



TRANSCRIPT

S3:E2 – Birthmarks - Haemangiomas and Port-wine stains

Dr Blake Mumford: Welcome to season three of the *Spot Diagnosis*, a podcast about all things dermatological, brought to you by the Skin Health Institute in Melbourne, Australia. I am Dr Blake Mumford, Education and Research Fellow at the Institute.

Dr Anneliese Willems: And I am Dr. Anneliese Willems. I'm a GP, medical educator and Research Fellow at the Skin Health Institute. Blake and I are your co-hosts today. And today we'll be exploring birthmarks. We'll cover what they are, the different types, and clinically significant aspects to care. As a reminder for our GP listeners, *Spot Diagnosis* has been accredited with RACGP and ACRRM.

There is one point per episode, so approximately 9 to 10 points per season. All you need to do is subscribe to the podcast, listen to all the episodes and fill in a brief evaluation and reflection form on spotdiagnosis.org.au

Blake: Our guest today is Dr Susan Robertson. Susan is a dermatologist with a special interest in birthmarks and genetic skin diseases. She holds public appointments at the Royal Children's Hospital, Royal Melbourne Hospital, and Monash Medical Centre, as well as working in her private consulting rooms.

She is actively involved in teaching and is a supervisor of dermatology training at the Royal Children's Hospital. Dr. Robertson is an honorary fellow of the Murdoch Children's Research Institute. Susan, welcome to *Spot Diagnosis*. Can you start us off by sharing an interesting fact about birthmarks with our listeners?

Dr Susan Robertson: Funny you should ask that, Blake. There are many folk tales in traditional beliefs. Birthmarks can be interpreted as relating to someone's personality or character. For example, in Asian culture, hairy birthmarks are sometimes seen as a sign of great wisdom. Those that are particularly superstitious won't cut the hair coming from the birthmark for fear of becoming less wise.

You may have seen some examples of this with older generation Asian men sporting very long hairs growing from their facial moles. Some European cultures believe that birthmarks are the product of a particular food craving their mother had during pregnancy, or even that the birthmark results from trauma in a previous life.

Blake: Well, for everyone out there looking for their life partner, clearly the only criteria that matters is the presence of a birthmark with exceptionally long hair.

Anneliese: And moving right along, Susan, I certainly have lots of questions on this topic. A time I particularly have lots of questions around birthmarks is when parents come in with an infant for a six-week check. Parents, especially new parents, will point out something on their beautiful new infant skin and ask, "What is this?" And most importantly, "Is this something I need to worry about?" So, Susan, my first question is, how do you even approach this topic? What exactly is a birthmark?

Susan: The term birthmark usually refers to a skin lesion or a different-looking patch of skin present at birth or arising shortly after birth.

Anneliese: Now, I have a birthmark on my leg. In high school, when I found that my classmate had a matching one, we became firm friends. Was it particularly special that I found someone else with a birthmark? And how common are they?

Susan: Birthmarks are actually very common. It is estimated that more than 80% of newborns have some form of birthmark.

Anneliese: So I'm not as special as I thought.

Blake: You're very special, Anneliese. So what are the main types of birthmark?

Susan: Birthmarks can appear in a variety of shapes, sizes, colors, and textures on or under the skin. The two main types of birthmark are vascular and pigmented birthmarks. Vascular birthmarks, being made up of blood vessels, are usually pink, red, or purple in colour. The most common vascular birthmark is the naevus simplex birthmark, which is otherwise known as the stork bite, salmon patch, or angel kiss.

This usually occurs as a pale pink, V-shaped mark on the glabella forehead and the nape of the neck, and has a tendency to fade in the first few years of life. The second main type of birthmark is a pigmented birthmark, and they can be seen as different skin tones. They're quite often tan or brown in colour. The most common pigmented birthmark is dermal melanocytosis, otherwise known as Mongolian blue spots, which usually present on the lower back or the buttocks in darker skin infants, and also has the tendency to fade over time.

Blake: There are many types of birthmarks, but today we're going to focus on infantile hemangiomas, port-wine stains, pigmented congenital naevi, and Becker's naevus. Let's start with infantile haemangiomas, what are they?

Susan: Infantile haemangiomas, also called strawberry naevi or strawberry marks, are a common type of vascular birthmark affecting about 5% to 10% of newborns. They are benign growths made up of capillaries. They may be present at birth or arise soon after birth, and can resemble a faint red mark or even be mistaken for a scratch on the skin. But they have a tendency to grow and get bigger and more bright red over time, forming a lump that resembles a strawberry.

