SYS Patient Voices Questionnaire

In an effort to identify unmet medical needs and treatment priorities for the SYS community, FPWR has created a questionnaire for parents and caregivers to complete. The survey results, with identifying information removed, will be shared with the SYS community, and may also be shared with representatives from the US Food and Drug Administration and representatives of pharmaceutical companies and academic institutions interested in developing new drugs for SYS.

If you are the primary caregiver of a person or persons with SYS and you wish to participate in this survey, please click the button below.

If you have more than one immediate family member with SYS, you will be able to provide information about the age of the second family member and the impact SYS has for that person at the end of the survey.

* 1. What is your relationship to the person(s) with Schaff-Yang syndrome (SYS)?

- ◯ Parent
- ◯ Legal Guardian
- ◯ Sibling
- ◯ Professional Caregiver
- ◯ Other (please specify)

* 2. What is the type of mutation?

- ◯ De novo (no other family history, new mutation)
- ◯ Inherited (others in the family may have SYS)
- ◯ Don't know

If you know the exact mutation in the MAGEL2, please enter it here.

* 3. What is the age of the first immediate family member with SYS? (If you have more than one family member with SYS you will be able to enter the age of the person at the end of the survey)

- [ ] 0-11 months
- [ ] 1 year old
- [ ] 2 years old
- [ ] 3 years old
- [ ] 4 years old
- [ ] 5 years old
- [ ] 6 years old
- [ ] 7 years old
- [ ] 8 years old
- [ ] 9 years old
- [ ] 10 years old
- [ ] 11 years old
- [ ] 12 years old
- [ ] 13 years old
- [ ] 14 years old
- [ ] 15 years old
- [ ] 16 years old
- [ ] 17 years old
- [ ] 18 years old
- [ ] 19 years old
- [ ] 20 years old
- [ ] 21 years old
- [ ] 22 years old
- [ ] 23 years old
- [ ] 24 years old
- [ ] 25 years old or older

* 4. What major symptoms has the person with SYS experienced so far? (check all that apply)

- [ ] Hypotonia / Weak muscles
- [ ] Contractures
- [ ] Feeding problems / Inability to eat independently
- [ ] Developmental delay / Intellectual disability
- [ ] Autism Spectrum Disorder
- [ ] Growth Hormone Deficiency
- [ ] Mobility problems / Difficulty walking
- [ ] Hypogonadism / Incomplete sexual development
- [ ] Scoliosis
- [ ] Gastrointestinal problems / Chronic constipation
- [ ] Osteoporosis / Weak bones
- [ ] Seizures
- [ ] Sleep problems
- [ ] Breathing problems
- [ ] Excessive hunger (hyperphagia)
- [ ] Communication / Speech delays or difficulties
- [ ] Difficult behavior
- [ ] Anxiety
- [ ] Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)
- [ ] None of the above

Other (please specify)

* 5. Please rate the importance of how these issues impact the life of the person with SYS in their day-to-day living right now?

	Not Important	Somewhat Important	Very Important	Most Important
Hypotonia / Weak muscles	○	○	○	○

	Not Important	Somewhat Important	Very Important	Most Important
Contractures	○	○	○	○
Feeding problems / Inability to eat independently	○	○	○	○
Developmental delay / Intellectual disability	○	○	○	○
Autism Spectrum Disorder	○	○	○	○
Growth Hormone Deficiency	○	○	○	○
Mobility problems / Difficulty walking	○	○	○	○
Hypogonadism / Incomplete sexual development	○	○	○	○
Scoliosis	○	○	○	○
Gastrointestinal problems / Chronic constipation	○	○	○	○
Osteoporosis / Weak bones	○	○	○	○
Seizures	○	○	○	○
Sleep problems	○	○	○	○
Breathing problems	○	○	○	○
Excessive hunger (hyperphagia)	○	○	○	○
Communication / Speech delays or difficulties	○	○	○	○
Difficult behavior	○	○	○	○
Anxiety	○	○	○	○
Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)	○	○	○	○
Other (as listed above in Question #4)	○	○	○	○

* 6. How much do you worry about these issues when thinking about the future of the person with SYS?

