect may be seen
- The surgical approach is made much clearer with MRI

ECG:

- Looking for Rt or Lt Axis Deviation

Doppler Echocardiogram

- Enables the imaging of flow directions in the defect

ABNORMALITIES ASSOCIATED WITH DOWN SYNDROME:

Congenital heart disease in 40%
GIT malformation: 10%
Vision Disorder in 46%
Conductive hearing loss
Hypothyroidism
Atlanto-axial instability 15%
(spinal cord compression in 1%)
undescended testes 50%
obesity 70% male, 96% female
poor oral health
coeliac disease 11%
psychiatric disorders 22%
EARLY ALZHEIMERS → 10% by 40y.o.

AMNIOCENTESIS:

A dangerous and invasive procedure which samples the **amniotic fluid (20mls)**
! Offered to the over-35s
→ ultrasound-guided trans-abdominal needle puncture
→ chromosome count reveals if there is any cause for concern.
Amniocentesis causes spontaneous miscarriage in 0.5% of instances

ULTRASONOGRAPHY CAN DETECT THE DOWNS "NUCHAL FOLD" IN UTERO !→ non-invasive Dx

Management

Immediately: Oxygen via nasal cannula or face mask
Palliatively: high-yield feeds to reduce necessary feed duration and thus facilitate return to normal head circumference and weight parameters
Surgical: by closure of the defect.

Prognosis

Life expectancy for a Down Syndrome sufferer with mild/moderate disability = 55 yrs

- ***75% of concepti with trisomy 21 die in utero***
- ***85% of infants survive to 1 year***
- ***50% can be expected to live longer than 50 years.***

The presence of congenital heart disease is the most significant factor that determines survival.

Epidemiology

8 in 1000 live births have some sort of congenital heart defect.
Approx. 1 in 800 live births has Down syndrome
The cause of Down syndrome is full trisomy 21 in 94% of cases

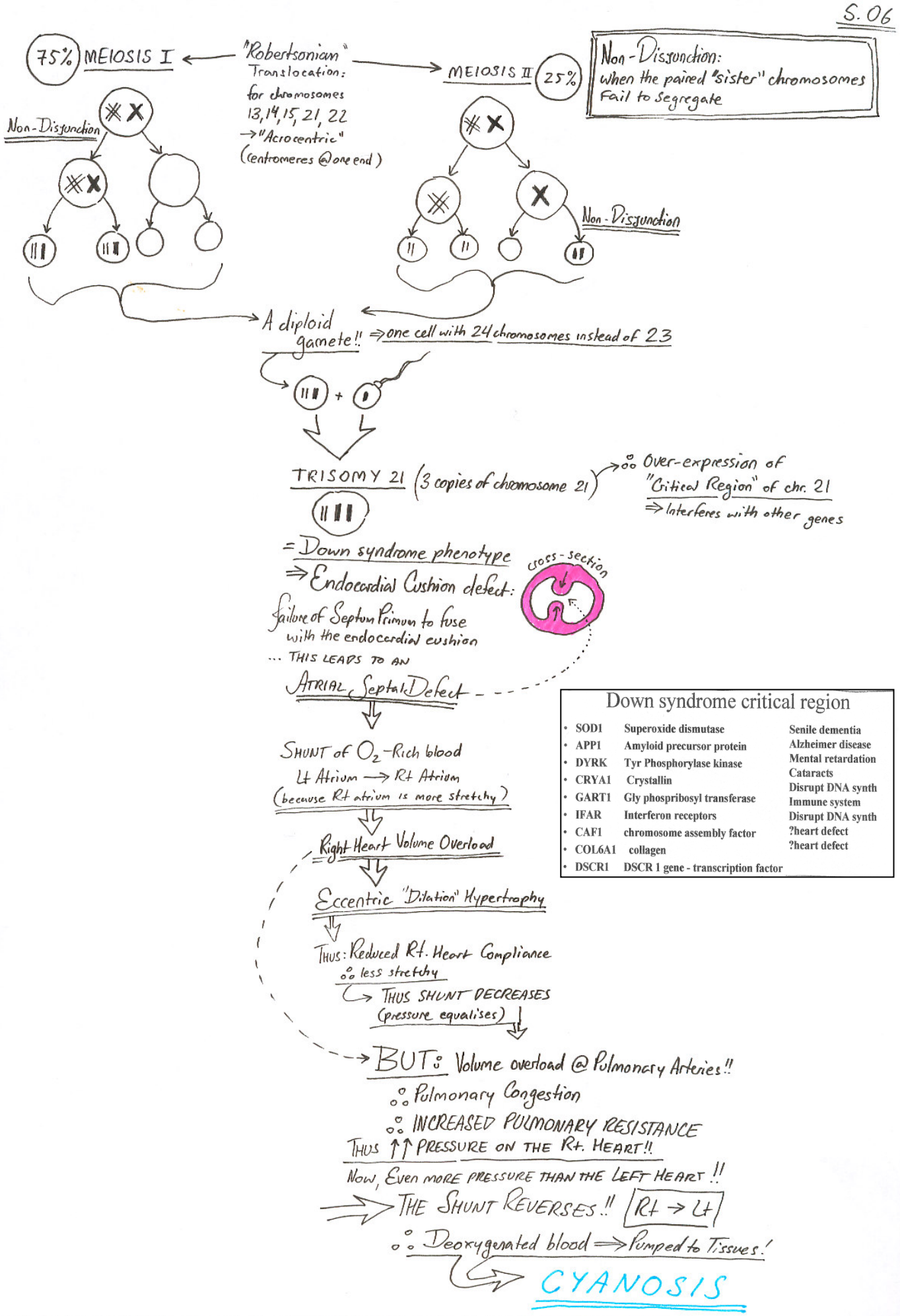
Sex: 15% more common in males
Race: there is no racial predilection

