

A close-up photograph of a young girl with light brown hair and green eyes. She is smiling, showing her teeth. On the left side of her face, there is a large, irregular, purple and pinkish lesion, which is a glomuvenous malformation. She is wearing a purple patterned top. The background is blurred.

Glomuvenous malformations

Information for families

Great Ormond Street Hospital for Children
NHS Foundation Trust

This leaflet explains about glomuvenous malformations, what causes them and how they can be treated. It also explains what to expect when your child comes to Great Ormond Street Hospital for assessment and treatment.

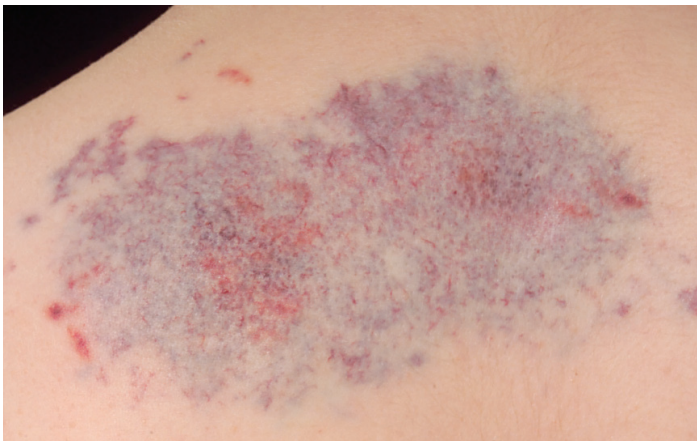
What is a glomuvenous malformation?

A glomuvenous malformation is a type of vascular malformation made of abnormal accumulations of blood vessels and glomus body cells. Glomus body cells as a normal part of the skin and are associated with temperature regulation. In cold weather, they regulate blood flow away from the surface to keep the body warm. In warm weather, they improve blood supply to the skin surface for cooling.

What do they look like?

Glomuvenous malformations can appear anywhere on the skin, either as single or multiple marks. They can be present at birth (congenital) or appear later in life. They are common on the fingers and toes, but can develop anywhere, including the mucous membranes inside the mouth and eyelids. Glomuvenous malformations do not appear within internal organs, but they can appear within the muscles underneath the skin.

Each child is different, although in most cases, they look like pink or blue raised



lumps. They usually thicken over time and turn darker blue in colour and more uneven in texture. The lump on the skin may swell when blood flow increases, for instance when crying, exercising or in hot weather. However, they do not lighten in colour (blanch) when pressure is applied to the area. Due to their colour, they can wrongly be confused with severe bruising. However, unlike bruises, they do not fade over a period of days. They can be painful, particularly if knocked or banged, or when there is an extreme change in temperature, or pressure on the mark.

How are glomuvenous malformations diagnosed?

They are best diagnosed with a combination of tests and scans. A skin biopsy is needed to confirm the presence of glomus body cells. An MRI scan gives a precise picture of the glomuvenous malformation, its location and extent. Giving an injection of a radioisotope dye before the MRI scan highlights the blood flow within the area.



What causes a glomuvenous malformation?

Research suggests that multiple glomuvenous malformations are inherited in an autosomal dominant way. This means that one parent carries the faulty gene. This has now been identified as a glomulin gene defect, which is involved in the regulation of smooth muscle cells surrounding specific blood vessels. This discovery may lead to improved diagnosis in the future.

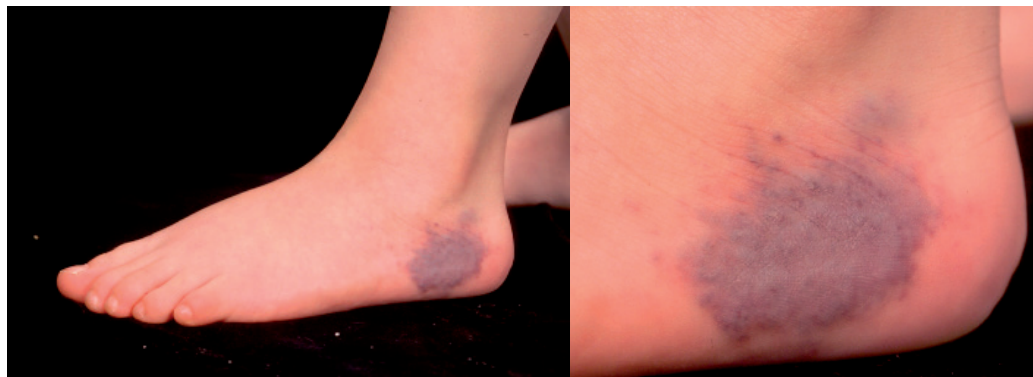
How common are they?

