



### What is Congenital Vascular Malformation?

#### Birthmarks, Vascular Malformations and Anomalies

Vascular anomalies occur in barely 1 percent of all births. Yet, because of their rarity, their proper diagnosis and treatment is difficult, as most physicians do not see these problems often enough to become knowledgeable about their management.

#### What are they?

“Vascular anomalies” is an all-inclusive term for vascular malformations, vascular tumors and other congenital vascular defects. The more commonly used term, Congenital Vascular Malformation (CVM), implies abnormally formed blood vessels that one is born with. However, in spite of its redundancy, CVM is a popular term and it will be used here.

Birthmarks occur on the surface of the body and are relatively easy to deal with. Other vascular malformations can develop from any type of blood vessel and develop in any part of the body, although most involve the extremities. They represent defects or development problems that occurred during embryonic growth. Depending on the state of development at the time this occurs, the result can involve arteries, veins, lymph vessels, or combinations of these.

#### Birthmarks

The difference between a CVM and a vascular tumor or hemangioma (the medical term), both of which are commonly called “birthmarks,” is very important to a child. Although they may initially appear the same, “all birthmarks are not the same”. Most birthmarks represent a superficial vascular malformation, consisting of abnormal collections of small blood vessels near the skin.

Typically, this CVM type of “birthmark” does not go away nor does it enlarge, growing only at the same rate as the child. It thus maintains the same size and appearance indefinitely, is not a health threat and requires no immediate treatment. Some birthmarks, because of their location, particularly around the face and neck or on some other exposed body part, may be considered unsightly. Fortunately, the characteristic reddish color coincides with the range of certain lasers that can be used for their removal. Another approach has been to cover them up by tattooing them a skin color.

The other type of birthmark may appear the same at first, but is actually a vascular tumor or hemangioma. In contrast, this type grows rapidly in the months after its discovery, but then it “involutates” or gets progressively smaller. The majority disappear completely in a few years, leaving behind a patch of shrunken elastic skin. This regression normally is completed between two and eight years of age, but not all of them completely disappear. During their growth phase, these “juvenile hemangiomas” can be alarming, particularly if they grow in a critical location, such as those on the face impinging on the eye, nose, or mouth, in which case they may require treatment. However, most juvenile hemangiomas do not require treatment. Rather the best advice is to do nothing but wait it out, giving it a chance to go away by itself.

Both of these types of birthmarks, or the remnants of them, can be greatly improved in appearance by plastic surgery, but this is only occasionally needed and can usually be done after the child grows up.

# What is Congenital Vascular Malformation?

## Arterial-Venous CVMs

Other CVM are formed during early developmental stages when large connecting channels or shunts between future arteries and veins exist and for some unknown reason these artery-to-vein connections, or a cluster of them persist. Such connections are called arteriovenous fistulas (AVFs), or if there is a cluster of them they are called arteriovenous malformations (AVMs). These are potentially the most serious type of CVMs because in shunting blood from arteries to veins, they bypass the small vessels that make up the normal circulation beyond that point. This not only robs or steals blood that would otherwise pass through to more distant tissues and nourish them, but doesn't allow for a gradual pressure drop from the high arterial pressure to the low pressure on the venous side. Thus, these AVFs represent a high flow short-circuit and, depending on their size and location, they may force the heart to work harder. They may also cause poor circulation in the limb beyond the point of the AVF. In time, these AVFs tend to get bigger and have greater effect on the circulation. For example, an AVF or AVM in an extremity can "steal" (reduce) blood flow to a foot or hand as much as a blocked artery would. Fortunately, AVFs located in the legs and arms are more common than elsewhere in the body and thus easier to deal with. Those involving pelvic vessels, or vessels to vital organs or the brain can be extremely difficult to treat without injuring the organs or tissues surrounding them. Although AVFs make up only one third of all CVMs, they attract the most attention because of the serious problems they create, and are the CVMs most likely to need interventional treatment.

