

CHAPTER 19 VASCULAR MALFORMATIONS

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Introduction

The difference between a **hemangioma** and **congenital vascular malformations (CVMs)**, both of which are commonly called “birth marks”, is important to recognize.

Hemangioma

Vascular **hemangiomas** are a big concern to the family and are rather common. A **hemangioma**, commonly known as a “**strawberry birthmark**”, is the most common benign tumor of infancy. They are often not seen right at birth and most are seen in the head and neck but can sometimes involve other parts of the body. They are more common in girls. These vascular growths have a very active **blood vessel** lining so grow very fast during the first year of life to the fear of the parents but then stop growing and actually get smaller very slowly so that most have disappeared by school age. Because the presence of the **hemangioma** can be alarming to the family, education and reassurance that it will go away is important information. In rare cases, since the head and neck is involved, vision problems, feeding problems and breathing problems can be seen. If the **hemangioma** involves the liver, heart or lungs, breathing problems or heart failure can occur. Generally, a drug used to reduce swelling (steroids) will take care of the problem. In very rare cases, **sclerotherapy** (injecting drugs directly into the tumor which can scar the insides) will decrease the size and allow normal bodily functions.

Congenital vascular malformations (venous malformations emphasis)

Overview

CVMs are different in that they have no rapidly growing **blood vessel** lining and do not go away. **CVMs** occur in approximately 1% of the general population. They occur for no apparent reason and rarely run in families (no genetic reason). **CVMs** involve **arteries** (**blood vessels** delivering oxygen and food to your cells from your heart), **veins** (**blood vessels** removing waste from your organs back toward the heart), and/or **lymphatic vessels** (**blood vessels** that clean up any excess in the tissues that the **veins** do not collect). A doctor’s meeting held in Hamburg, Germany, came to an agreement that **congenital vascular malformations** should be separated into five different groupings, namely 1) predominantly involving **arterial defects**; 2) predominantly **venous defects**; 3) combined **artery** and **vein defects** (shunting of blood between the two) 4) predominantly **lymphatic defects**; 5) combined **vascular** (all the above in addition to **capillaries** which are the normal connections between **arteries** and **veins**) **defects**. Each

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is further divided into when the normal development of normal **blood vessels** were stopped (arrested) during pregnancy. If stopped early on the way to forming the normal **arteries** and **veins**, the incompletely formed **blood vessels** have some of the growth properties of primitive **blood vessels** which means that they can grow when influenced by puberty, pregnancy or surgery. The lesion seen can be very small and well defined or more extensive involving muscle and even bone. In contrast, the more mature form does not grow, has main channels formed but they are formed abnormally leading to a total lack of normal **vessels** (aplasia), blockage where there should be normal **blood vessels** or the **blood vessels** can be too big (dilation). It is important to separate the different forms since the doctor must treat them differently. **Venous malformations (VM)** account for nearly 50% of all **CVMs** and there are a few more ladies than men affected.

VMs which stop maturing later in pregnancy

VMs which stop maturing later in pregnancy are present at birth but often **symptoms** are not noticed until the late teens and are most often seen in the lower leg. **Symptoms** (pain, swelling, skin changes and even skin wounds called ulcers) common to all **venous disorders** brings the patient to the doctor. The patient may have early **varicose veins** or locally enlarged **veins (phlebectasia)**. There can be blockage of **deep veins** either completely (**aplasia**) or partially (**hypoplasia**). **Aplasia / hypoplasia** of the **deep veins** may require the **superficial veins** as the exit route of blood from the legs or arms. Removing the **superficial veins** in these patients could cause major problems. Commonly, these patients develop a “**marginal vein**” on the outside of the leg which is not usually present and may be very important to getting blood out of the leg. Narrowing of the left **common iliac vein** (the big vein in the pelvic and abdomen which allows blood to exit the left leg) can happen as the right **common iliac artery** crosses over it to get to the right leg. It is called the **May-Thurner syndrome** or **iliac compression syndrome**. Compression and repeated hitting of the left **common iliac vein** from the right common iliac artery as it hits the underlying spine damages the **vein**. The majority of children affected are teenage girls on oral contraceptives showing up with a swollen left leg. If **blood clots** form because of the narrowing and injury, it is called **deep venous thrombosis**. The **deep veins** can be too large. **Avalvulia** or the lack of **vein valves** in both the **deep** (in the deep muscles) and **superficial** (just under the skin) **veins** often runs in families (has a genetic cause) seen as swelling and **varicose veins** when the child starts to walk. Significant **venous reflux** (backwards flow of blood in the **veins** into the lower leg) and sustained high pressure in the **veins** leads to swelling and **varicose veins** often before puberty. There can be very focal dilations called **aneurysms** which can become filled with **blood clot (deep venous thrombosis)**. These local dilations can happen in the legs, **veins** in the abdomen or even in the chest. Early studies to show that a **VM** is the problem should be a **magnetic resonance imaging (MRI)**, a study using magnets to see into the body). A **venous duplex** study (using sound waves to see into the body) will aid in determining what **veins** are involved and how the body is being affected. **Venography** (an X-ray study using drugs injected into the **vein** which help to see the **veins**) may be used to clearly show **vein** connections and prevent incorrect treatment during procedures. In general, these tests tell what **veins** are affected, if they are working well and what can be done to help **vein** blood flow. If acute **blood clots** are the problem, then ways to