	It is not a worry	I worry about it a little bit	I worry about it a lot	I worry about this the most
Hypotonia / Weak muscles	○	○	○	○
Contractures	○	○	○	○
Feeding problems / Inability to eat independently	○	○	○	○
Developmental delay / Intellectual disability	○	○	○	○
Autism Spectrum Disorder	○	○	○	○
Growth Hormone Deficiency	○	○	○	○
Mobility problems / Difficulty walking	○	○	○	○
Hypogonadism / Incomplete sexual development	○	○	○	○
Scoliosis	○	○	○	○
Gastrointestinal problems / Chronic constipation	○	○	○	○
Osteoporosis / Weak bones	○	○	○	○
Seizures	○	○	○	○
Sleep problems	○	○	○	○
Breathing problems	○	○	○	○
Excessive hunger (hyperphagia)	○	○	○	○
Communication / Speech delays or difficulties	○	○	○	○
Difficult behavior	○	○	○	○
Anxiety	○	○	○	○
Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)	○	○	○	○
Other (as listed above in question #4)	○	○	○	○

* 7. Overall, how would you rate the impact of SYS on the day-to-day life of person with SYS so far?

- ◯ Little or no impact
- ◯ Mild - causes occasional disruptions or manageable challenges in normal daily living
- ◯ Moderate – causes regular disruptions or occasional difficult challenges in normal daily living
- ◯ Severe – causes frequent and severe disruptions in normal daily living

* 8. Overall, how would you rate the impact of SYS on the PRIMARY CAREGIVER of the person with SYS so far?

- ◯ Little or no impact
- ◯ Mild - causes occasional disruptions or manageable challenges in the life of the primary caregiver
- ◯ Moderate – causes regular disruptions or occasional difficult challenges in the life of the primary caregiver
- ◯ Severe – causes frequent and severe disruptions in the life of the primary caregiver

* 9. Overall, how would you rate the impact of SYS on the FAMILY of the person with SYS so far?

- ◯ Little or no impact
- ◯ Mild - causes occasional disruptions or manageable challenges in the lives of family members
- ◯ Moderate – causes regular disruptions or occasional difficult challenges in the lives of family members
- ◯ Severe – causes frequent and severe disruptions in the lives of family members

* 10. How would you rate the impact of SYS on the ability of the person with SYS to reach long term goals? (education, job opportunities, independent living)

- ◯ Little or no impact
- ◯ Mild – has caused small changes to long term goals
- ◯ Moderate – has caused some changes to long term goals
- ◯ Severe – has caused major changes to long term goals

* 11. To what degree has SYS affected these aspects of the PRIMARY CAREGIVER'S life?

	Little or no impact	Mild	Moderate	Severe	Not applicable
Financial Impact	◯	◯	◯	◯	◯
Social Impact	◯	◯	◯	◯	◯
Day-to-day living	◯	◯	◯	◯	◯
Ability of the primary caregiver to work outside of the home	◯	◯	◯	◯	◯
Long term goals of the primary caregiver	◯	◯	◯	◯	◯

* 12. What treatments are you currently using to manage SYS? (Check all that apply)

- [ ] Growth Hormone
- [ ] CPAP
- [ ] G-tube or NG tube
- [ ] Scoliosis brace
- [ ] Splints, bracing or foot orthoses for contractures
- [ ] Tonsillectomy and/or adenoidectomy
- [ ] Physical therapy
- [ ] Speech therapy
- [ ] Occupational therapy
- [ ] ABA therapy
- [ ] GI medications (reflux, constipation)
- [ ] Seizure medications
- [ ] Anxiety medications
- [ ] None of the above

Other (please specify)

* 13. In thinking about all of the treatments you have used for the person with SYS (including therapies, supplements and drugs) how well do you feel these treatments able to control the symptoms of SYS?

