Sturge-Weber Syndrome: Frequently Asked Questions

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Q: What percentage of those with a Port Wine Birthmark (PWB)/Port Wine Stain (PWS) near the eye will be suspect for Sturge-Weber Syndrome (SWS)?

A: About 25% depending on the size, pattern, and extent. A midline pattern on the forehead has about a 30% risk. A hemifacial birthmark covering the forehead, the eye, and the cheek on one or both sides has the highest risk. The risk extends up to 50% with large bilateral birthmarks.

Q: Is the V1, V2, V3 diagnostic criteria still relevant? If not, what is the new criteria for location of stain in relation to probability of SWS?

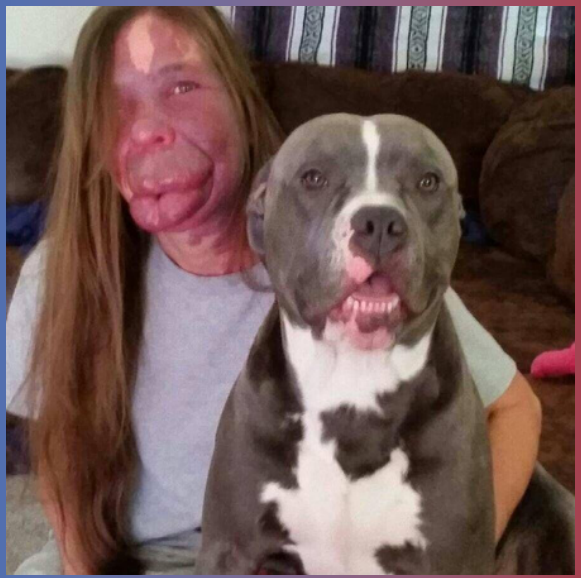
A: It's the upper eyelid, temple area, lower eyelid, and forehead; we call a PWB in this area of the face a "high-risk birthmark." Patients can have stains on the scalp and not be at risk for SWS. If there is a PWB/Capillary Malformation in any of these areas there is a risk, no matter how small. A tiny birthmark on the forehead can signal SWS.

Q: Does the density of the stain have any relationship to either an increase or decrease in probability of SWS?

A: There might be, but this has not been well-researched. The main issue is the location.

Q: What are the components of each type of SWS?

A: The Type 1, 2, or 3 for SWS are all caused by the same mutation, whether eye only, brain only, skin and/or any combination. The main issue is the location; they are always at-risk to develop eye or brain involvement. Brain involvement may be ruled out later, but a patient with a high-risk PWB is always at-risk for eye involvement and glaucoma. This is a spectrum disorder. Since the diagnosis is often confused, it is best to rely on the risk and location of involvement while in treatment.



Q: Is SWS the same as encephalotrigeminal angiomatosis?

A: Yes. That is another name that can be used, but SWS is the most common and current name.

Q: Is the GNAQ gene mutation the cause of SWS/PWB?

A: This is the most common cause by far. A small population can have a GNA11 mutation, and a very small number are caused by an undefined mutation.

Q: Is it typical that the brain involvement will be on the same side as the stain?

A: That is typical, although not always.

Q: Do you need to have a PWB on the face to have SWS?

A: No. 10% of patients have brain involvement without skin or eye involvement.

Q: When should a newborn with a facial PWB have testing done for SWS? Which type of testing should be done?

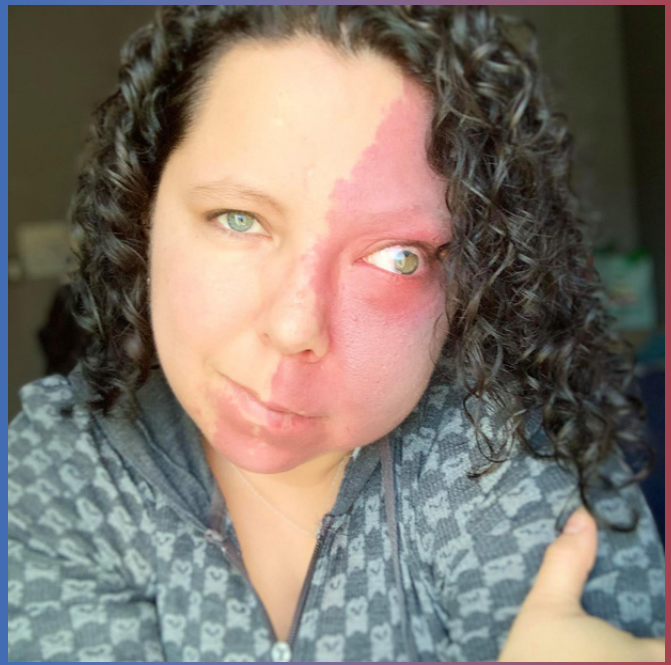
A: Eye involvement should be evaluated in the newborn with an eye exam to check for glaucoma and vasculature of the eye.