## Venous CVMs

CVMs composed entirely of veins are the most common, comprising almost half of the total, and are of two basic types. The more primitive ones appear as thin-walled lakes in which venous blood collects and when they develop in groups or clusters, they may form a mass consisting of a collection of grape-like clusters of these venous lakes. This type usually does not affect the venous circulation which returns blood to the heart, but



these malformation can be unsightly, cumbersome or be the site of a type of blood clot (not the type that travel to the heart or lungs). They are not a serious threat, but may be worth treating if the mass is large and causes local problems, for example, interfering with walking.

The other type of venous defect involves the large deep or central veins and often interferes with their function. Segments of major veins may be absent or narrowed. Some segments may be greatly widened and expanded (dilated), called a venous aneurysm. Treatment depends upon how severely they affect venous return or contribute to deep vein thrombosis (DVT). Most of the venous malformations involve only short venous segments and do not require treatment.

## Arterial CVMs

CVMs of the arteries are the least common and responsible for only one to two percent of the total. The most common arterial defect involves a segment that did not develop. The result is that a normal arterial segment is missing and instead blood flows through an undeveloped side channel or collateral artery, which persists rather than withers. Although this allows a bypass of the blockages, the enlarged bypassing segment often becomes more vulnerable to compression and injury, developing into an aneurysm or suddenly clotting off. The most common example of this is the so-called persistent sciatic artery.

## Symptoms of CVM

When located in an extremity, CVMs may show up as a birthmark, a visible or palpable mass of blood vessels, may stimulate the development of collateral blood vessels in the form of varicose veins, or produce an enlargement of the limb or a lengthening of the limb by stimulating its bony growth centers. The localized masses may be of various sizes from small to huge. At their surface the vessels may be vulnerable to injury and bleed or may even break down and ulcerate. AVFs may cause “ischemic” pain, which is the medical term for pain that results when circulation is so restricted that the tissues and the nerves serving them, do not get enough blood.

## How is CVM Diagnosed?

Years ago, the only definitive way to evaluate blood vessel problems was by the injection of contrast dye that would make them visible on x-ray, called an angiogram. However, since most CVMs do not need treatment, or it is delayed until the need for treatment is more obvious, it is now rarely necessary to get angiograms as a first step. They may ultimately be needed, but only when an intervention is required and even then are best obtained just before or at the time of treatment.

Fortunately, great strides have been made in less invasive forms of vascular imaging. The localized superficial CVMs can often be initially studied by a form of ultrasound imaging, called a color duplex scan. Larger mass lesions can best be studied by magnetic resonance imaging (MRI) which images in multiple planes (view angles), determines the anatomic extent of the malformation and importantly, whether its involvement of surrounding tissues (muscles, nerves, bones and joints) might preclude or complicate surgical treatment.

## CVM Treatment Options

As a general rule, CVMs should be treated for specific indications: persistent pain, ulceration, bleeding, blood clots, obstruction of major vessels, causing progressive limb asymmetry by overgrowth, and for cosmetic indications or because the vascular mass is cumbersome and leads to a badly misshapen limb or interferes with extremity function in a mechanical way. Since most of the patients with the worst CVMs show up early in life, the timing of any intervention should be planned according to a child’s growth and development. Often it is better to delay operating on very young children, if possible.

In the past, the only treatment for these vascular anomalies was surgical removal. However, of the CVMs that are significant and justify surgery, only 10-15 percent are removed. Removing even the simplest of these vascular malformations could lead to significant blood loss and is a surgical risk.

Surgery may still be appropriate for localized, accessible lesions, but in the last few decades, techniques using catheters have been developed. Catheters are placed (usually through a groin vessel) into the lesions and the malformed vessels are blocked, or embolized, with a variety of injectable particles, substances or devices such as polyvinyl foam, biological glues and absolute alcohol.

These catheter embolization techniques can be used to control lesions without surgery. They can also shrink larger CVMs to make them more treatable by surgery. Laser therapy may also be effective for small, localized birthmarks (port wine stains). Patients with a rare venous malformation (Kleppel–Trenaunay Syndrome) of the limbs, frequently benefit from elastic garments and bandages used for compression of the large veins. After careful evaluation, surgery or less invasive therapy of the enlarged superficial veins can also be helpful.