remove the **blood clot** (clot busting drugs) or drugs used to thin the blood and stop more clot from forming (**anticoagulants**) are the correct treatment. If swelling is the only problem, **compression stockings** (external force pushing from the skin) can sometimes take away the pain, decrease and even control the swelling. If the stockings are not working and only the **superficial veins** are allowing backward flow of blood resulting in **varicose veins**, there are many ways to eliminate the **vein** including removing it by pulling it out (**stripping**) or more recently by scarring the inside of the **vein** with heat (**lasers** or **radiowaves**) or damaging drugs (**sclerotherapy**). The latter is usually done only when the child is somewhat older. If **deep veins** are involved, **superficial veins** can only be removed if the **deep veins** are open to allow blood to get out of the leg. If the **deep veins** are blocked, dilating the **vein** (**percutaneous venoplasty**) and placing a device in the vein to keep it open (a **stent**) can sometimes correct the problem. If the **deep veins** have no **valves** and **compression** is not working, then sometimes the surgeon can fix or replace the **valve** but this is uncommonly needed. Whether simple **vein** dilation should be removed or not is a decision your doctor must make with the patient's thoughts taken into consideration and that decision is determined by where the dilation is and what other structures lay close.

VMs which stop maturing early in pregnancy

Those **VMs** with growth that has stopped early in pregnancy do not look much like normal **veins** but more like a bundle of soft, spongy blood lakes that can flow into and become part of muscle, bone, fat and other organs. Since they go into fat, muscle, bone and other organs; it is very important the diagnostic studies are used which can picture where the abnormality is located, how the **veins** meet up and where, and what other organs are involved so a proper plan for treatment can be made. This usually means that **MRI** and invasive studies (studies using needles placed into the body) which directly put drugs easily seen by X-ray into the **veins** (**venogram**) and sometimes arteries (**angiogram**). The desire is to get rid of the malformations with as little damage to surrounding body parts. When causing symptoms from swelling and when compression is not working, the usual way to get rid of the abnormal cluster of **veins** is to put drugs that damage the **VM** directly into the **veins**. This means placing a needle right into the **VM** or by placing a needle into the **vein** which is far away and directly a very small plastic tube (a catheter) into the problem area. When properly in place; a drug (**sclerosant**), which can damage the **VM**, is pushed into it. The **sclerosant** can be high grade alcohol or a number of other drugs. Unusual problems sometimes seen after injecting these drugs are open areas and infection in the skin, **blood clots** within the **veins**, and even **blood clots** moving to the lungs (**pulmonary embolism**) that can stop you from breathing causing death or even strokes. Rarely is open surgery useful and in many cases simple compression and elevation is the best treatment.

Complex Venous Malformations

Venous malformations that have other **congenital vascular malformations** (complex **VMs**) as a part of the overall syndrome are rare but of concern if a person in the family has the problem. Although we are trying to move away from naming particular disorders,

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many people have come to know special conditions. **Complex venous malformations** are divided into those without (**Maffucci Syndrome, Proteus Syndrome, Klippel-Trenaunay syndrome**) and those with direct artery to vein connections (**arteriovenous shunting**) (**Parkes-Weber Syndrome**).

Maffucci syndrome

The **Maffucci syndrome** has a **venous malformation** (often large and in the fatty tissue just under the skin (subcutaneous)), abnormal growth of cartilage (a type of soft bony part of the body), and bone disfigurement. About 20% of patients can have changes into cancer which means that careful following of the patient is needed to catch any such changes early. If this does happen, removing the lesion is needed since other therapies do not work.

Proteus syndrome

The **Proteus syndrome** has several **blood vessel malformations** (**capillary, venous and lymphatic malformations**) combined with abnormal growth of bones, muscles, and fatty tissues. Children are usually born without deformity. As they age; tumors, skin, and bony growths appear, get worse and may involve more than half the body. Connective tissue nevi (birth marks) on the abdomen, hands, or nose are telltale signs of the disease. **Venous malformations** play a big role in the risk for **blood clot** in the **deep veins** (**DVT**), **blood clots** traveling to the lungs (**pulmonary emboli**), and premature death. No cure exists.