Q: If an MRI is clear two times in a row, does that mean the patient will never get SWS?

A: it depends on the age of the patient when the MRI is done and how the MRI is done. An MRI prior to age one is not definitive for SWS. It needs to be done with contrast and with proper sequencing. It's not about getting two done to rule it out. It is about getting them done properly, at the right age, and with proper MR sequences.

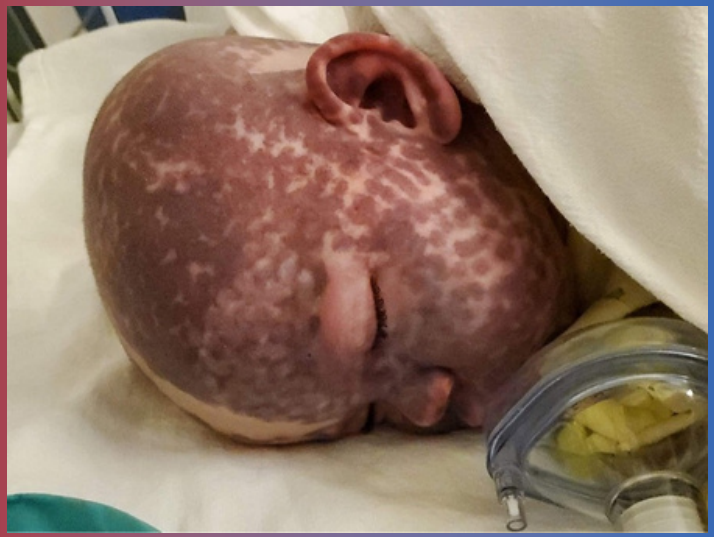
Q: Can SWS-related glaucoma or brain involvement present at any time in life?

A: Eye involvement can present at any age and must be followed by an eye doctor for life. Brain involvement can be determined on proper MRI imaging (see question above). In children with SWS brain involvement, 10% will have an onset of neurological symptoms after age two. However, these symptoms can occur at any age.



Q: What is the typical treatment for SWS-related glaucoma?

A: Typical treatment begins with medical (drug) management using eye drops (i.e. beta blockers). If medical management is unsuccessful, there are surgeries that can drain fluid from the eyes to alleviate pressure.

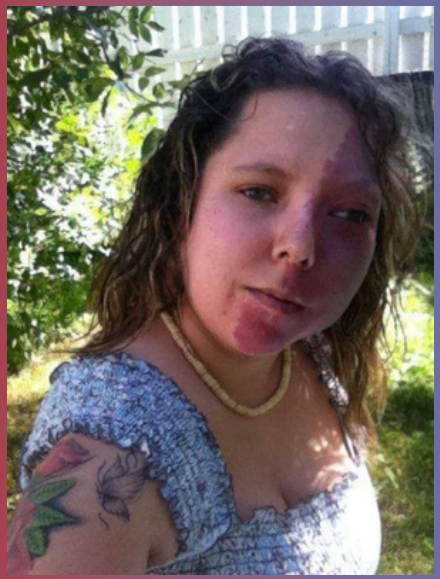


Q: What age should laser therapy begin for the birthmark present in SWS?

A: Laser treatments do not have to be delayed because of SWS. Treatment should be tailored to individual SWS cases.

Q: If the PWB is lasered to 90-100% clear, does that mean the patient will never get glaucoma or brain involvement?

A: No. Removing the birthmark does not have anything to do with preventing or reducing the risk of brain or eye involvement.



Q: Can SWS-related glaucoma present on the globe of the eye as bloodshot?

A: Yes. If the eye has increased blood vessel appearance this can indicate involvement. This patient should be examined and monitored closely.

Q: What is the latest treatment for controlling SWS seizures?

A: Seizure medication continues to be the standard treatment. The usual protocol at Kennedy Krieger Institute is to start with an anti-seizure drug and a low-dose aspirin.

Q: What is the risk of cognitive impairment from SWS-related seizures?

