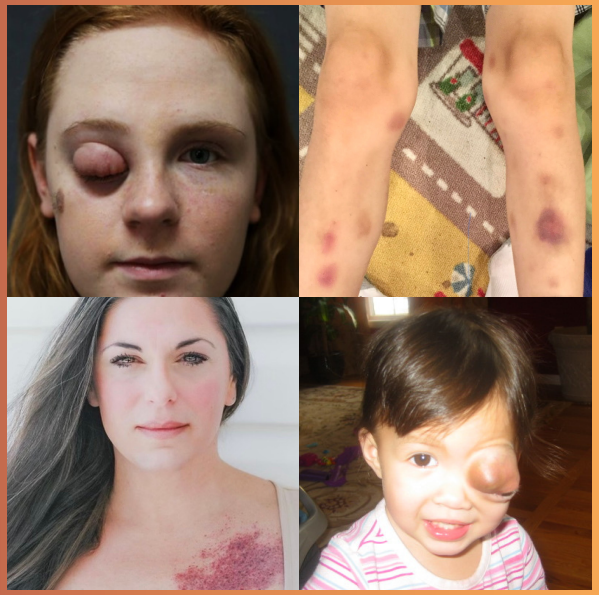

Lymphatic Malformations (LM) and Associated Anomalies: Frequently Asked Questions

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Lymphangioma, Cystic Hygroma, Kaposiform Lymphangiomatosis (KLA), Lymphangioma Circumscriptum (LC)

Note about LMs

The lymphatics serve, among other things, as collecting and transfer systems for tissue fluids. They collect excess fluid from tissues and transport it through a series of small vessels back into the venous system.

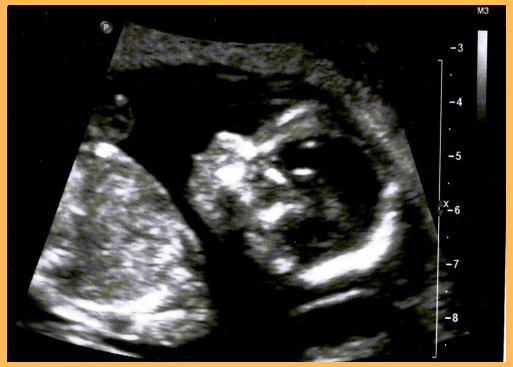
Furthermore, debris from dead cells, bacteria and viruses are removed from the interstitia and conveyed to lymph nodes to be degraded and analyzed. For this to properly function, the lymphatic vessels must be fully functioning. With an LM, the transfer of this fluid through these vessels is slowed down (most of the time), owing to a genetic mutation. The excess fluid accumulates within these vessels and dilates them, resulting in a swelling of the affected area. This is what you see with an LM. Sometimes, bone is also involved and is thicker than normal due to infiltration of the bone from the LM (very rare) or because an excess of growth factors will permeate from the stagnant fluid and influence the growth of the bone.

Q: Is a Lymphatic Malformation the same as a Lymphangioma and/or a Cystic Hygroma?

A: Yes. "Lymphangioma" and "Cystic Hygroma" are now referred to as a Lymphatic Malformation, which is a congenital malformation, or anomaly, of the lymphatic system which can result in dilated vessels that form a localized (focal) or diffused mass.

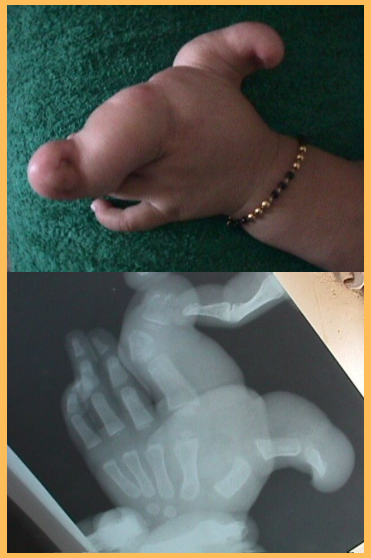
Q: Are these lymphatic malformations formed during fetal development? Can they be seen on ultrasound, and is it true that some spontaneously disappear just before birth?