They can also develop under the skin and appear as a slowly growing skin-colored or slightly blue lump. The natural history is for quite rapid growth over the first couple of months of life, which then stabilises around 6 to 12 months of life. Then the lesions start to get reabsorbed by the body in a process called involution. This is a slow process, which can take many years.

Blake: If some of these involute spontaneously, does that mean not all of these need to be treated?

Susan: Yes. In most cases, infantile haemangiomas do not cause any problems and will eventually shrink by themselves without treatment. However, sometimes they can cause problems including ulceration, scarring, and disfigurement. Additionally, if the tissues around the eye are involved, blindness can result. So there are some instances when infantile hemangiomas require urgent assessment and treatment. Such cases include when the infantile haemangiomas threatens function or is likely to cause significant disfigurement.

For example, lesions involving any part of the face, particularly the eyelids, nose, lips, and ears, and also the chest or nipple area. Infantile haemangiomas involving the nappy area also need to be monitored closely as they are at a high risk of becoming ulcerated. Infantile haemangiomas on any body's side that are very large or have already ulcerated and are bleeding also require urgent medical attention. Such lesions are incredibly painful and require specialist multidisciplinary management, including dressings, laser treatment, beta-blockers or even surgery.

Blake: Dermatologists love showing off by diagnosing systemic diseases by their cutaneous manifestations. Can infantile haemangiomas be a marker of an underlying syndrome?

Susan: Yes. There are some instances when infantile haemangiomas on the skin can be a marker for underlying internal problems that require urgent investigation. Firstly, large, flat haemangiomas that cover a whole body region. These are termed segmental haemangiomas. These types of lesions can be associated with haemangioma syndromes termed PHACE, as in P-H-A-C-E, and LUMBAR, L-U-M-B-A-R syndromes, which can involve abnormal structure and vasculature of the central nervous system, cardiac, and multiple visceral abnormalities.

Secondly, infants with multiple, as in five or more, lesions are at increased risk of extracutaneous haemangiomas, mostly involving the liver. Interestingly, the visceral involvement can be significant, even if the cutaneous lesions are small. And if these visceral lesions are left undiagnosed and

untreated, they can lead to problems such as high output cardiac failure, consumptive hypothyroidism, and may even be fatal.

Blake: It's my favorite time of the podcast. First **skin tip** time, infantile haemangiomas involving the eye area, nappy area, or particularly large or numerous lesions should be referred for further investigation.

Anneliese: In recent years, there has been a real change in how infantile haemangiomas are managed. Can you please tell us a bit more about this?

Susan: Historically, we didn't have any good treatments for infantile haemangiomas. But this changed dramatically in 2008, when there was an accidental discovery made that propranolol, a beta-blocker medication, can be helpful. The first case was an infant in France who had haemangioma affecting the nose, and also had cardiomyopathy, which required propranolol.

Miraculously, within 24 hours of commencing propranolol, the child's doctors noted that the haemangioma changed colour and started to soften. Over the ensuing months, it continued to improve and completely flatten. A lot of research and clinical trials have taken place since that time. And we now know that it is not just propranolol, which is helpful, but a class effect of the beta blockers.

The most common medicine used to treat small and flat haemangiomas is Timolol, a topical beta-blocker. In more severe cases, oral medication is required, namely propranolol or atenolol. It is not understood exactly how beta-blockers work, but they are particularly effective in reducing the growth, size and redness of the infantile haemangioma.

Anneliese: Prescribing beta-blockers in children is not a risk-free affair, how should this treatment be initiated?

Susan: Beta blocker therapy for infantile haemangiomas is an off-label treatment that is usually prescribed by a specialist paediatric dermatologist. I, in fact, know a lot of general dermatologists are uncomfortable prescribing it. The reasons for this are the need for diagnostic certainty prior to treatment. And then the treatment does come with some precautions and the need for monitoring.

The children are usually managed in conjunction with a Paediatrician or a Paediatric Cardiologist. If there are cardiac issues or if the child is underweight, then there is a higher risk of complications, and treatment may need to be initiated under monitoring in hospital. Hypoglycemia is an important side effect that I warn parents about because it can manifest as increased lethargy, reduced conscious state or even seizures and death.

The risk of hypoglycemia is significantly higher when the child is unwell or not feeding properly. And for this reason, systemic beta-blockers should only be administered with feeds and when the child as well.

