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)
Oon't know
If you know the exact mutation in the MAGEL2, please enter it here.

# O AMb at in the case of the first income distant family one	
* 3. What is the age of the first immediate family me	
member with SYS you will be able to enter the age of	<u> </u>
0-11 months	13 years old
1 year old	14 years old
2 years old	15 years old
3 years old	16 years old
4 years old	17 years old
5 years old	18 years old
6 years old	19 years old
7 years old	20 years old
8 years old	21 years old
9 years old	22 years old
10 years old	23 years old
11 years old	24 years old
12 years old	25 years old or older
Contractures Feeding problems / Inability to eat independently	Seizures Sleep problems
Feeding problems / Inability to eat independently	Sleep problems
Developmental delay / Intellectual disability	Breathing problems
Autism Spectrum Disorder	Excessive hunger (hyperphagia)
Growth Hormone Deficiency	Communication / Speech delays or difficulties
Mobility problems / Difficulty walking	Difficult behavior
Hypogonadism / Incomplete sexual development	Anxiety
Scoliosis	Eye problems (ex. myopia, esotropia, strabismus, pto
Gastrointestinal problems / Chronic constipation	cortical vision impairment)
	None of the above
Other (please specify)	
5. Please rate the importance of how these issues im	pact the life of the person with SYS in their dav-to-d
ving right now?	
Not Important Somewhat	Important Very Important Most Important
Hypotonia / Weak	
muscles	

	Not Important	Somewhat Important	Very Important	Most Important
Contractures	\bigcirc	\bigcirc	\bigcirc	
Feeding problems / Inability to eat independently	0	0	0	0
Developmental delay / Intellectual disability	\bigcirc	\bigcirc	\bigcirc	
Autism Spectrum Disorder	\circ	0	0	0
Growth Hormone Deficiency	\circ	\circ	\circ	\circ
Mobility problems / Difficulty walking	\circ	0	\circ	0
Hypogonadism / Incomplete sexual development	\bigcirc	\bigcirc	\bigcirc	\circ
Scoliosis		\circ		
Gastrointestinal problems / Chronic constipation	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Osteoporosis / Weak bones	\circ	0	\circ	\circ
Seizures				
Sleep problems			\bigcirc	
Breathing problems		\bigcirc	\bigcirc	
Excessive hunger (hyperphagia)	\circ	\bigcirc	\circ	
Communication / Speech delays or difficulties	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Difficult behavior				
Anxiety				
Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)				
Other (as listed above in Question #4)	\bigcirc	\bigcirc		

	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	\circ	0	\bigcirc	
Contractures		\bigcirc	\bigcirc	
Feeding problems / Inability to eat independently	0		0	
Developmental delay / Intellectual disability	\bigcirc		\bigcirc	
Autism Spectrum Disorder			\bigcirc	
Growth Hormone Deficiency	\bigcirc		\bigcirc	\bigcirc
Mobility problems / Difficulty walking			\bigcirc	
Hypogonadism / Incomplete sexual development	\bigcirc		\circ	\bigcirc
Scoliosis		\bigcirc	\bigcirc	
Gastrointestinal problems / Chronic constipation	\bigcirc		\circ	\bigcirc
Osteoporosis / Weak bones	\circ		\bigcirc	0
Seizures			\bigcirc	
Sleep problems				
Breathing problems		\bigcirc		
Excessive hunger (hyperphagia)	\circ	0	0	
Communication / Speech delays or difficulties	\bigcirc		\bigcirc	\bigcirc
Difficult behavior		\bigcirc		
Anxiety				
Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)	0		0	0
Other (as listed above in question #4)	\circ		0	