Advanced maternal age remains the only well-documented risk factor for maternal meiotic nondisjunction.

- *With a maternal age of **35 years, risk is 1 in 385.***
- *With a maternal age of **40 years, risk is 1 in 106.***
- *With a maternal age of **45 years, risk is 1 in 30.***

PATHOGENESIS: Downs Syndrome associated Heart Defect

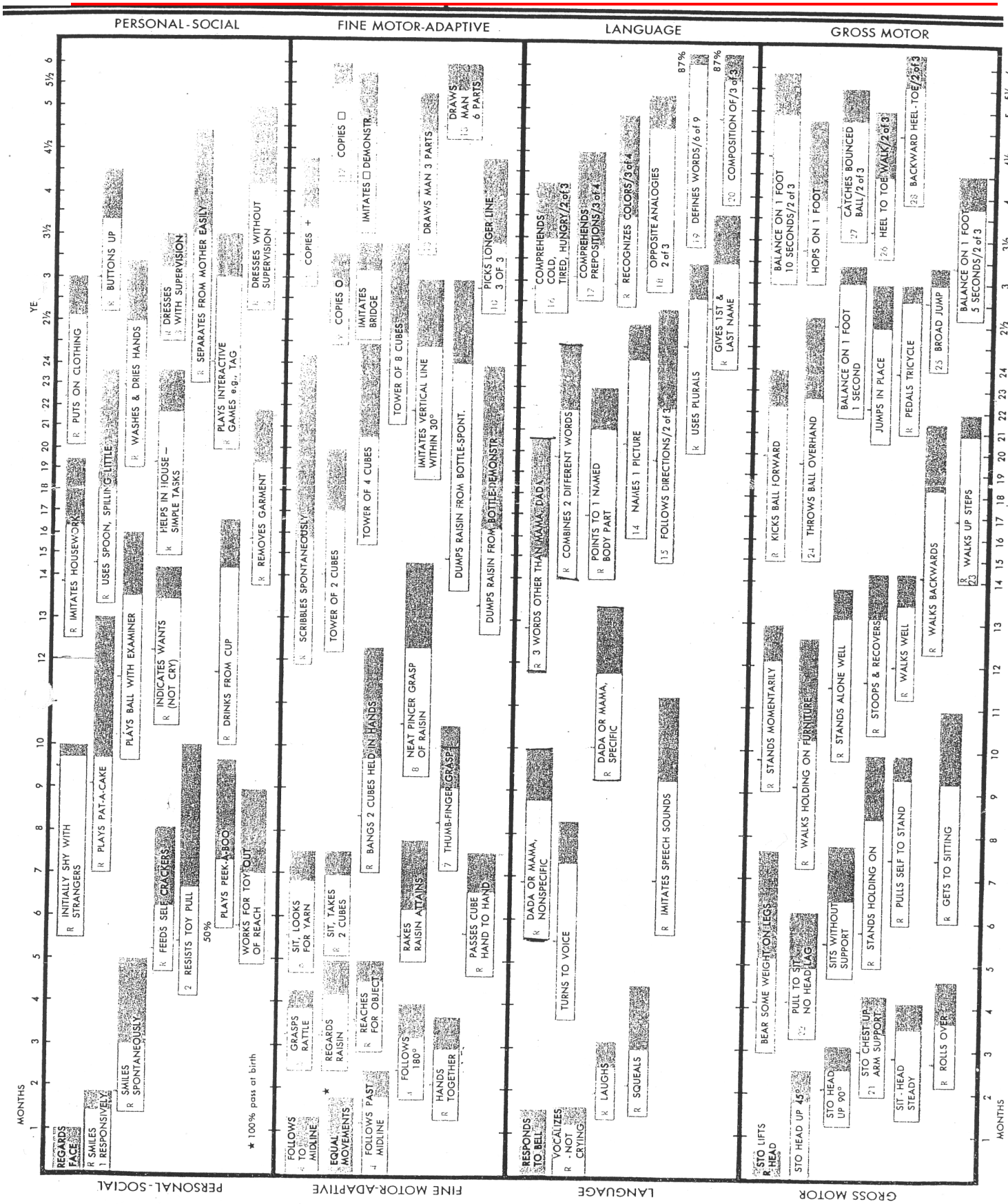
S.06



Down syndrome critical region

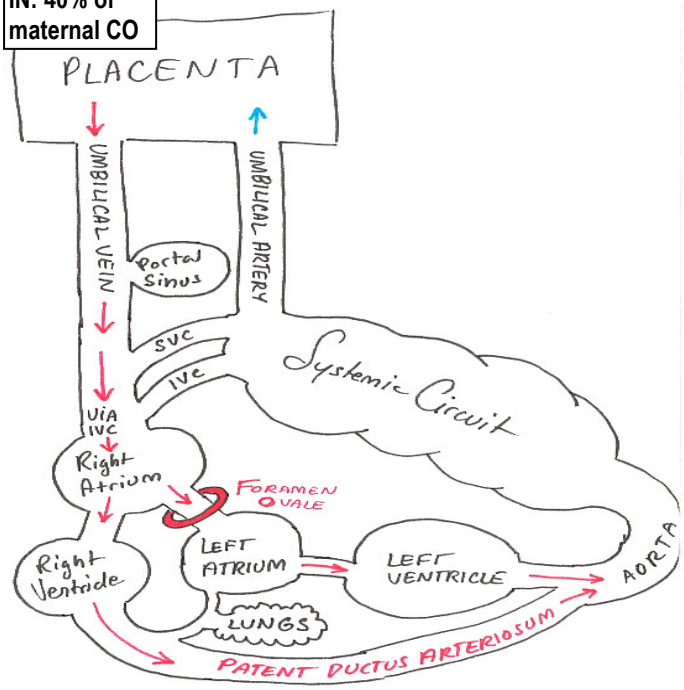
• SOD1	Superoxide dismutase	Senile dementia
• APP1	Amyloid precursor protein	Alzheimer disease
• DYRK	Tyr Phosphorylase kinase	Mental retardation
• CRYA1	Crystallin	Cataracts
• GART1	Gly phospribosyl transferase	Disrupt DNA synth
• IFAR	Interferon receptors	Immune system
• CAF1	chromosome assembly factor	Disrupt DNA synth
• COL6A1	collagen	?heart defect
• DSCR1	DSCR 1 gene - transcription factor	?heart defect

DEVELOPMENTAL MILESTONES



FOETAL CIRCULATION

IN: 40% of maternal CO



BEFORE BIRTH:

Circulation bypasses liver and lungs

Placental resistance is low → 40% of the total maternal cardiac output passes through it; fetal hemoglobin harvests the O2

PLACENTA → Umbilical vein → IVC → RA → RV, LA , LV → systemic circuit → umbilical artery → PLACENTA

Why the foetal lungs are shut down:

Alveoli filled with fluid: much tougher + denser than air!