A: The risk of cognitive impairment increases with the frequency and severity of seizures. Onset of seizures under one year of age is associated with a higher risk of neurological issues. However, cognitive levels can improve with prolonged seizure control.

Q: Can a PWB be associated with cognitive impairment without SWS?

A: This can happen, but we do not have good data on PWB patients having cognitive impairment without SWS. It is not certain if the Port Wine Birthmark and cognitive impairment, or autism, are related when there is no SWS brain involvement with abnormal blood vessels.

Q: Can cognitive impairment be reversed or improved in SWS individuals?

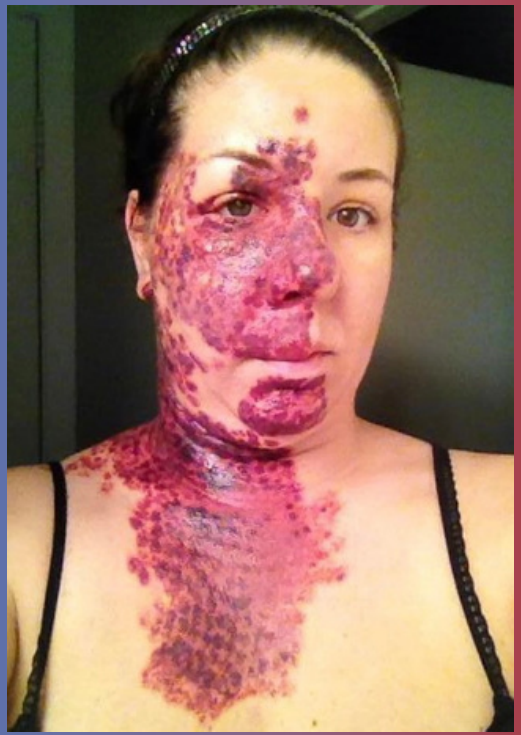
A: Yes, if seizures are well-controlled for a year or more.

Q: Does early diagnosis and intervention lessen the chance of serious complications?

A: There is some evidence that early intervention for glaucoma and seizures does improve outcomes. However, further study is needed.

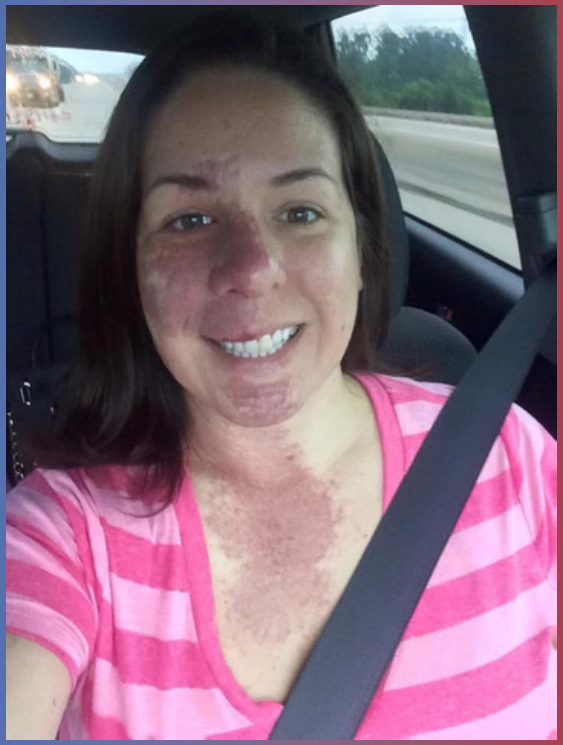
Q: Can an individual with SWS have a seizure from laser treatments?

A: Most SWS individuals tolerate laser treatments without any issues. Upon rare occasions, a patient may have issues that seem to be triggered by laser treatments. In those cases, laser treatments should be delayed, spaced out, or stopped.



Q: Should a pregnant woman with SWS take certain precautions?

A: There is not a lot of data regarding pregnant women with SWS. A woman with SWS who is pregnant should be cared for by a high-risk pregnancy obstetrician if possible.



Q: Do hormone changes such as puberty, pregnancy, and menopause affect SWS?

A: Puberty can definitely worsen seizures, migraines, stroke-like incidents, and mood issues. There is no conclusive data for pregnancy or menopause.

Q: Are there growth/stature differences in males vs females with SWS?

A: In general, there is an increased risk of hypothyroidism and growth hormone deficiency in SWS individuals compared to the general population, regardless of sex. However, there is data to suggest that males have a higher risk for cognitive issues. Further study is needed.

