

CARDIOLOGY IN CHARGE SYNDROME: FOR THE PHYSICIAN

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TYPE AND FREQUENCY OF CONGENITAL HEART DEFECTS (CHDS) IN CHARGE

- ◆ 75% - 85% have a CHD
- ◆ Almost every type of CHD has been described, including "typical" VSD, ASD, PDA. Many children have multiple, complex congenital heart defects .
- ◆ There are proportionally more conotruncal and aortic arch CHDs (30-40% of those with a CHD)

Type of CHD (below)	Lin	Wyse	Tellier	Round #
# patients studied *	53	50	40	
Conotruncal, aortic arch (1)	42%	47%	33%	30-40%
Simple shunts, R/L obstruction (2)	32%	50%	57%	40-50%
AV/endocardial cushion (3)	15%	10%	10%	10%
Miscellaneous (4)	10%		5%	5%

* many had more than one CHD

(1) Conotruncal: tetralogy of Fallot, double outlet right ventricle, aberrant subclavian artery, right aortic arch, truncus arteriosus, interrupted aortic arch (type B), conoventricular VSD.

(2) Shunts: atrial septal defect, ventricular septal defect, patent ductus arteriosus, pulmonic stenosis/atresia, tricuspid stenosis/atresia, aortic stenosis, mitral stenosis, coarctation, hypoplastic left heart syndrome.

(3) AV canal: complete atrioventricular canal, atrial septal defect, primum.

(4) Misc: complex single ventricle, anomalous pulmonary venous return, others not specified.

DIAGNOSTIC TESTS AND CAVEATS

1. Echocardiography . In addition to intracardiac anatomy, imaging should look for aortic arch anomalies, such as vascular ring and aberrant subclavian artery.
2. Catheterization provides additional information about pressures and anatomy. In one study, 1/4 of the children with CHD also had renal anomalies. Although renal ultrasound and IVP/VCUG are more definitive, **delayed abdominal films** at the time of a cath may be useful in screening for urinary tract malformations.
3. Electrocardiogram (ECG) to study electrical activity. May be supplemented by longer term Holter monitoring.
4. Occasionally: treadmill stress testing, MRI scanning.

MEDICAL MANAGEMENT WITH CAVEATS

1. In one study of 50 CHARGE patients with CHD, 75% required surgery.
2. Children with CHARGE may be resistant to chloral hydrate sedation.
3. Anesthetic risk is increased in children with airway involvement such as **choanal atresia**, or **laryngotracheomalacia** (both common in CHARGE). Children with choanal atresia and complex heart defects have the highest rate of serious complications and/or poor outcome.
4. Swallowing problems with increased secretions (presumably due to involvement of cranial nerves IX and X) may present an additional risk of aspiration.
5. Prostaglandin administration may be risky because of a high mortality in CHARGE following prostaglandin administration.
6. Hypocalcemia may be due to the absence of the parathyroids in the small number of CHARGE children with classic DiGeorge sequence. However, hypocalcemia may also be due to hypoparathyroidism in other patients.
7. Heart problems are only one component of growth failure. Others include feeding problems, frequent illnesses (especially chronic otitis media and respiratory infections), and possibly growth hormone deficiency.

NON-MEDICAL ISSUES

Many children with CHARGE syndrome have dual sensory impairment (hearing and vision loss) in addition to the medical problems. This combination (medical concerns along with sensory impairment) has a tremendous impact on development. All of these children will be developmentally delayed. With appropriate medical and educational intervention (including a deafblind specialist), many of these children will eventually function in the normal to above normal range of intelligence. Do not assume mental retardation based on early developmental delay.

