CENTRE CARDIO-THORACIQUE DE MONACO
Patient information sheet

Congenital heart disease

Characteristics, diagnosis, and treatment

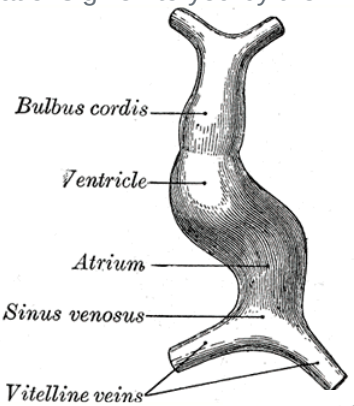
You or your child may be suffering from congenital heart disease. Understanding this pathology better will help you deal with this difficult situation. The cardiologists and entire health care team at the Cardio-Thoracic Centre of Monaco are here to provide support.
They will explain to you the specifics of the disease as well as treatment options.

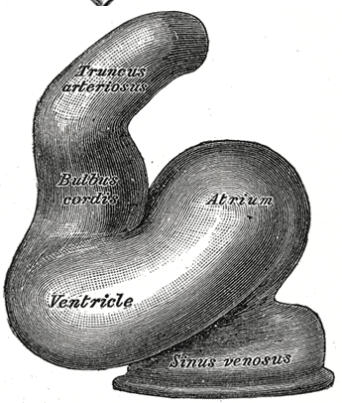
This document contains general information, intended to help you begin to understand the disease affecting you, what causes it, how it impacts health, as well as the techniques used in its diagnosis and treatment. It will provide the foundations so that you are able to research additional information more easily and ask us any **questions you may have, which are always most welcome**. Information contained in this document is in no way intended to replace the explanations given to you by the medical team.

What causes congenital heart disease?

The heart is formed between the **3rd and 8th weeks of pregnancy:**
initially it resembles a tube, which twists back onto itself and progressively develops into a structure comprised of **4 chambers** (2 ventricles and 2 atria) and **4 valves.**
It then functions as a **"pump" which circulates blood around the body** at every contraction.

Development of the foetal heart can sometimes be disrupted. This is the case in approximately **8 out of 1,000 births**.
9 times out of 10, it is impossible to determine the exact cause of the anomaly.





In 1 out of 10 cases, this may be caused by:

- a genetic condition or chromosomal abnormality,
- an infection (e.g. maternal rubella infection in an individual without protective immunity),
- intoxication (caused by certain medications, smoking, drug or alcohol abuse etc.).

The various types of congenital disease

Twelve underlying pathologies account for 90% of all heart defects.

These anomalies may be caused by

- the development of blood channels **which do not normally exist** within the heart walls (these are referred to as **"shunts"),**
- or a **total absence of partitions**,
- a **leaky** valve or one which remains **closed**,
- **blood channels which are too narrow** (forming regions of **"stenosis"),**
- the development of a **single ventricle,**
- or **poor connections** between veins or arteries and the heart,

When certain defects cause inadequate oxygenation of the blood circulating in the body, the blood appears darker and imparts a **bluish colour** to the skin, lips, tongue, nails.
This is known as **"cyanotic"** heart disease.

Some diseases are a combination of several deformities. These are known as **"complex"** defects.

Age of onset and age at diagnosis vary greatly depending on the type of defect.

Some surgical procedures need to be performed immediately after birth, whilst others require the child to reach a specific weight before surgical intervention can be considered.

In the case of multiple defects, surgery may sometimes have to be performed in several stages.

How are heart defects identified?

Diagnosis

Only 10% of the most visible heart defects are detected **by foetal ultrasound before birth**.

When a heart defect is suspected, the obstetrician will ask a paediatric cardiologist to perform a **detailed cardiac foetal ultrasound** to confirm the diagnosis.

- in cases of a deformity detected *in utero*, a **more extensive examination** can be carried out to detect any other potentially associated defects and to check for chromosomal abnormalities,
- in cases of heart defects deemed incompatible with life or likely to lead to a very severe handicap, on the parents' request, termination of pregnancy may be considered in agreement with the medical team,
- identification of all other defects prior to birth provides an opportunity to prepare the best possible strategies for the monitoring and birth of the child, including **delivery in a centre where specialised care can be provided from the onset**.

In 90% of cases, heart disease is diagnosed **after birth**, or even later during childhood or adulthood.