THUS – compress pulmonary vasculature,

THUS the resistance is too great for blood to flow down the lung circuit, especially seeing as there is a path of less resistance (patent ductus arteriosum)

CHANGES DUE TO BIRTH:

Baby becomes hypoxic → medullary centre stimulates respiration: Most of the lung fluid expelled with first few high-pressure breaths; THUS →**THE LUNGS ARE NOW THE PATH OF LEAST RESISTANCE**

And the blood flows down into the newly opened pulmonary circuit

Patent Ductus closes within 10-15 hrs (permanently within 2-3 weeks)

Foramen Ovale closes immediately (due to sudden filling of left atrium with lung blood , plus decreased Rt atrium pressure because the placental vein no longer supplies it → THUS pressure imbalance forces the **foramen ovale** shut)

Ductus Venosus closes after 3-7 days

Probably due to reduced flow pressure from the umbilical vein (nothing to keep it inflated open)

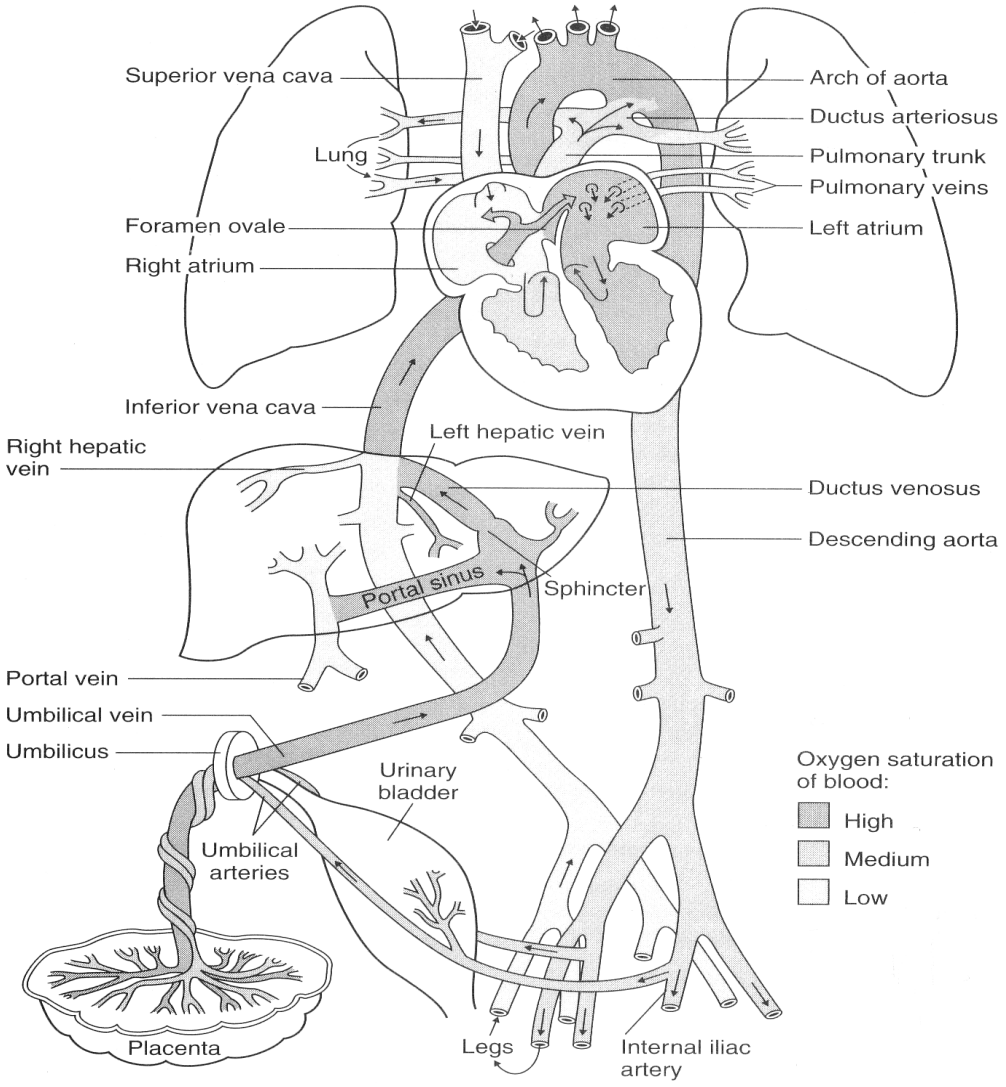


Figure 16.10. **The fetal circulation, as described in the text.** Arrows indicate the direction of blood flow. Three shunts (ductus venosus, foramen ovale, and ductus arteriosus) allow the majority of the blood to bypass the ungs and liver during fetal life, but cease to function shortly after birth. (Modified from Moore KL, Persaud TVN. The Developing Human. Philadelphia: WB Saunders, 1993:344.)

Postnatal Course with Abnormal Transition of Blood Flow:

Diagnosis relies on timing of presentation:

IMMEDIATELY:

Almost always due to parenchymal lung disease

WITHIN 4 HOURS:

Inadequate pulmonary blood flow due to Rt Heart Hypoplasia

AFTER 4 HOURS:

Likely to be duct-dependant lesion

Normal transition complicated by premature birth:

Means that it's a Patent Ductus Arteriosum problem

Basic Sciences: ASSORTED HEART DEFECTS in order of prevalence

ACYANOTIC: LEFT →RIGHT SHUNTS

VENTRICULAR SEPTAL DEFECT 1.5 in 1000

Blood from LV shunted to RV, thus volume overload in the RV, pulmonary circuit, LA, and thus LV… This reuslts in hypertrophy of all the above chambers plus pulmonary congestion → eventually leading to symptoms of heart failure !! If pulmonary resistance increases for whatever reason, you will get Eisenmenger syndrome: Rt→Lt shunting; CYANOSIS