	+				
Little or no impac	casional disruptions or m	anagaahla challa	nges in normal daily livi	na	
	es regular disruptions or				
			•	ually living	
Severe – causes	frequent and severe dis	ruptions in norma	i daliy living		
* 8. Overall, how wo	ould you rate the im	pact of SYS o	n the PRIMARY CA	REGIVER of th	e person with SYS
Little or no impac	rt.				
Mild - causes occ	casional disruptions or m	anageable challe	nges in the life of the pr	imary caregiver	
Moderate – cause	es regular disruptions or	occasional difficu	ılt challenges in the life	of the primary carec	giver
Severe – causes	frequent and severe dis	ruptions in the life	of the primary caregive	er	
* 9. Overall, how wo	•	pact of SYS o	n the FAMILY of the	e person with SY	/S so far?
Mild - causes occ	casional disruptions or m	anageable challe	nges in the lives of fami	ly members	
Moderate – cause	es regular disruptions or	occasional difficu	ılt challenges in the live	s of family members	S
	frequent and severe dis		-	•	
Little or no impac	t				
Moderate – has c	d small changes to long caused some changes to used major changes to lo	o long term goals	the PRIMARY CAF	REGIVER'S life?	
Moderate – has c	caused some changes to loused major changes to louse the louse SYS affected the	o long term goals ong term goals ese aspects of			
Moderate – has c	caused some changes to	o long term goals	the PRIMARY CAR	REGIVER'S life? Severe	Not applicable
Moderate – has cau Severe – has cau 11. To what degree h	caused some changes to loused major changes to louse the louse SYS affected the	o long term goals ong term goals ese aspects of			
Moderate – has cau Severe – has cau 11. To what degree h Financial Impact	caused some changes to loused major changes to louse the louse SYS affected the	o long term goals ong term goals ese aspects of			
Moderate – has cau Severe – has cau 11. To what degree h Financial Impact Social Impact	caused some changes to loused major changes to louse the louse SYS affected the	o long term goals ong term goals ese aspects of			

	-	itly using to mana	ige SYS? (Check		
Growth Hormone			Speech thera	ру	
СРАР			Occupational	therapy	
G-tube or NG tub	e		ABA therapy		
Scoliosis brace			GI medication	ns (reflux, constipation))
Splints, bracing o	or foot orthoses for co	ntractures	Seizure medi	cations	
Tonsillectomy and	d/or adenoidectomy		Anxiety medic	cations	
Physical therapy			None of the a	bove	
Other (please specify) 13. In thinking about	all of the treatme	nts you have used	d for the person w	rith SYS (including	therapies,
upplements and drug		-		-	
Growth Hormone	ellective	Somewhat	Пеіріці	Significant	treatment
CPAP	0		0		
G-tube or NG tube	0				
Scoliosis brace			0	0	
Scoliosis brace Splints, bracing or foot orthoses for contractures	0	0	0	0	0
Splints, bracing or foot orthoses for contractures Tonsillectomy and/or	OOO	0	0	0	0
Splints, bracing or foot	OOOO	OOOO		0	OOOO
Splints, bracing or foot orthoses for contractures Tonsillectomy and/or adenoidectomy					OOOOO
Splints, bracing or foot orthoses for contractures Tonsillectomy and/or adenoidectomy Physical therapy					
Splints, bracing or foot orthoses for contractures Tonsillectomy and/or adenoidectomy Physical therapy Speech therapy					
Splints, bracing or foot orthoses for contractures Tonsillectomy and/or adenoidectomy Physical therapy Speech therapy Occupational therapy ABA therapy (Applied					
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,					
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)					

eatment for SYS?				
	Not at all important	Somewhat important	Very important	Most Important
mproves bone health	\circ		\bigcirc	\circ
mproves ability to communicate / speech mprovement	\circ	\bigcirc	\bigcirc	\circ
mproves behavior			\bigcirc	
mproves mobility				
mproves gastrointestinal health, reduces constipation	0	0	0	0
mproves positive social nteraction	\bigcirc	\bigcirc	\bigcirc	\bigcirc
mproves sleep				
mproves intellect / development	\bigcirc	\bigcirc	\bigcirc	\bigcirc
mproves stamina / activity		\circ	\bigcirc	\bigcirc
_	nons Foundation and Glob	al PWS Registry		
16. In what country	do you live?			
•	e additional commen	s on the impact of SYS	on your loved one, o	n you or on your
y.				
8. Feel free to provide ymptoms of SYS.	e additional comment	s on whether current tre	eatments are adequa	

oritized.					
* 20. Do you h	ave more than 1 im	madiata famil	memher with	SVS2	
	ave more man i m	imediale family	member with	313:	
O No					
Yes					