Q: What is the difference between a SWS seizure and an epileptic seizure?

A: They both result from abnormal electrical activity in the brain. The most important difference is that SWS seizures can harm the brain if they continue, as the seizing further impairs blood flow in the brain. There is no vascular anomaly in epilepsy, therefore there is a lower risk of stroke injury with prolonged seizures.

Q: Are there any alternatives to anti-seizure medications for controlling or minimizing the effects of seizures?

A: Yes. We frequently use Epidiolex®, a pharmaceutical grade cannabidiol oil. However, there are other useful alternatives such as:

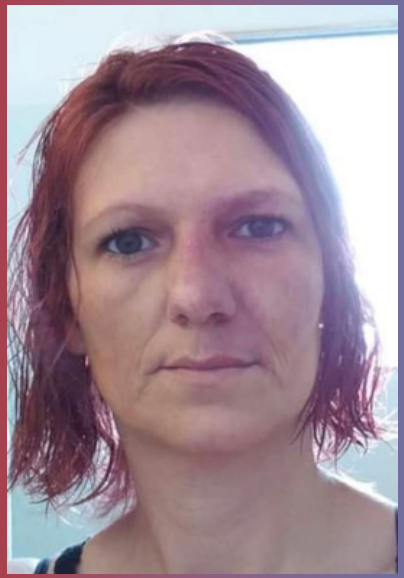
The Atkins and ketogenic diets are also helpful for controlling seizures, and there are even ketogenic baby formulas. However, it will only be effective if families are committed to keeping the patient on this diet.

VNS (Vagal nerve stimulation) is an option for patients who are not candidates for resection surgeries. In very select cases, other surgeries can be beneficial.

In terms of supplements, multivitamins and vitamin D are important. It is also very important to stay hydrated, get proper sleep, and eat healthy.

Q: Can cognitive damage occur from repeated seizures?

A: Yes, because prolonged/repeated seizures can cause further decreases in blood flow, which can injure the brain.

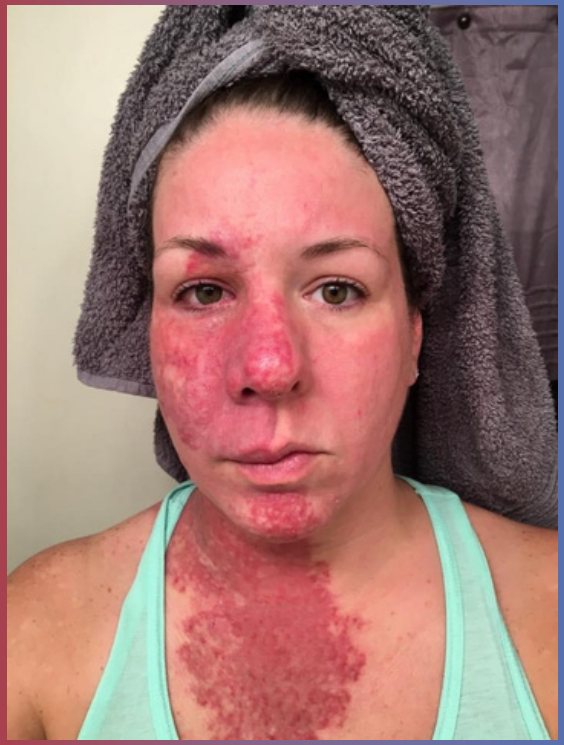


Q: Can cognitive damage from repeated seizures be prevented or reversed?

A: There is evidence that if the patient remains seizure-free for a year or more, that neurological and cognitive functioning can improve. Studies are currently underway to assess preventative measures.

Q: Can SWS result in bone or tissue overgrowth in the head and neck area?

A: Yes. The facial bones underlying the birthmark can thicken and can be associated with soft tissue or boney hypertrophy. This is most likely to happen with an extensive facial PWB.



Q: Can SWS affect ear, nose, and throat issues if the stain is in those areas?

A: Yes because the drainage of the ear and sinuses can be affected as well as the nasal cavity and inner ear. Snoring, sleep apnea, and repeated ear and sinus infections can occur.

Q: Are there interactions with drugs and alcohol that could impact SWS patients?

A: Recreational drugs and alcohol can interact with the metabolism of medications in the liver and can increase the risk of seizures, stroke, and migraines.

Q: Are there any activities that should be limited or avoided by individuals with SWS?