Harsh systolic murmur @ lt sternal edge (small defect = louder)

ATRIAL SEPTAL DEFECT: 1 in 1500

Blood from LA shunted to RA (thus no cyanosis)
= this is an uncomplicated ASD ;
→ **RA enlargement** due to overload results in **hypertrophy**, loss of compliance and subsequently increased pressure, plus **pulmonary congestion** possibly as part of the Eisenmenger syndrome → the shunt reverses and becomes Rt→Lt, thus CYANOSIS RESULTS (that's what happens to the 5.06 baby)

RV Heave, split S2, pulmonary valve murmur

PATENT DUCTUS ARTERIOSUS 1 in 2500

Blood flowing from the Aorta via the patent ductus into the pulmonary circulation (which its meant to bypass)→
Thus → pulmonary congestion, LA + LV hypertrophy
→ LEFT-SIDED HEART FAILURE
Once again, if pulmonary resistance increases, you will get EISENMENGER's SYNDROME with shunting from pulmonary artery to the aorta, and thus CYANOSIS

Continuous machine-like murmur @ Lt subclavicular region

CONGENITAL AORTIC STENOSIS

Forever narrow aortic valve = much resistance to cardiac outflow into the aorta, thus concentric hypertrophy of LV and a powerful jet of blood ejected into the aorta may dilate the aorta past the narrowing; thus → aneurysm.

Harsh systolic crescendo-decrescendo murmur, radiates to the neck

PULMONIC STENOSIS

Obstruction to RV ejection → RV hypertrophy

RV heave, elevated JVP, crescendo-decrescendo murmur @ P area

COARCTATION OF AORTA 1 in 6000

= discrete narrowing of the aortic lumen; thus LV faces a MASSIVE PRESSURE AFTERLOAD → if the coarctation is after the brachiocephalic branching, the circulation is preserved in the upper body; but the lower body is cyanotic. To compensate, the LV becomes hypertrophied.

HEART FAILURE ENSUES EARLY AFTER BIRTH

Collateral circulation is established via the internal thoracic costal arteries, thus → characteristic notches on the inner inferior margin of the ribs.