Anneliese: I think it's time for our second **skin tip**. Infantile haemangioma can be treated with topical or oral beta blockers. Treatment is not without potential significant side effects and should be ideally completed through a specialist paediatric dermatologist in a multidisciplinary setting.

Blake: The next birthmark we will discuss is pigmented congenital naevi. Susan, what are pigmented congenital naevi?

Susan: Congenital naevi are pigmented birthmarks made up of melanocytes, the pigment making cells in our skin. Similar to what are commonly known as moles, but they are present at birth and can be quite large and more lumpy and raised. They can range in colour from a light tan to even black.

Blake: Dermatologists have no doubt developed an elaborate classification system for this. Am I right?

Susan: They are usually classified based on size, which is according to their predicted largest diameter in adulthood. Small congenital naevi are less than 1.5 centimeter diameter, medium between 1.5 to 20 centimeters and large between 20 to 40 centimeters. Lesions measuring larger than 40 centimeters predicted adult size have been referred to as giant congenital melanocytic naevi.

Anneliese: Are there any congenital naevi in particular that we should be concerned about?

Susan: Solitary small and medium congenital naevi are essentially harmless, but can be a cosmetic nuisance. It is the large and giant lesions that can cause some problems. There is a 10% to 15% lifetime risk of melanoma developing within a large or giant congenital naevus. Also, individuals born with multiple naevi are at risk of Neurocutaneous melanosis, which is where the birthmark also involves the central nervous system.

This can lead to neurological problems such as seizures and abnormal development, and there is also a risk of melanoma developing within the central nervous system. Management of such patients is complex and multidisciplinary. We would recommend referring infants with large, giant or multiple congenital naevi for expert advice.

Blake: The next birthmark is a favorite of latte sipping Melbourne dermatologists, what are café-au-lait macules?

Susan: Café-au-lait macules are pigmented birthmarks that usually appear before a child is one year of age. They're usually flat tan marks, which is slightly darker than the rest of the skin. And they grow proportionately to the child.

Anneliese: Indeed. I used to think these were called café-au-latte macules. Should we ever be concerned about café-au-lait macules, Susan?

Susan: The café-au-lait birthmark is itself harmless and there is no malignant potential. However, multiple café-au-lait macules, particularly when there are six or more, can be a sign that the individual has an underlying genetic condition. Most commonly this is neurofibromatosis type 1, but there are a number of other genetic conditions such as Bloom syndrome and Noonan syndrome, which can be associated with multiple café-au-lait macules. Also, very large café-au-lait macules located on the trunk can be associated with McCune-Albright syndrome.

Blake: All right, before you go out and get yourself a latte, I think it's time for another **skin tip**. Multiple café-au-lait macules can be a sign of underlying genetic conditions.

Anneliese: And moving on to our next birthmark for discussion, port-wine stains. Now, when I think of port-wine, I'm taken back to a trip I had to Porto and the surrounding Douro Valley. Well, I'm not currently sitting in the Douro Valley, sipping on a flight of local port-wines. I am excited to be here discussing port-wine, or at least port-wine stains. Susan, what are these?

Susan: Port-wine stains are a common type of vascular birthmark seen in about 3 in every 1,000 newborns. They are a type of capillary malformation and usually look like a well-defined pink or red patch of skin, which is present at birth, usually affecting only one side of the body. Some lesions are small, while others can be quite large. It should be noted that the port-wine stain capillary malformation is a separate entity from the more common naevus simplex birthmark, otherwise known as stork bite, angel kiss or salmon patch, which we mentioned earlier.

They can sometimes be very difficult to distinguish. Unlike naevus simplex, port-wine stain capillary malformations will not fade, and if left untreated, they will grow proportionally as the child grows. In adulthood, they can progressively darken, thicken and become lumpy and may bleed. I have a fun fact for you. The most famous port-wine stain in the world belonged to former Soviet leader, Mikhail Gorbachev. He had a port-wine stain on his forehead.

Blake: And why should we be concerned about port-wine stains in particular, Susan?

Susan: Port-wine stain birthmarks may occur on any body site, but are commonly seen on the head and neck. If the port-wine stain is located on any part of the forehead, including the temple and upper eyelid area, there is about a 30% risk of epilepsy and or eye problems such as glaucoma. This

association is called Sturge-Weber syndrome. Since these problems can be present from birth, newborns with a port-wine stain birthmark affecting the forehead require urgent investigation.

Anneliese: And if we are concerned about a port-wine stain, how is it investigated?