Several **indications** can lead to the early diagnosis of heart disease:

- **respiratory disorders** (very difficult and rapid breathing),
- **cyanosis** (bluish appearance)

- a **murmur** (an audible vibration detected by the physician during auscultation with a stethoscope, comparable to the purring of a cat) which may reflect the passage of blood through a **narrowed orifice** (stenosis) or through an **abnormal blood channel** (shunt).

The presence of any one of these signs requires a consultation with a paediatric cardiologist if the patient is a child and **three painless additional basic tests:** an **electrocardiogram** (ECG), a **chest x-ray** and **Doppler echocardiography. The latter is a key examination which should be performed by a paediatric cardiologist**.

- An electrocardiogram records the **electrical activity of the heart** by means of electrodes attached to the limbs (with cuffs) and to the chest (with small suction cups). This enables the physician to investigate the **rhythm of** the heartbeat, to identify any abnormalities (speed, regularity) as well as the heart chambers with an increased workload, which may help to pinpoint **the causative deformity** and to extrapolate its **impact** on overall myocardial function.

- An x-ray is used to assess the **volume and shape** of the heart and to determine how the deformity **affects** the **lung vessels**.

- Doppler echocardiography provides more specific information relating to the **anatomy of** the defect and heart **function**. This examination is not dangerous or painful to the child and can be repeated several times.
  If the recording is plotted out against time, as is the case with an electrocardiogram, the **mode of contraction of the heart muscle** and the **function of the valves** can be investigated: this represents the **"tm"** echo (or time motion) reading.
  The **ultrasound** beam quickly sweeps an entire plane of space in a continuous back and forth motion and converts signals from the various parts of the heart into an image which can be visualised, in "two dimensions" and in "real time" on a screen, which displays a reconstruction of the defective heart's anatomy, by combining multiple cross-sectional captures.
  The **Doppler** technique also analyses the **movement of** blood (the "**blood flow"**) present inside the chambers and vessels of the heart. This provides an estimate of **blood flow rates** and **intra-cardiac pressures**. The "colour Doppler" displays the **direction of blood flow** and the **different flow speeds** on the screen in real time.

The echocardiogram and Doppler have become so important in the diagnosis of heart defects, that in some instances, cardiac catheterisation is no longer necessary.

Specialised tests:

Heart catheterisation is still frequently necessary prior to surgery. In many cases, it facilitates intervention with no need for surgery. It is generally performed in a hospital setting. Since it is performed either under general anaesthesia or after administration of a sedative with a local anaesthetic, it is not a painful procedure for the child.

This technique is classed as "invasive", as it involves the introduction of a very thin tube inside the vessels and the heart, which are generally accessed via a vein or artery from the groin region.

The probe is advanced into the heart and its progress is visualised on a display monitor.

It enables **blood samples** to be collected for **oxygen content measurements** and to record **pressures** in the various heart **chambers**.

Injection of a "contrast" agent (opaque to X-rays) via the probe into the catheter, enables **the cardiac chambers to be filmed in detail**

and enables visualisation of the majority of **defects** and their effect on **the heart's** blood **circulation**.

New-generation **angiocardiography** techniques, known as "digital" angiocardiography, have a comparable image resolution without the need for injection of a contrast agent into the heart itself, but instead by intravenous injection, accompanied by an ultrasound. This approach can avoid any repeat catheterisation procedures, since heart disease progression and the obligation to assess the outcome of surgery can in fact mean that several surgical explorations are necessary over the years, even in children.

Other tests may be necessary for the diagnosis of congenital heart disease. They may require the injection of radioactive substances or radioisotopes to investigate the circulation and function of the heart in more detail.

- It is often important to understand the function of the heart during exercise which can be performed as from the age of 4 or 5.
  A **stress test** is an electrocardiogram recording taken whilst the patient walks on a treadmill or pedals lying down on a stress table.

- Cardiac MRI
  This technology does not emit radiation and is very safe. It can be used as many times as necessary in children from 4 or 5 years of age. MRI is the ideal follow-up examination for congenital heart dysfunctions, enabling **detailed analysis of the anatomy of the heart**, even in cases where interpretation of ultrasound is compromised by successive interventions.

- Scan
  This completely painless examination **detects coronary and vascular abnormalities**, at any age, even in babies.
  The advantage of combining new-generation "Flash" technologies with the accuracy of triggering image capture during diastole, is that the examination only takes between 5 to 30 seconds and therefore significantly reduces exposure to radiation.