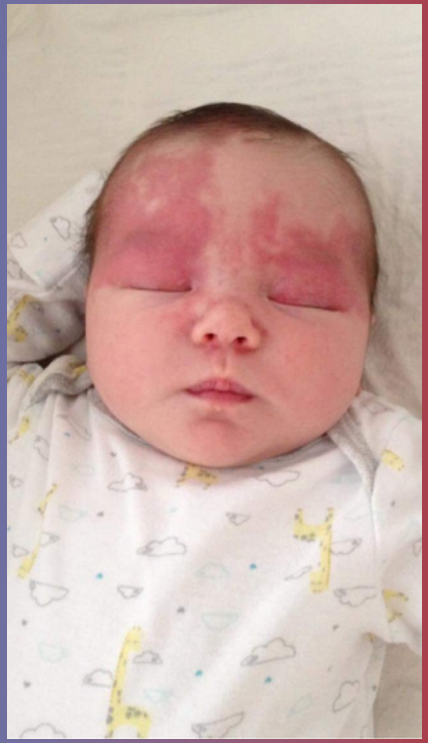
A: Individuals with SWS with brain involvement should steer away from aggressive contact sports or activities that can cause brain trauma. Specifically, sports that require the use of a helmet and involve frequent blows to the head (football, hockey, soccer, lacrosse, rugby, etc.).

Q: Should SWS patients notify their primary care doctor about their condition?

A: Yes. Primary care doctors should be involved in the treatment plan. They have an important role in making sure the child gets all vaccinations, and grows and develops properly.

Q: What is the earliest that SWS can be ruled out in a baby?

A: Brain involvement can be ruled out after age one with appropriate MRI imaging done with or without contrast and appropriate sequences. Eye involvement can never be ruled out and should be checked annually.



Q: Can individuals living with SWS collect disability?

A: Yes. They can in some cases as this is a medical condition that can affect a person's ability to work.

Q: What percentage of individuals with SWS are considered disabled?

A: Unknown

Q: Is SWS hereditary? Can the GNAQ gene be hereditary?

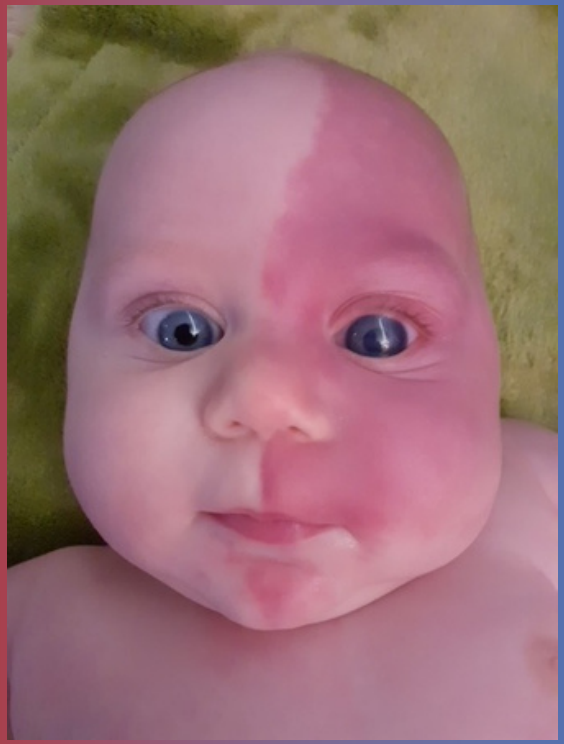
A: No, the current understanding is that it is not a hereditary condition.

Q: Has SWS ever been diagnosed in utero?

A: It has been retrospectively suspected. There have been some rare cases in which after a child with SWS was born, a review of the ultrasounds revealed brain involvement or a PWB. However, diagnosis prior to birth is not routinely common.

Q: If an individual with SWS has only a stain plus glaucoma, can they still develop brain involvement later in life?

A: If they have had the appropriate imaging after age one, they will likely not develop it later. Most of this is based on having definitive testing with proper SWS experts.



Q: What should parents of a new baby that may have SWS know?

A: They should have a SWS expert and center monitor the baby as this is a rare disorder and the knowledge of this condition is very limited within general pediatric providers.

Q: Will a baby who is “fisting” and exhibiting early signs of possible seizures display this “fisting” on the same side of the birthmark?

A: The “fisting” is usually on the opposite side of the birthmark. This is because, for example, the left side of the brain controls the right side of the body.