	Treatment is not effective	Treatment helps somewhat	Treatment is very helpful	Treatment is helpful but side effects are significant	Not receiving this treatment
Growth Hormone	○	○	○	○	○
CPAP	○	○	○	○	○
G-tube or NG tube	○	○	○	○	○
Scoliosis brace	○	○	○	○	○
Splints, bracing or foot orthoses for contractures	○	○	○	○	○
Tonsillectomy and/or adenoidectomy	○	○	○	○	○
Physical therapy	○	○	○	○	○
Speech therapy	○	○	○	○	○
Occupational therapy	○	○	○	○	○
ABA therapy (Applied Behavior Analysis)	○	○	○	○	○
GI medications (reflux, constipation)	○	○	○	○	○
Seizure medications	○	○	○	○	○
Anxiety medications	○	○	○	○	○
Other (as reported in question #12)	○	○	○	○	○

* 14. Assuming there is no complete cure for SYS, what specific things would you look for in an ideal treatment for SYS?

	Not at all important	Somewhat important	Very important	Most Important
Improves bone health	○	○	○	○
Improves ability to communicate / speech improvement	○	○	○	○
Improves behavior	○	○	○	○
Improves mobility	○	○	○	○
Improves gastrointestinal health, reduces constipation	○	○	○	○
Improves positive social interaction	○	○	○	○
Improves sleep	○	○	○	○
Improves intellect / development	○	○	○	○
Improves stamina / activity	○	○	○	○

15. Are you participating in an SYS registry?

- ○ Yes, the Simons Foundation Magel2 Registry
- ○ Yes, the Global PWS Registry
- ○ Yes, both the Simons Foundation and Global PWS Registry
- ○ No, I am not participating in a registry

16. In what country do you live?

17. Feel free to provide additional comments on the impact of SYS on your loved one, on you or on your family.

18. Feel free to provide additional comments on whether current treatments are adequately addressing the symptoms of SYS.

19. Feel free to provide any additional comments about your views on what SYS research should be prioritized.

* 20. Do you have more than 1 immediate family member with SYS?

- ◯ No
- ◯ Yes

SYS Patient Voices Questionnaire

* 21. What is the age of the second immediate family member with SYS?

- [ ] 0-11 months
- [ ] 1 year old
- [ ] 2 years old
- [ ] 3 years old
- [ ] 4 years old
- [ ] 5 years old
- [ ] 6 years old
- [ ] 7 years old
- [ ] 8 years old
- [ ] 9 years old
- [ ] 10 years old
- [ ] 11 years old
- [ ] 12 years old
- [ ] 13 years old
- [ ] 14 years old
- [ ] 15 years old
- [ ] 16 years old
- [ ] 17 years old
- [ ] 18 years old
- [ ] 19 years old
- [ ] 20 years old
- [ ] 21 years old
- [ ] 22 years old
- [ ] 23 years old
- [ ] 24 years old
- [ ] 25 years old or older

* 22. What major symptoms has the second immediate family member with SYS experienced so far? (check all that apply)

- [ ] Hypotonia / Weak muscles
- [ ] Contractures
- [ ] Feeding problems / Inability to eat independently
- [ ] Developmental delay / Intellectual disability
- [ ] Autism Spectrum Disorder
- [ ] Growth Hormone Deficiency
- [ ] Mobility problems / Difficulty walking
- [ ] Hypogonadism / Incomplete sexual development
- [ ] Scoliosis
- [ ] Gastrointestinal problems / Chronic constipation
- [ ] Osteoporosis / Weak bones
- [ ] Seizures
- [ ] Sleep problems
- [ ] Breathing problems
- [ ] Excessive hunger (hyperphagia)
- [ ] Communication / Speech delays or difficulties
- [ ] Difficult behavior
- [ ] Anxiety
- [ ] Eye problems (myopia, esotropia, strabismus)
- [ ] None of the above

Other (please specify)

* 23. Please rate the importance of how these issues impact the life of the second immediate family member with SYS in their day-to-day living right now?