A: Yes. LMs are considered genetic mutations and as such they are present in the womb, even if they are not evident until years after birth. Most times they can be seen on ultrasound, if the baby is cooperating. There are reports of upwards of 12% of LMs spontaneously disappearing just prior to delivery, but no one knows why this happens.



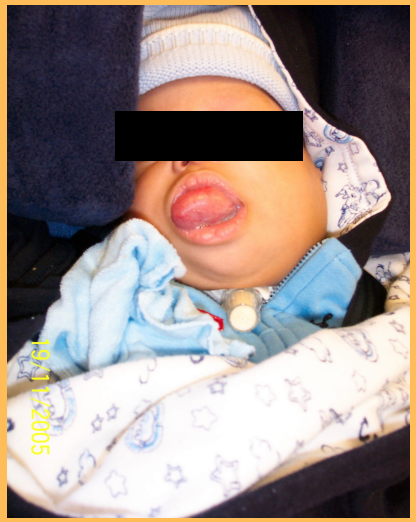
Q: Are LMs always present at birth or can they happen later in life?

A: The defect is there, and although it may not be visible at birth, it will manifest at some stage. Some are present at birth but others can become noticeable following infection, trauma, or during times of hormone changes. They are very hormonally-driven lesions so times of hormonal fluctuations, such as puberty and pregnancy, can see “changes” in these lesions. Most are apparent upon imaging.



Q: What is the difference between microcystic, macrocystic, and mixed LM?

A: Microcystic LMs (pure) have small cystic spaces and, therefore, are often not treatable with sclerotherapy as they need a more prominent cystic space, which is evident in macrocystic LMs. Frequently, LMs are "mixed," which is a combination of both.



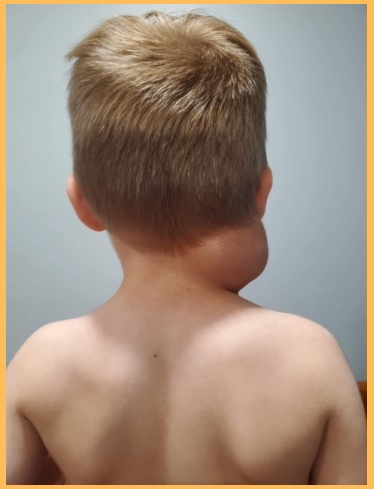
Q: Will an LM ever go away on its own, or does it always need treatment?

A: No two lesions are alike. Most will require some intervention but it is always the patient's right to observe or treat. There are anecdotal reports of spontaneous resolution of LMs in the early ages after birth.

Q: My son had his LM surgically removed as a baby as it was very large. Will it come back? He is 5 now and there has been no new growth but I'm worried about when he enters puberty. His doctor wants me to put him on Sirolimus for the rest of his life to keep it from growing again. I don't know what to do.

A: There are cases where surgically removed LMs can result in a complete resolution to the lesion, though many experts are reluctant to say they are "cured" as they can appear during hormone changes, trauma, or sickness. Some experts believe that certain medications can keep LMs from growing, but there is not sufficient evidence to support putting someone on this drug unless there is active disease that is not amenable to surgery or sclerotherapy.

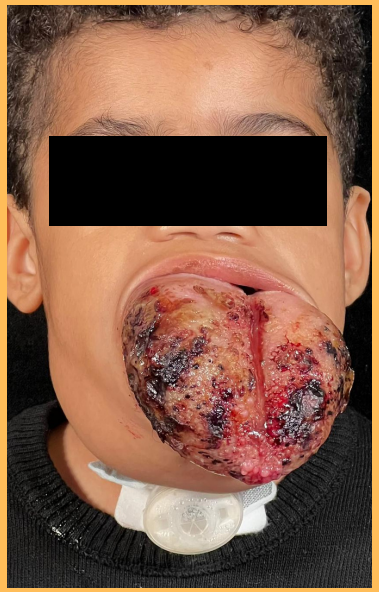
Q: Every time my son has a fever his LM in his neck enlarges, but then it goes back down when he is better, especially if he is on an antibiotic. However, it never fully goes away. It seems like each time it swells and shrinks it stays a little bigger and bigger. If he has surgery, the next time he has a fever will it come back or will surgery cure it?