CYANOTIC: RIGHT →LEFT SHUNTS

TETRALOGY OF FALLOT:

Results from *ONE SINGLE ABNORMALITY*

→ an abnormal anterior and superior displacement of the **ventricular outflow tract area of the septum**

THUS → 4 characteristic abnormalities

1. **Ventricular Septal Defect**
2. **Aorta receiving blood from both ventricles**
3. **Pulmonic Stenosis below the valve**
4. **RV hypertrophy (due to the pulmo0nic stenosis)**

THUS: Increased rsistance of the pulmonary circuit
→ shunting of RV blood via the VSD into the LV,
→**CYANOSIS results**
="boot-shaped" heart on Chest Xray

whenever you get systemic vasodilation, an increased Rt→Lt shunt result s→ children alleviate this symptom by squatting and thus "kinking" femoral arteries and thus reversing the shunt

phys exam:
chronic hypoxia, thus clubbing, plus RV heave, plus pulmonary murmur

TRANSPOSITION OF GREAT ARTERIES (7% of congenital heart defects)

STUPIDLY, the AORTA ORIGINATES IN THE RIGHT VENTRICLE, AND THE PULMONARY ARTERY – IN THE LEFT VENTRICLE.
→ this SEPARATES the pulmonary and systemic circuits!
Now, they are in parallel rather than in series!
THUS oxygeneated blood never makes it to the tissues, and the systemic blod never gets oxygenated! → **LETHAL CONDITION!!**
...UNLESS the foramen ovale and the patent ductus remain open, in which case you may live awhile.
→ rapidly progressive cyanosis makes this condition obvious
RV heave (RV faces systemic pressure) and loud S2 sound

PATENT FORAMEN OVALE

Is **normally kept closed** by the comparatively higher L.A pressure. **BUT !! when the RA pressure rises** eg. in pulmonary hypertension or Tetralogy of Fallot-
a **Rt→ Lt shunt is formed!!**
This causes inexplicable cyanosis and sometimes
Paradoxical Emboli

EISENMENGER SYNDROME

= severe pulmonary vascular obstruction

results from a chronic left-to-right shunt (hypertension in the lungs causes the thickening of the vessel walls thereoin, thus increased resistance to blood flow→ thus higher pressure in the Rt ventricle → this causes the reversal of an originally Lt→Rt shunt
THUS → CYANOSIS
Typically, huge pulmonary arteries are seen on chest X-rays

What the hell is CYANOSIS anyway? 5.06

→ **blue colouration of skin due to extra DEOXYHEMOGLOBIN**
→ from either **less oxygen** or more deoxygenation (due to **slowed blood flow**)

LESS OXYGEN → central cyanosis
SLOW CIRCULATION → peripheral cyanosis

Detectable @ O_2 saturation of 67%
(DANGEROUS! This is below the normal venous O_2 saturation!)
(PaO_2 35 mmHg)- even worse when anaemic!

DEVELOPMENTAL MILESTONES: quick reference 5.06

APPROXIMATE PHYSICAL GROWTH

DOUBLE BIRTH WEIGHT AT 5 - 6 MONTHS

TREBLE BIRTH WEIGHT AT 12 MONTHS

150% BIRTH LENGTH AT 12 MONTHS

Average age of walking 19.6 months (range 16 - 28 months)

Quietens in response to sounds	4 weeks
Vocalises to talked to	6 - 8 weeks
Two or three words with meaning	12 months
Five - 20 words (recognises many words)	18 months

Deaf babies babble initially

ALWAYS take a parent's report of possible deafness SERIOUSLY

FINE MOVEMENT

Reaches	16 weeks
Feet and hands in mid-line	16 weeks
Plays with feet	20 weeks
Evolution of grasp	5-9 months
Transfers	6 months
Pincer grip	9 months
Two cubes	13 months
Ten cubes	3 years

HEARING

Responds to sound from birth	
Starts to consistently turn head to soft sound at ear level	12 weeks
Turns to soft sound 40 - 50 cm from ear	9 months

SOCIAL DEVELOPMENT AND UNDERSTANDING

Smiles socially	4 - 6 weeks
Shows pleasure in familiar, pleasurable situations, eg bottle, bath	12 weeks
Laughs	16 weeks
Stranger anxiety	6 - 7 months
Concept of permanence	9 months
Plays pat-a-cake and peek-a-boo	9 - 10 months
Knows and turns to own name	12 months
Domestic mimicry	15 months