Klippel-Trenaunay syndrome

The **Klippel-Trenaunay syndrome** is a mixed **capillary, venous, and lymphatic malformation**. Symptoms vary from mild **varicosities** to massively enlarged unilateral lower limb involvement. A skin blemish or hyperpigmented area (dark birth mark on the skin) is commonly seen. X-ray studies may find lower leg **deep vein** abnormalities like to small or absent **veins** in the leg or abdomen and the presence of a **vein** which should have been replaced during the pregnancy (persistent **sciatic vein**). Mild **varicosities** and symptoms are managed best with **compression stocking** and leg elevation. If a particular skin malformation bleeds often then **sclerotherapy** might be a good treatment. Although recurrence of **venous varicosities** after removing the **veins** (**open, laser, radiowave removal**) is common, such treatment can help to decrease local pain, swelling and improve cosmetic image if the deep system is good enough to remove blood from the lower leg.

Parkes Weber syndrome

The **Parkes Weber syndrome** is distinguished by **high-flow** (rapid blood flow in the abnormal **blood vessels**) **arteriovenous fistulas** and problems that occur with such connections. Brightly stained skin which is warm to the touch as well as certain noises (bruit and thrills) which your doctor listens for can make the doctor think of this problem.

Problem with heart failure usually happen later in life but if the connections are very large this can occur even in infants. Most of these direct **artery** to **vein** connections are not easily gotten to for surgical removal. Thus, **sclerotherapy** has become the way to control symptoms.

Conclusions

A **hemangioma**, commonly known as a “**strawberry birthmark**”, is the most common benign tumor of infancy. These vascular tumors grow very fast during the first year of life to the fear of the parents but then stop growing and actually get smaller very slowly during childhood so that they are usually gone by school age. Only in rare cases is any treatment needed. **Congenital vascular malformations** are the result of **blood vessels** not maturing (going to full development) while the infant is still in the mother’s womb. If this happens early, the abnormal **blood vessels** do not have the form usually seen with **blood vessels** and appears more like a spongy mass which can involve neighboring body parts. If this happens later in the pregnancy, the **blood vessels** look more normal but are abnormally small, abnormally large or have unusually connections with other **blood vessels**. How bad the symptoms are will determine the need for treatment. As a part of **congenital vascular malformations**, the **venous malformation** is the most common and possibly the easiest to manage. **Congenital vascular malformations** do not go away and will require a lifetime of care.

Commonly asked Questions

My child has a reddish spongy mass on the cheek, what should I do?

You should see your pediatrician. He can tell if this is a **hemangioma** or what some people call a “**strawberry birthmark**” which may look bad now but will go away with time. This is a rather common problem in children. There are other much less common problems which have a similar appearance but must be managed differently. One such condition is the **congenital vascular malformation**, which results from an abnormal maturing of **blood vessels** as the baby develops the mother’ womb. The doctor will know how to tell the difference and when more testing or treatment is needed.

My doctor thinks that my baby had a congenital venous malformation, what does this mean?

A **congenital venous malformation** means that some of the **blood vessels** which should have matured into normal **veins** did not make it to the final stage of growth. If this happens early, the abnormal **blood vessels** do not have the form usually seen with **blood vessels** and appears more like a spongy mass which can involve neighboring body parts. If this happens later in the pregnancy, the **blood vessels** look more normal but are abnormally small, abnormally large or have unusually connections with other **blood vessels**. How bad the symptoms related to the malformation are will determine the need for further study and/or treatment.

My seven year old has a birth mark on the left leg and back, a varicose vein that I just noticed that goes down the outside of the leg, and his leg on that side may be a little bigger than the other side. What could be the problem?

Your child might have a syndrome called the **Klippel-Trenaunay syndrome** which involves an abnormal maturing of **veins, lymph blood vessels** and the normal connection between **arteries** and **veins** (the **capillary blood vessels**). It is a **congenital vascular malformation** which has been present since birth but just noticed now because your child is up walking and playing. X-ray studies may find lower leg **deep vein** abnormalities like too small or an absent **vein** in the leg or abdomen and/or the presence of a **vein** which should have been replaced during the pregnancy but remains to help get blood out of the leg. The lateral **varicose vein** may also be helping to remove blood from the leg so is important to have. Early on all that is needed to take care of symptoms is the use of support stockings. Your doctor will be able to tell you if this is the problem and, if so, if other treatment will be needed.

My child has a congenital venous malformation, are we at risk for other children with the same problem?

Most **congenital venous malformations** do not run in the family, in other words, are not genetically determined. There are some that do have a genetic basis especially certain syndromes. You would have to discuss this with your doctor to know for sure if this is a concern for your family.