A: This is a tough one to answer. Some LMs do not return if the surgery is done properly by an expert. The same is true with proper sclerotherapy. The accurate treatment of these lesions can yield great results but only when done by an expert with experience. Lesions that persist should be considered for surgery or sclerotherapy.

Q: My son has a tongue LM. We are currently managing it as it is stable. He is 18 months old. He has had no treatment to date. Should we only get one vaccine at a time or can we do multiple shots at once? Should we do live vaccines or inert? What about the COVID vaccine?

A: What we do know about these lesions is that sickness can make them worse. However, there is no scientific evidence to support any type of approach (one vs multiple vaccines). There is a general agreement that COVID vaccine is recommended for anyone with a vascular anomaly.



Q: I am 21 years old and I have a recurring LM in my lower lip and tongue. I have had a lot of surgeries and sclerotherapy. I am on birth control but my OB/GYN is concerned that the birth control is making my LM worse. Is it safe to be on birth control? Does it make it worse or better? Should I be on a specific birth control?

A: There are reports that birth control can make LMs worse. If a correlation can be established in a patient, then another form of birth control is preferable.

Q: Do LMs happen everywhere on the body? I mostly see them on the face, in the discussion groups, but my son has a large LM on one of his buttocks.

A: While most LMs occur in the head and neck region, they can appear almost anywhere.

Q: My 7-year-old daughter was diagnosed with an LM on one side of her neck after she had an accident on her bicycle. It came out a few days after the accident. The ENT we are seeing said that it was always there but the accident traumatized that area and made it more noticeable. Is this true? Can she get more in the future? She is currently on Sirolimus but I would like other options.

A: Trauma can make an LM "present itself" for the first time, even though nothing was ever noticed prior to this trauma. Some LMs are just dormant until a triggering event like trauma, sickness, surgery, etc. For LMs of the head and neck, surgery and/or sclerotherapy should be considered.

Q: My 9-year-old son has a mixed LM in his cheek that goes down along his jawline. He has been on Sirolimus for a year and it is stable but he constantly has mouth sores and does not want to take the medication. What other options do we have?

A: As previously mentioned, surgery and/or sclerotherapy are alternative options for treating LMs. However, an indication to which protocol is better suited for the patient must be the result of a multidisciplinary discussion. Mouth sores are a reported frequent complication of Sirolimus.

Q: I am a 72-year-old male who has had an LM of my head and neck since I was 9 years old. I had many treatments but it is still there. Am I too old to try the new drug that begins with an "S"? Will the drug shrink the bone overgrowth in my jaw? My chin sticks out and feels very heavy sometimes. Is that from the LM?

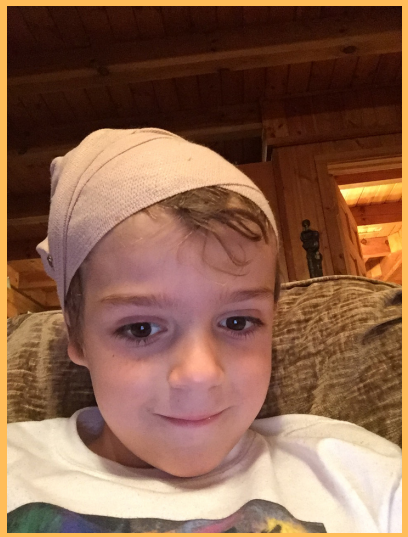
A: The drug you are referencing is called Sirolimus. You can try this drug if a multidisciplinary team believes it will be beneficial. However, there is no drug that will shrink bone overgrowth. Only surgery can correct bone overgrowth.

Q: My daughter is 14 and has an LM of her eye area. It pushes her eyeball out. The Hematologist wants her on Sirolimus but the surgeon wants to remove it. Both disagree with each other. I don't know what to do to save my daughter's vision. I don't want to make the wrong decision.

A: Sometimes, the best approach is a combination of surgery and drug therapy. A multidisciplinary team discussion is imperative in such cases. It is critical that any vascular anomalies of the orbit be treated to prevent permanent vision loss or impairment.