Susan: Firstly, if you are not sure whether the birthmark is a port-wine stain or a naevus simplex birthmark, I would recommend referring to a specialist Paediatric Dermatologist, because it can be tricky to differentiate between these birthmarks at times. And we do not want to be causing unnecessary concern for the family or performing unnecessary investigations in these children.

For the high risk port-wine stains, which are those involving any part of the forehead, I recommend urgent referral to an Ophthalmologist, as the eye problems such as glaucoma can be present at birth. An urgent intervention may be required.

The screening for neurological involvement is more complicated and there currently isn't any international consensus on what tests should be done and when. Our preferred approach now is to refer the infants with high risk port-wine stains to the Paediatric Neurologists, regardless of symptoms, as the emphasis has been taken away from MRI scanning and put more towards counseling the parents about the symptoms and signs to look out for that could indicate early seizure development.

Seizures are the earliest presenting neurological feature of Sturge-Weber syndrome, and have significant consequences for neurodevelopment, so early recognition and treatment is critical.

Blake: Do port-wine stains ever fade spontaneously, like infantile haemangiomas?

Susan: Unfortunately not Blake, port-wine stain birthmarks are permanent. They do not shrink by themselves or disappear spontaneously. They grow proportionally to the child and may become darker or thick and lumpy after many years. There may be significant psychosocial impact on the child and effects on self-esteem, particularly for facial birthmarks.

This impact increases with age and has been shown to improve with treatment. We do tend to recommend treatment of port-wine stains involving the face and neck as early as possible, so that optimal improvement can be achieved by the time the child starts school.

Blake: And Susan, what form does this treatment take?

Susan: Vascular laser therapy, particularly with pulsed dye laser, is a safe, non-scarring treatment, which may significantly lighten these birthmarks. Multiple treatments are usually required.

Blake: It's time for another **skin tip**. High-risk port-wine stains in infants are those which involve any part of the forehead. These should have urgent referral to a Paediatric Dermatologist. Cosmetically sensitive port-wine stains should also be referred early for consideration of vascular laser treatment.

Anneliese: I've come across another more unusual birthmark called Becker's naevus. What exactly are these? Are they just another name for congenital melanocytic naevi?

Susan: No, they are quite different. Becker's naevi are in fact very odd. They are named after Samuel William Becker, an American Dermatologist, who first described these lesions. We characterise them as a birthmark, but they are not usually present at birth. They are kind of like a delayed birthmark because they often don't appear until puberty. They are a type of pigmented birthmark and are made up of melanocytes, hair follicles, and sometimes smooth muscle cells.

They appear as a large brown and sometimes hairy patch, usually located on the shoulder or trunk, and they can be prone to acne. There sometimes can be underlying soft tissue and bony abnormalities. Because of their development at puberty, we hypothesise that Becker's naevi are sensitive to hormones, particularly androgens.

Blake: I imagine this can be quite challenging. If a Becker's naevus suddenly becomes present in a young adolescent. How do we treat these?

Susan: Unfortunately, there are not many good treatments for Becker's naevi. Pigment laser treatment can be tried, but it tends to not be effective in the majority of cases. The hair can be removed by shaving or depilatory creams, and the acne can be treated with standard acne therapies. Another fun fact for you today, actor Richard Gere's birthmark on his left shoulder, which first featured in the film *American Gigolo* is arguably the most famous Becker's naevus, and it hasn't bothered him or affected his popularity in the slightest.

Anneliese: We've covered several different types of birthmarks today. When should I, as a GP, be monitoring or even referring birthmarks?

Susan: As a general rule, it would be good practice to refer any facial birthmarks to a Pediatric Dermatologist. The reasons for this are that there may be functional or cosmetic concerns which develop, and early intervention can be helpful, such as beta-blocker treatment for infantile haemangiomas, and early laser treatment for port-wine stains.

Also, facial birthmarks are at a higher risk of syndromic associations.

This is because the facial skin, particularly the forehead and nose areas, develop from the same part of the embryo as the brain and the eye. So if a birthmark forms early on in embryogenesis, it can

affect these internal structures as well as the facial skin, as is the case with Sturge-Weber syndrome. I would also suggest referring any unusual looking, large, or multiple birthmarks for evaluation as they may herald an underlying genetic condition.

Similarly, if there are any unusual features associated with the birthmark, such as over or undergrowth of body parts or concerns regarding development, this may be a clue to underlying syndromes.

