PORT – WINE STAIN

Yüzümle Mutluyum Derneği

1. What is a Port-Wine Stain?

A port-wine stain (medically known as *nevus flammeus*) is a congenital capillary malformation usually seen on the face. It typically manifests as a reddish-purple birthmark on one side of the face, covering the forehead or eyelid.

This birthmark can be the most distinct sign of a rare neurocutaneous (affecting the nervous and skin systems) condition called Sturge-Weber Syndrome (SWS). However, not every individual with a port-wine stain has Sturge-Weber Syndrome; the risk of developing this syndrome is related to the location of the stain on the face (especially the forehead and around the eyes)

2. What are the Symptoms of a Port-Wine Stain?

- **Skin Findings:** A pink, red, or purple mark present at birth, usually unilateral, and whose color can darken with age.

Skin Characteristics:

Critical Sign: The stain covering the forehead or upper eyelid is the most important sign indicating the need to evaluate the risk of brain and eye involvement (Sturge-Weber Syndrome).

Persistence: It is distinct at birth and, unlike some other spots that appear during infancy, it does not fade or disappear over time.

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Apart from the stain itself, if the condition is associated with Sturge-Weber Syndrome, the following symptoms may be seen:

- **Eye Problems (Glaucoma):** Glaucoma (eye pressure) characterized by increased intraocular pressure can be seen in 30% to 60% of patients. If this condition is not treated early, it can lead to vision loss
- **Neurological Symptoms:**

  - **Seizures (Epilepsy):** Seizures are seen in approximately 80% of patients, usually starting in the first year of life (average 6th month)
  - **Muscle Weakness:** Weakness on one side of the body (hemiparesis) may develop.
  - **Developmental Status:** Learning difficulties, attention deficit, or developmental delays may be seen

3. How is Port-Wine Stain Diagnosed?

Diagnosis is usually suspected at birth upon observing the characteristic stain on the baby's face. If the stain is particularly in the forehead or upper eyelid region, the risk of brain and eye involvement should be evaluated.

The following methods are used to determine the definitive diagnosis and the scope of the disease:

- **Magnetic Resonance Imaging (MRI):** Contrast-enhanced brain MRI is the "gold standard" for imaging vascular abnormalities (angiomas) in the brain.
- **Computed Tomography (CT):** Can be helpful in showing calcifications in the brain.
- **Examination:** It is essential for measuring intraocular pressure and evaluating the risk of glaucoma.
- **EEG (Electroencephalogram):** Used to monitor seizure activity.

4. Genetic Transmission: Is My Family at Risk?

One of the most common questions asked by families encountering this situation is "Did I pass this on to my child?" or "Will it occur in my other children?"

- **It Is Not Hereditary:** Sturge-Weber Syndrome and the associated port-wine stain are not a hereditary disease (passed down from mother or father). It is not passed from family to child
- **Cause:** This condition is caused by a "somatic mosaic" mutation in a gene called *GNAQ*.

- **When Does It Occur?:** This mutation occurs randomly during the very early stages of the baby's development in the womb. Therefore, there is no problem with the mother and father's genetic structure.

5. Counseling and Family Planning

Since the disease occurs "sporadically" (randomly):

- **Recurrence Risk:** The risk of the same situation recurring in the parents' subsequent pregnancies is no different from the risk in the general population.
- **Prenatal Diagnosis:** Prenatal ultrasounds usually appear normal, so it is difficult to detect during routine pregnancy follow-ups.
- **Counseling:** Genetic counseling can be reassuring for the family to understand that this condition is not their "fault" and that they do not carry a hereditary burden.

6. Risks and Follow-up (Why is it Important?)

Early diagnosis and regular follow-up are critical to improving the quality of life. Treatment is entirely symptom-directed and requires a multidisciplinary (different specialties working together) approach.

- **Eye Health:** Regular lifelong eye examinations are recommended. If glaucoma (eye pressure) is detected early, vision loss can be prevented with eye drops or surgery.
- **Neurological Follow-up:** Controlling seizures is very important to protect cognitive development. Medication treatments and, if necessary, surgical interventions can be applied.
- **Skin Treatment:** Laser treatments (photocoagulation) for the facial stain can be applied from infancy to lighten the color of the stain and control vascular dilations.
- **Psychosocial Support:** psychological support for children and families due to visible differences facilitates the processes of adaptation to school and social life.

This booklet is a general information resource prepared with the aim of increasing knowledge and raising awareness about Port-Wine Stain and Sturge-Weber Syndrome. However, it should be kept in mind that the information here does not substitute for professional medical advice, diagnosis, or treatment. The follow-up of the disease should be personalized according to your individual health status, the location of the stain, and the emerging clinical findings. The screening methods and intervals included in the booklet reflect general standards; your doctor may move these periods forward or change them according to your clinical needs. For any decisions and follow-up processes related to your health, please be sure to consult your specialist physician.

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