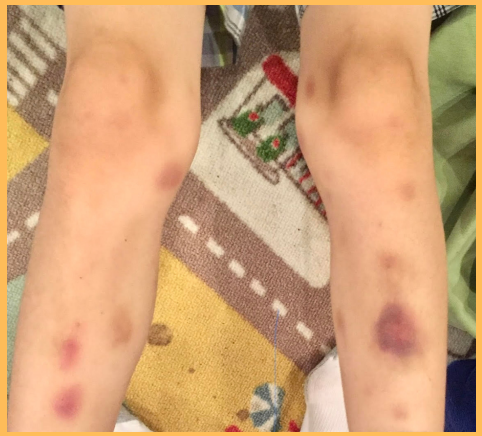
Q: What is KLA and how does it differ from other LM conditions?

A: Kaposiform Lymphangiomatosis (KLA) is a subtype of Diffused Lymphatic Malformation, just like the Generalized Lymphatic Anomaly (GLA) or the Central Conducting Lymphatic Anomaly (CCLA). It is probably caused by an abnormal development of the lymphatic system. However, KLA involves multiple parts of the body, especially the lungs and chest.



Q: What are the symptoms of KLA?

A: KLA usually starts in early childhood and can include shortness of breath and cough due to accumulation of fluid around the lungs and heart. Other common symptoms include chest and body pain, abnormal bleeding and bruising, and soft LM masses under the skin. The most feared complication is called Purpura, a disease caused by massive activation and consumption of platelets. This can lead to severe hemorrhages and constitutes a medical emergency.



Q: What causes KLA?

A: Recent evidence has shown that many cases of KLA are caused by mutations in a gene called NRAS. It is not inherited nor transmissible and is diagnosed based on symptoms, laboratory testing, and a biopsy of the LM lesion which has to be investigated for the probable mutations.

Q: What is the treatment for KLA?

A: Treatment is based on management of symptoms. Some physicians are using Sirolimus with or without other drugs such as vincristine and steroids. Surgery and interventional approaches can be used on large LMs that are part of the KLA.

Q: What is LC and how does it differ from the other LM lesions or any other vascular lesion?

A: Lymphangioma Circumscriptum (LC) is an LM confined to the skin and subcutaneous tissue. When primarily flat, it can appear as a stain similar to a port wine stain (capillary malformation) but it is not. You can usually tell because it has tiny bumpy vesicles and can burn, weep, and/or itch. A PWS does not behave like an LC.

Q: What is the natural history of LC, especially if left untreated?

A: Lymphangioma Circumscriptum is a progressive lesion, similar to all vascular malformations in that over time it can thicken and develop more vesicles which can actually ooze, burn, crust and bleed, if untreated.

Q: Is LC related to other Lymphatic Malformations?

A: Lymphangioma Circumscriptum is part of the Lymphatic Malformation family and is referred to as a Microcystic Lymphatic Malformation (MLM).

Q: Is an LC always superficial, deep, or both?

A: LC is a hamartomatous malformation of the lymphatic channels of the skin. It is always benign and has two components: a superficial vascular staining type lesion that can be seen, and a deeper cisternal element.

Q: What are the standard treatments for LC lesions?

A: Standard treatments such as laser (Pulse Dye), sclerotherapy and surgery have all been used with varying success. An LC lesion can be managed successfully. Radiofrequency ablation therapy is a new form of treatment that has recently emerged for an LC, but there is not enough data to report its success to date. Depending on the extent, surgery may be an option. Surgery, if radical, can cure LC. Bleomycin can also be injected into the affected area and can be quite successful.

Q: What is the future hope for LC?

A: The future hope for the management of LC is a combination of topical drug therapy and some form of laser or radiofrequency device which, together, may bring the best outcome. There is currently a research project which involves the application of a topical drug therapy, which is showing promising results. VBF will share outcomes from the new drug when the results are published.

Q: For any vascular anomaly in the lymphatic area, what type of doctor should be sought out for proper care?

A: These lesions are complex and as such, require a multidisciplinary approach. Unless the individual is an expert in treating and managing LMs, then a team should be sought out that manages the various aspects of LMs. Some of these experts may include a Surgeon, Hematologist, and an Interventional Radiologist. You may ask to see before/after photos of similar cases so that you can get an idea of what to expect. Whether or not these lesions can be 100% "cured" is controversial, although some experts have a history of treatments that have resulted in no further recurrence of the LM.

About the Authors

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