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CARDIOLOGY: THE HEART IN CHARGE SYNDROME: PARENT INFORMATION

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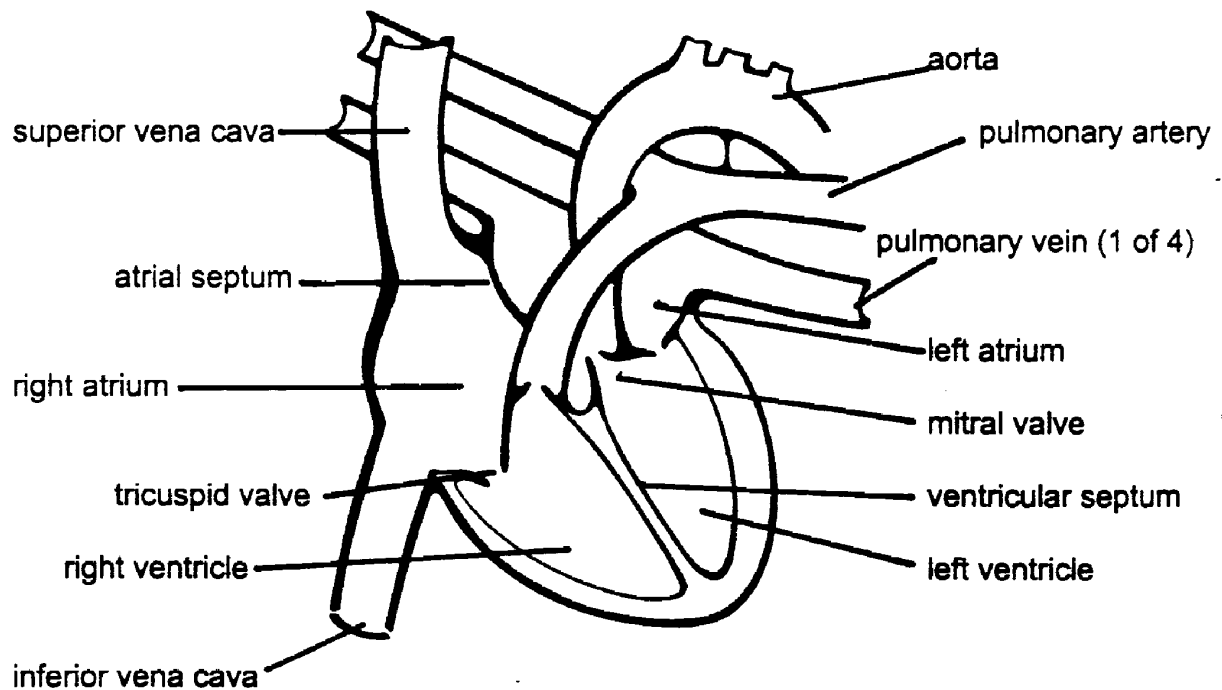
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NORMAL STRUCTURE AND FUNCTION OF THE HEART

The heart is a muscular pump (myocardium) with inflow & outflow pipes (blood vessels). It can be thought of as a house with:

- ◆ Two upper rooms (right atrium [RA], left atrium [LA])
- ◆ Upstairs dividing wall (atrial septum [AS])
- ◆ Two lower rooms (right ventricle [RV], left ventricle [LV])
- ◆ Downstairs dividing wall (ventricular septum [VS])
- ◆ Two one-way swinging doors between upstairs and downstairs rooms (tricuspid valve, mitral valve)
- ◆ Large pipes entering upper right room from body (superior vena cava, inferior vena cava)
- ◆ Smaller pipes entering upper left room from lungs (pulmonary veins)
- ◆ Two large pipes, one exiting each ventricle (pulmonary artery, aorta)
- ◆ Electrical system (conduction system)
- ◆ Internal plumbing (coronary arteries) which supply blood to the heart muscle itself



BLOODFLOW IN THE NORMAL HEART:

- 1) Blue blood returns from the body through veins to the **superior and inferior vena cava**, which empty into the **right atrium**. This blood passes through the **tricuspid valve** to the **right ventricle**.
- 2) The right ventricle pumps the blood through the **pulmonary valve** into the **pulmonary arteries**, which carry the blue blood to the lungs. The blood picks up oxygen in the lungs and turns red.
- 3) Red blood returns from the lungs through the **pulmonary veins** to the **left atrium** of the heart. From the left atrium, the blood passes through the **mitral valve** into the **left ventricle**.
- 4) From the left ventricle, the blood is pumped through the **aortic valve** into the **aorta** and from there throughout the body.