Treatment

1 - Medical treatment

Medical treatment is required when heart defects are poorly tolerated and primarily focuses on preventing **heart failure**.
Other complications such as **heart rhythm disorders** or **blood anomalies** due to poor oxygenation may also require treatment.

Heart failure is treated with diuretics and sometimes with a group of medication known at vasodilators.

- Diuretics function by eliminating water, which accumulates in the tissues following heart failure.
- Vasodilators, indicated after major heart failure, decrease the heart's workload by dilating blood vessels as much as possible; their use in children is very complex and recommended only in a hospital setting. Maintenance therapy is based on diuretics and vasodilators which are administered in **very specific doses**, and adapted to each child. This requires **specifically dosed capsules** to be prepared by the pharmacist. The same approach applies to other medications for the treatment of heart rhythm disorders, which contain very active compounds.

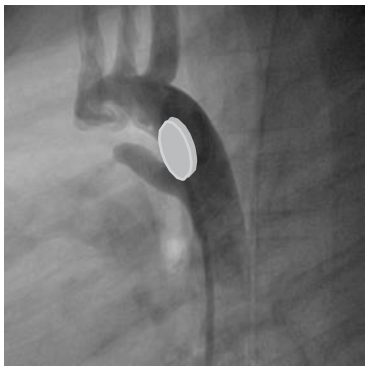
- In very particular cases, an anticoagulant treatment to prevent blood clot formation, also needs to be administered.
  This necessitates **frequent blood tests** and a **follow-up diary** showing the dose administered based on the latest blood test results.

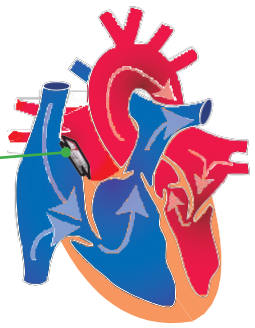
2 - Interventional catheterisation treatment

Currently many cardiovascular abnormalities can be treated by **interventional catheterisation**, with no need to resort to surgery.

It is possible to dilate constrictions of the pulmonary or aortic arteries with a **balloon catheter** introduced through the femoral vein or artery.

For abnormal blood flow between the left and right sides of the heart, such as patent ductus arteriosus (PDA) or atrial septal defects (ASD), specific "prostheses" have been developed: which, when deployed in the affected blood vessel, completely block the abnormal blood flow.





Digital angiography room

However, not all cardiovascular abnormalities can be treated using such an approach and many cases still require surgical intervention.

3 - Heart surgery

Many congenital heart conditions necessitate surgical treatment, but this does not mean that all children with heart defects require surgery. Heart surgery is indicated for children with poorly tolerated heart conditions which impede daily life, or for those who appear to tolerate their heart disease well, but whose defect has a high probability of poor tolerance at some point in the future.

Any surgical procedure involves some element of **risk**, even if it is relatively minimal, as is the case for the treatment of several specific heart defects (i.e. patent ductus arteriosus, atrial septal defect). The level of risk varies depending on a number of factors (age of the child, defect type, level of intolerance, possible complications, etc.) and will be **assessed on a case to case basis**.

The physician will explain to the child's parents how the surgical risk is significantly lower than the risk incurred by the child should he/she not undergo surgery.

Remedial intervention

Remedial intervention aims to restore as normal a heart anatomy and functioning as possible. Other than for several relatively routine cases (patent ductus arteriosus, aortic narrowing), this involves **"open-heart"** surgery.

During this type of surgery an **“extracorporeal circulation"** (ECC) system is implemented. An “artificial heart-lung” machine carries out the work whilst organs are stopped thereby enabling surgery on a heart emptied of its blood and at rest.

Post-surgery

The child is admitted to the **intensive care unit** for several days, where he/she progressively wakes up, but remains calm and sleepy thanks to the administration of sedatives.

"Artificial ventilation" is provided through a cannula intubated in the trachea and connected to a ventilator. Various sensors and infusion lines are connected to electronic monitoring devices and vials or pumps to administer appropriate medication and provide nutrient solutions.
The physiotherapists will initiate respiratory rehabilitation sessions in intensive care.