	Not Important	Somewhat Important	Very Important	Most Important
Hypotonia / Weak muscles	○	○	○	○
Contractures	○	○	○	○
Feeding problems / Inability to eat independently	○	○	○	○
Developmental delay / Intellectual disability	○	○	○	○
Autism Spectrum Disorder	○	○	○	○
Growth Hormone Deficiency	○	○	○	○
Mobility problems / Difficulty walking	○	○	○	○
Hypogonadism / Incomplete sexual development	○	○	○	○
Scoliosis	○	○	○	○
Gastrointestinal problems / Chronic constipation	○	○	○	○
Osteoporosis / Weak bones	○	○	○	○
Seizures	○	○	○	○
Sleep problems	○	○	○	○
Breathing problems	○	○	○	○
Excessive hunger (hyperphagia)	○	○	○	○
Communication / Speech delays or difficulties	○	○	○	○
Difficult behavior	○	○	○	○
Anxiety	○	○	○	○
Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)	○	○	○	○
Other (as listed above in question #20)	○	○	○	○

* 24. How much do you worry about these issues when thinking about the future of the second immediate

family member with SYS?

	It is not a worry	I worry about it a little bit	I worry about it a lot	I worry about this the most
Hypotonia / Weak muscles	○	○	○	○
Contractures	○	○	○	○
Feeding problems / Inability to eat independently	○	○	○	○
Developmental delay / Intellectual disability	○	○	○	○
Autism Spectrum Disorder	○	○	○	○
Growth Hormone Deficiency	○	○	○	○
Mobility problems / Difficulty walking	○	○	○	○
Hypogonadism / Incomplete sexual development	○	○	○	○
Scoliosis	○	○	○	○
Gastrointestinal problems / Chronic constipation	○	○	○	○
Osteoporosis / Weak bones	○	○	○	○
Seizures	○	○	○	○
Sleep problems	○	○	○	○
Breathing problems	○	○	○	○
Excessive hunger (hyperphagia)	○	○	○	○
Communication / Speech delays or difficulties	○	○	○	○
Difficult behavior	○	○	○	○
Anxiety	○	○	○	○
Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)	○	○	○	○

	It is not a worry	I worry about it a little bit	I worry about it a lot	I worry about this the most
Other (as listed above in question #20)	○	○	○	○

* 25. Overall, how would you rate the impact of SYS on the day-to-day life of second immediate family member with SYS so far?

- ○ Little or no impact
- ○ Mild - causes occasional disruptions or manageable challenges in normal daily living
- ○ Moderate – causes regular disruptions or occasional difficult challenges in normal daily living
- ○ Severe – causes frequent and severe disruptions in normal daily living

* 26. How would you rate the impact SYS has on the second immediate family member with SYS to reach long term goals? (education, job opportunities, independent living)

- ○ Little or no impact
- ○ Mild – has caused small changes to long term goals
- ○ Moderate – has caused some changes to long term goals
- ○ Severe – has caused major changes to long term goals

* 27. What treatments are you currently using to manage SYS for the second immediate family member? (Check all that apply)

- ☐ Growth Hormone
- ☐ CPAP
- ☐ G-tube or NG tube
- ☐ Scoliosis brace
- ☐ Splints, bracing or foot orthoses for contractures
- ☐ Tonsillectomy and/or adenoidectomy
- ☐ Physical therapy
- ☐ Speech therapy
- ☐ Occupational therapy
- ☐ ABA therapy
- ☐ GI medications (reflux, constipation)
- ☐ Seizure medications
- ☐ Anxiety medications
- ☐ None of the above

Other (please specify)

* 28. In thinking about all of the treatments you have used for the second immediate family member with SYS (including therapies, supplements and drugs) how well do you feel these treatments able to control the symptoms of SYS?

	Treatment is not effective	Treatment helps somewhat	Treatment is very helpful	Treatment is helpful but side effects are significant	Not receiving this treatment
Growth Hormone	○	○	○	○	○
CPAP	○	○	○	○	○
G-tube or NG tube	○	○	○	○	○
Scoliosis brace	○	○	○	○	○
Splints, bracing or foot orthoses for contractures	○	○	○	○	○
Tonsillectomy and/or adenoidectomy	○	○	○	○	○
Physical therapy	○	○	○	○	○
Speech therapy	○	○	○	○	○
Occupational therapy	○	○	○	○	○
ABA therapy (Applied Behavior Analysis)	○	○	○	○	○
GI medications (reflux, constipation)	○	○	○	○	○
Seizure medications	○	○	○	○	○
Anxiety medications	○	○	○	○	○
Other (as reported in question #24)	○	○	○	○	○