Anneliese: I think it's time for our final **skin tip** of today. We recommend referring any facial birthmarks to a paediatric dermatologist, as these may have significant functional or cosmetic implications.

Blake: Okay. You've heard the last skin tip, but don't turn off the podcast just yet. We've got a fun section coming up. We're going to put Susan into the hot seat for a bit of a quiz.

Blake: Your first question for \$1,000. Are you ready, Susan?

Susan: Do I really get the \$1,000?

Blake: No.

Anneliese: Monopoly money.

Blake: You see a three-month-old baby girl with a strawberry naevus on her left nose. Over the last few weeks, this has started to grow and it is now one centimeter in diameter. Her parents are beside themselves. What can be done here?

Susan: Infantile haemangiomas on the nose are a high-risk site for long-term complications if they are not treated. This is because they can cause irreparable stretching of the skin and deformation of the nasal cartilage when they grow. In this circumstance, I would recommend referral to a paediatric dermatologist for consideration of systemic beta-blocker therapy. If the lesion ulcerates, this is yet another indication for systemic beta-blocker therapy.

Additionally, other treatments that may be required, such as regular dressings and pain relief until the ulceration has healed. We also sometimes utilise vascular laser treatment to help heal ulcerated hemangiomas. Down the track, if there was any residual baggy skin or scarring, further treatment with laser or surgery may be needed to help improve the appearance.

Anneliese: Your next question for \$10,000 in monopoly money, a six-year-old girl is brought in by her parents with a congenital nevus on her left lateral thigh. It measures about two centimeters in diameter and is dark. The parents want this removed. How do you manage this situation?

Susan: We actually see this quite a lot. The congenital melanocytic naevus, in this case, would be classified as medium-sized. In the first instance, I would be exploring the reasons why the parents want it removed. Sometimes it is because of concerns that the lesion is cancerous or could become cancerous in the future. I'm happy to be very reassuring to parents that medium-sized congenital melanocytic naevi such as this lesion are not of any increased risk of developing melanoma when compared to other parts of the skin. There, therefore, is no medical benefit from removing such a lesion.

However, if their concerns are cosmetic in nature, that is another thing entirely. The only way to remove congenital naevi effectively is complete surgical excision. This would leave a surgical scar in place of the birthmark and the family would need to be sure that they would prefer the appearance of the scar rather than the birthmark in order to proceed. Sometimes it is helpful to suggest waiting until the child is old enough to gauge their opinion on the matter.

Blake: Can the scar kind of grow with the child and become more prominent with age?

Susan: Yes, it can.

Blake: It's time for the final million-dollar question. A 20-year-old male presents with a Becker's naevus extending over the left anterior chest to the left upper arm. The lesion has been mildly elevated and hairy since he was 14 years old. He is a keen swimmer and finds it uncomfortable when the girls stare at it when he is at the pool. He asks what options there are for cosmetic removal.

Susan: This is a very tricky situation because Becker's naevi are typically non-responsive to laser treatment. I would be exploring what component of the birthmark causes him the most concern. If it is the hairiness, that's an easy fix. Shaving, depilatory creams, and sometimes laser hair removal can be helpful for managing the hair. If it is the pigmentation, then I'd have to be honest with him, that laser treatments are not likely to be effective.

If he is determined to give it a try, then my approach would be to do some laser test patches on a small area of the birthmark to gauge if it is likely to respond, prior to committing him to the pain and expense of treating the whole area with laser. Importantly, it would also be very helpful to refer patients for psychological counselling in order to help them come to terms with, and accept their birthmarks, particularly ones that are not able to be removed.

Blake: All right, well that concludes our episode on birthmarks. We hope it has left a mark.

Anneliese: Thank you, Susan, for your time and sharing your expertise with us.

Susan: Thank you. It was a pleasure. Thanks for having me.

Blake: We would like to thank Associate Professor Alvin Chong at the Skin Health Institute for intellectual oversight of this podcast. Also a huge thank you to Jo Coughlin from our education team at the Skin Health Institute.

Anneliese: We hope you have enjoyed this episode of *Spot Diagnosis*. Remember, these podcasts are not meant to replace medical advice. If you have a skin condition that requires attention, we strongly encourage you to see your medical practitioner.

Blake: For ambitious and intellectual listeners who want more information on this subject, a transcript of this episode and links to other resources can be found on our website, spotdiagnosis.org.au.

Anneliese: Please share *Spot Diagnosis* with your friends and colleagues. Rate and review us, let us know what you think. We would really appreciate your feedback and any suggestions. Thank you.

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