STURGE-WEBER SYNDROME

Sturge-Weber syndrome (SWS) is a rare neurological and skin disorder characterized by nervous system problems and a permanent birthmark, known as a Port Wine Birthmark (PWB), usually on the face.¹

SWS AFFECTS

1 IN 40,000 - 400,000 WORLDWIDE²
< 200,000 IN THE U.S.³
1 IN 50,000 LIVE BIRTHS IN THE U.S.⁴

PWB IS MORE COMMON

3 IN 1,000 NEWBORNS⁵
8% TO 15% OF PEOPLE WITH PWB DEVELOP SWS⁷
85% OF SWS CASES AFFECT ONLY ONE SIDE OF THE BODY OR BRAIN⁶

50-75%
72-93%
44-62%
30-71%

developmental delay
of all children with SWS have seizures
experience headaches
develop glaucoma

COMPLICATIONS OF SWS⁴

THERE IS NO CURE FOR SWS. APPROVED TREATMENTS INCLUDE:
LASER THERAPY
ANTICONVULSANT MEDICATIONS
NEUROSURGERY to remove or disconnect the affected part of the brain
PHYSICAL THERAPY for paralysis or weakness
EDUCATIONAL THERAPY to treat developmental delays
EYE DROPS OR SURGERY to treat glaucoma

THE SWF IS A DRIVING FORCE IN DISCOVERY, EXCELLENCE, ENROLLMENT AND FUNDING:

25 YEAR JOURNEY TO DISCOVER GNAQ GENE MUTATION THAT CAUSES SWS⁸

$88,000 OVER A TWO YEAR PERIOD ISSUED IN 2014 FOR TWO RESEARCH GRANTS TO CONTINUE RESEARCH

10 CENTERS OF EXCELLENCE ACROSS U.S.

HERE’S HOW YOU CAN HELP:
VISIT STURGE-WEBER.ORG

● BECOME A SUPPORT VOLUNTEER
● BECOME A FAMILY DAY COORDINATOR
● HOLD AN AWARENESS EVENT
● DONATE

ABOUT THE STURGE-WEBER FOUNDATION

The Sturge-Weber Foundation’s international mission is to improve the quality of life and care for people with SWS and Port-Wine birthmark conditions through collaborative education, advocacy, research and friendly support.⁴

For more information about SWS and The Sturge-Weber Foundation, please visit http://www.sturge-weber.org/.

REFERENCES

A survey on behalf of Global Genes reflects a growing appreciation among primary care physicians (PCPs) and specialists of the societal impact of rare diseases, while also underscoring the burden shouldered by patients and family members.

7.3
Average number of physicians seen before receiving a diagnosis.

"I welcome the challenge that rare diseases bring and want to be part of finding a diagnosis."

19% of PCPs felt their knowledge of rare disease was excellent or good at time of diagnosis.
59% rated their current level of rare disease knowledge as excellent or good.

59% of specialists felt their knowledge of rare disease was excellent or good at time of diagnosis.
77% rated their current level of rare disease knowledge as excellent or good.

"It would be helpful to receive additional training in rare diseases."

"I lack sufficient time to do a workup for a rare disease even when I suspect the patient may have one."

44 percent of patients said that because of a slow diagnosis, treatment was delayed and the impact on their condition has been negative.

About the Survey
This survey was conducted by Engage Health, Inc. and provided data for 920 patients in 26 countries, and 367 HCPs in 13 countries. 805 patients, parents and spouses provided evaluable data, including those with more than one child with a rare disease.

The survey was administered using a standardized questionnaire and required special security measures to ensure patient confidentiality. The survey was conducted over a period of 12 months and closed in August of 2013.

Find the article here.

Reference