We know that glomuvenous malformations are rare, although we have no exact figures for the number of people diagnosed with them.

Single glomuvenous malformations are more common than multiple ones. They can affect anyone of any age. They are also more common in females than males but more research is needed to discover why.

What day-to-day care do glomuvenous malformations need?

Generally, lesions do not need any special care on a daily basis. They can be painful, however, particularly if knocked or banged, or when there is an extreme change in temperature. Sometimes they start to hurt for no reason. Pain can be treated by applying a cool compress and by taking pain relief such as paracetamol or stronger analgesia may be required. The lesions should not bleed any more than any other cut or graze so normal first aid should be used, that is, applying pressure until the bleeding stops. Applying pressure may be painful but may not be needed for small cuts and grazes.



How are glomuvenous malformations treated?

There are various options for treatment depending on the number of lesions, their size and location. Treatment should be considered if the lesions are causing problems. One option is to remove them surgically, although this may not be possible for multiple or extensive glomuvenous malformations. Laser treatment with a multiplex laser may be of benefit in some cases. The multiplex laser is a combination of pulsed dye and Nd:YAG lasers, which may help to shrink the blood vessels. Evidence of other treatment is anecdotal so more research is needed to explore these further.



Before laser treatment



After after treatment

In this child, the diagnosis of glomuvenous malformation was confirmed by biopsy.

What is the outlook for children with glomuvenous malformation?

Glomuvenous malformations can be painful but removing them surgically or treating them with lasers may be helpful. The colour of single glomuvenous malformation can be disguised using cosmetic camouflage but is not particularly successful for thickened areas.

If treatment is not possible, due to their size and number, the lesions may get bigger and move over time. Painful symptoms would be treated as they occur.

Conclusion

Vascular malformation research is an area of medicine that is continually advancing; studies have already given us improved options for treating glomuvenous malformations and continue to tell us more about how and why they develop. We hope that this leaflet has been helpful in learning more about your child's glomuvenous malformation; if you have any comments about it, please contact the Birthmark Unit.

Where to get further information

At Great Ormond Street Hospital

Birthmark Unit

Great Ormond Street Hospital, London WC1N 3JH

Tel: 020 7405 9200 ext. 0064

Support groups

Birthmark Support Group

BM The Birthmark Support Group, London WC1N 3XX

Tel: 0845 045 4700

Email: info@birthmarksupportgroup.org.uk

Website: www.birthmarksupportgroup.org.uk

Changing Faces

The Squire Centre, 33-37 University Street, London WC1E 6JN

Tel: 0845 4500 275

Email: info@changingfaces.co.uk

Website: www.changingfaces.co.uk

Notes

Bethan-May's story

I find it annoying people staring at me and asking if I have drawn on myself. I get a bit fed up at times having to take medicines and do creams and go to hospital. But I have met some fun people because I have GVM. The Doctors and Nurses are really nice to me and I get a day off school and I get to make things in the playroom at GOSH. It is okay having GVM but my Mummy and Daddy help make things easier for me.

Jacqui's story

Being a parent of a child with GVM is a learning process with each year with increasing concerns of function for her and how society and her peers will accept her looking so different. Daily as a Mum I am aware of the environment for her, from temperature, activities and risk to injury. I help her to pace herself so not to over exerting herself, tiring. Helping her to be comfortable, reducing pain by position, cool packs, paracetamol, and TLC, giving medications, applying creams, checking skin are all just part of every day. Keeping the school on board to her needs and making sure they follow the instructions and pass on information. Teaching Bethan-May how to deal with the general public, in a positive way. Making sure when she is in other's care they support her emotional side as well as physical.

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Compiled by the Birthmark Unit

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