Post intensive care

Progress is generally rapid and respiratory physiotherapy sessions are continued as required, according to the child's bronchial condition, to improve independent respiration. It is performed under the close supervision of the physiotherapist.
In most cases, surgery enables resumption of daily activities, including all the collective physical activities engaged in by children of a similar age.

In some cases, a minor lesion remains, which however does not interfere with everyday life.

Repeat surgery may be necessary to correct either a residual or a more significant lesion.

In all cases cardiac monitoring is necessary, since a late-stage complication (such as heart rhythm disorder) can never be completely excluded.

It can sometimes be helpful to perform a catheterisation check-up, but monitoring is chiefly carried out using techniques which do not require hospitalisation (echocardiography, stress test).

Surgical scars are generally not a significant aesthetic issue.
In the case of open-heart surgery, a vertical incision is made in the middle of the sternum and extends its full length. In the case of closed heart surgery, an incision is made in the side, in the armpit between two ribs.

Cardiac and cardiopulmonary transplantation

In extremely serious life-threatening heart conditions, that are not amenable to restorative surgery, cardiac or cardiopulmonary transplants can be considered, the latter only if lung function is also affected (hypoplasia of the pulmonary arteries, tardive cyanosis).

Although these types of transplant are recent procedures and infrequent in children, short-term results are encouraging. They do however require constant and meticulous medical surveillance and the administration of anti-rejection medication for life.

Patients requiring a transplant, as well as pregnant mothers awaiting a baby with a significant heart defect, are referred to centres that have access to grafts or with a level III maternity ward connected to the paediatric cardiac surgery unit.

For further information on congenital heart defects refer to:

Fédération Française de Cardiologie
(French Federation of Cardiology):
http://www.fedecardio.org

Association Nationale des Cardiaques Congénitaux
(French Association for Congenital Heart Diseases):
http://www.ancc.asso.fr/
http://www.heartandcoeur.com/

References

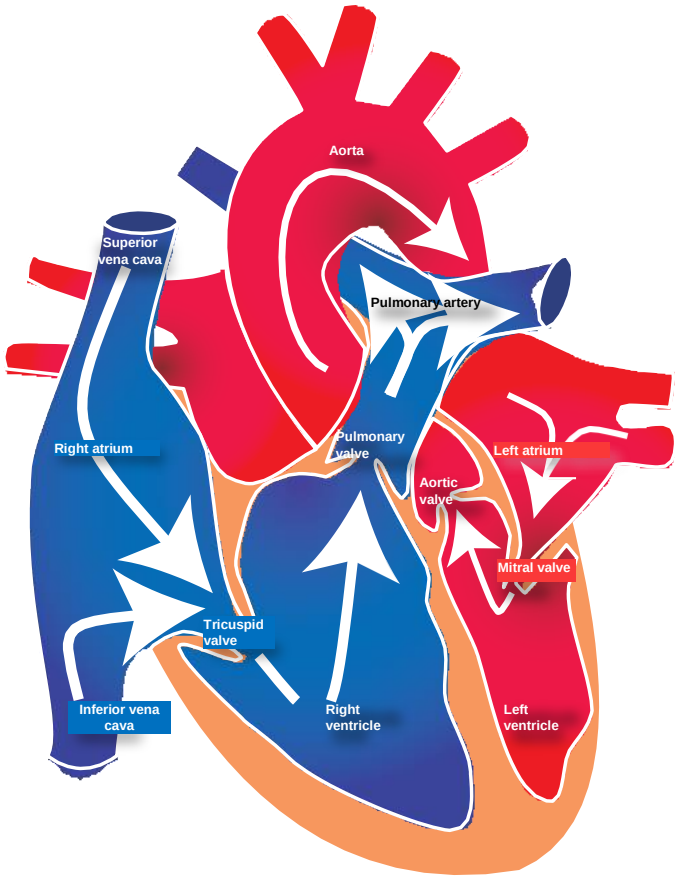
Fédération Française de Cardiologie
(French Federation of Cardiology)

Cardiologie Pédiatrique Pratique
(Paediatric Cardiology Practices)
Du fœtus à l'adulte
(From foetus to adulthood)
Alain Batisse, Laurent Fermont, Marilyne Levy.
Editions Doin

CENTRE CARDIO-THORACIQUE DE MONACO (cardio-thoracic centre of Monaco)
April 2017

Details of the pathology concerning:

(See more detailed sheet)



Monaco _____

Doctor _____