TYPES OF CONGENITAL HEART DEFECTS (CHD)

A. Holes in dividing walls, which can allow blue and red blood to mix

ASD: atrial septal defect (hole between upper rooms)

VSD: ventricular septal defect (hole between lower rooms)

AV canal/cushion defect: atrioventricular septal defect (large hole between upper rooms and lower rooms, including a hole in the floor)

B. Major plumbing problems with pipes, which can transport blood to incorrect chambers or restrict flow of blood through a vessel : ** These are very common in CHARGE

Truncus: arteriosus (1 pipe instead of 2 leaving the lower rooms)

Transposition: transposition of the great vessels (pipe positions switched leaving the lower rooms)

ToF: tetralogy of Fallot (1 pipe narrow, 1 pipe overriding, with hole in wall)

DORV: double outlet right ventricle (both pipes exiting leaving the right lower room)

Coarct: coarctation of the aorta (narrow pipe heading towards the body)

Interrupted aortic arch (pipe flow cut off heading towards the body)

TAPVR: total anomalous pulmonary venous return (pipe hooked to wrong room)

C. Leaky valves, which can allow backflow of blood through the doorway

Tricuspid or mitral regurgitation (backflow between lower and upper rooms)

Pulmonic or aortic regurgitation (backflow into lower room from pipe)

D. Tight valves, which can restrict flow

Tricuspid or mitral stenosis (narrowing of doorways between upper and lower rooms)

Pulmonic or aortic stenosis (narrowing of pipes)

E. Abnormal rooms

Single ventricle (one large lower room)

HLH: hypoplastic left heart syndrome (very small lower left room)

HRH: hypoplastic right heart syndrome(very small lower right room)

CHDs in CHARGE syndrome

How common are heart defects in CHARGE?

Approximately two-thirds (60-80%) of children with CHARGE have a CHD. Many, but not all, of them are serious.

Is there a typical pattern of heart defects in CHARGE?

Any heart defect is consistent with CHARGE, but ToF, DORV, and VSD are especially common. Some children with CHARGE have extremely complex heart defects that do not easily fit into just one category.

Are the typical heart defects mentioned unique to CHARGE? No, they are also common in:

VCFS/DiGeorge complex/del22q11

Hemifacial microsomia/Goldenhar syndrome

Retinoic embryopathy

DIAGNOSTIC TESTS WHICH MIGHT BE DONE

Chest x-ray

Electrocardiogram (EKG), Holter monitoring

Echocardiogram (echo, ultrasound)

fetal/prenatal

postnatal

Cardiac catheterization (cath)

Exercise test (stress test)

MANAGEMENT OF CONGENITAL HEART DEFECTS

Medication

- ◆ digoxin, to help the heart pump stronger
- ◆ diuretics, to get rid of extra fluid
- ◆ antibiotics, to prevent infection
- ◆ anticoagulants, to thin blood

Surgery

- ◆ to repair major plumbing problems (truncus, interruption of the aorta, ToF, DORV)
- ◆ to close holes in walls (ASD, VSD, AV canal)
- ◆ to repair loose valves (regurgitation)
- ◆ to repair tight valves (stenosis)
- ◆ to increase aorta blood flow (coarctation)

OUTCOME OF CHDS IN CHARGE

Medical outcome

Some heart defects can be totally repaired by surgery, while others can only be improved. Some children will end up with no heart problem at all, some will be much better, and others will continue to have problems with their heart. A few children with CHARGE will remain medically fragile for a long time, in part due to the heart defects.

The outcome and risks associated with heart surgery in CHARGE depend on the type of defect, the type of surgery, and on the presence of other serious health problems, especially choanal atresia tracheo-esophageal atresia, or cleft lip/palate. Many children with CHARGE have unusual reactions to anesthesia.

Developmental effects of heart defects

Remember that any child who is in the hospital for an extended period of time or who undergoes many procedures is under great stress. One outcome of the stress is delayed development. Some of the early delayed development in children with CHARGE may be attributed to multiple hospital stays and multiple surgical and diagnostic procedures. Some children actually lose milestones while hospitalized, only to regain them later. Do not be discouraged by early developmental delays, however extreme. Many children with CHARGE are truly "delayed" and will catch up over the years to come.