Q: If a baby has a bilateral Port Wine Birthmark covering both eyes, will they still only exhibit “fisting” on one side or will they display bilateral “fisting”?

A: It can be one side or both sides depending on where the brain involvement is.

Q: Is there anything that the parents could have done to prevent this mutation?

A: Not that we are aware of at this time.

Q: Should individuals with SWS take baby aspirin every day? Why?

A: Many patients with SWS take low-dose aspirin to manage their condition. Low dose aspirin interferes with platelet function and is thought to improve blood flow through the abnormal blood vessels.

Q: Why are some SWS individuals sensitive to cold and others are sensitive to heat?

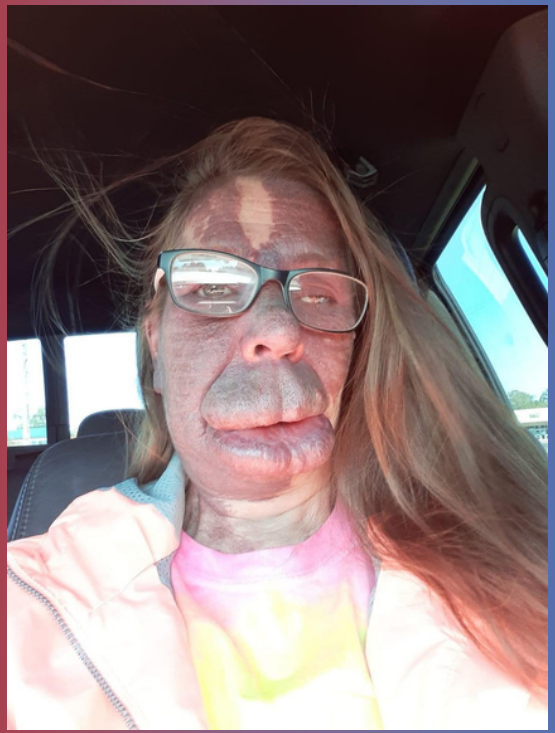
A: This phenomenon has been observed but not fully understood. It is normal for your skin to flush when you are warm and go pale when you are cold. With SWS, in many cases, the opposite phenomenon is observed. When a SWS patient is cold, skin can get darker and when warm, skin can get lighter. More research is needed to fully understand this.

Q: What cautions should individuals with SWS take if they have a high fever? Can this impact seizures?

A: A high fever can lower a seizure threshold, so patients may want to take Tylenol or Motrin and contact their doctor to assess the reason for the fever.

Q: Should individuals with SWS have routine imaging to track progression? How frequent?

A: We do not recommend “routine” imaging to monitor a SWS patient. Imaging should only be done if there is a significant clinical change or to address other issues that may require new imaging.

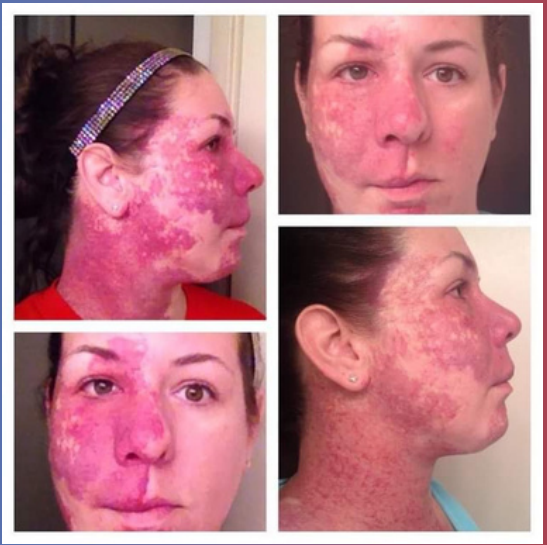


Q: Do individuals with SWS have a shortened life expectancy?

A: There is no published data on this at this time.

Q: Should women with SWS use hormonal birth control?

A: Birth control pills can be used by some patients but they should be aware that the pill can affect seizure medication. There is an increased risk of stroke with estrogen use, and the lowest possible estrogen dosage should be used. The decision must be weighed individually by each patient in discussion with their physician. There are other alternatives that are not hormone-based.



Q: When will there be a cure for SWS?

A: The best hope for a cure will be in gene therapy, a treatment approach that is currently successful for other conditions. There are researchers thinking about the possibility of this for SWS.

About the Authors

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https://www.kennedykrieger.org/patient-care/centers-and-programs/sturge-weber-syndrome-center

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