0.11 months	illy member with SYS?
0-11 months	13 years old
1 year old	14 years old
2 years old	15 years old
3 years old	16 years old
4 years old	17 years old
5 years old	18 years old
6 years old	19 years old
7 years old	20 years old
8 years old	21 years old
9 years old	22 years old
10 years old	23 years old
11 years old	24 years old
12 years old	25 years old or older
i inai annivi	
Hypotonia / Weak muscles	Osteoporosis / Weak bones
	Osteoporosis / Weak bones Seizures
Hypotonia / Weak muscles	
Hypotonia / Weak muscles Contractures	Seizures
Hypotonia / Weak muscles Contractures Feeding problems / Inability to eat independently	Seizures Sleep problems
Hypotonia / Weak muscles Contractures Feeding problems / Inability to eat independently Developmental delay / Intellectual disability	Seizures Sleep problems Breathing problems
Hypotonia / Weak muscles Contractures Feeding problems / Inability to eat independently Developmental delay / Intellectual disability Autism Spectrum Disorder	Seizures Sleep problems Breathing problems Excessive hunger (hyperphagia)
Hypotonia / Weak muscles Contractures Feeding problems / Inability to eat independently Developmental delay / Intellectual disability Autism Spectrum Disorder Growth Hormone Deficiency	Seizures Sleep problems Breathing problems Excessive hunger (hyperphagia) Communication / Speech delays or difficulties
Contractures Feeding problems / Inability to eat independently Developmental delay / Intellectual disability Autism Spectrum Disorder Growth Hormone Deficiency Mobility problems / Difficulty walking	Seizures Sleep problems Breathing problems Excessive hunger (hyperphagia) Communication / Speech delays or difficulties Difficult behavior
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	Seizures Sleep problems Breathing problems Excessive hunger (hyperphagia) Communication / Speech delays or difficulties Difficult behavior Anxiety
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	Seizures Sleep problems Breathing problems Excessive hunger (hyperphagia) Communication / Speech delays or difficulties Difficult behavior Anxiety Eye problems (myopia, esotropia, strabismus)

	Not Important	Somewhat Important	Very Important	Most Important
Hypotonia / Weak muscles		0	0	0
Contractures				
Feeding problems / Inability to eat independently	0	0	0	0
Developmental delay / Intellectual disability	\bigcirc	\bigcirc		\bigcirc
Autism Spectrum Disorder		\circ		\circ
Growth Hormone Deficiency	\bigcirc	\bigcirc	\bigcirc	\bigcirc
Mobility problems / Difficulty walking	\circ	\circ	\circ	\circ
Hypogonadism / Incomplete sexual development	\bigcirc			\bigcirc
Scoliosis				
Gastrointestinal problems / Chronic constipation	\bigcirc	\bigcirc	\circ	\circ
Osteoporosis / Weak bones	0	0	0	0
Seizures				
Sleep problems				
Breathing problems				
Excessive hunger (hyperphagia)		\circ		\circ
Communication / Speech delays or difficulties	\bigcirc		\bigcirc	\bigcirc
Difficult behavior				
Anxiety				\bigcirc
Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)				0
Other (as listed above in question #20)		\bigcirc	\bigcirc	\bigcirc

family member with SYS	S?			
	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	0	0	0	0
Contractures	\bigcirc	\circ	\bigcirc	\circ
Feeding problems / Inability to eat independently	0	0	0	
Developmental delay / Intellectual disability	\bigcirc	\bigcirc	\bigcirc	
Autism Spectrum Disorder	0	\circ	\circ	
Growth Hormone Deficiency	\bigcirc	\bigcirc	\bigcirc	
Mobility problems / Difficulty walking	\circ	0	\circ	
Hypogonadism / Incomplete sexual development	\bigcirc		\bigcirc	
Scoliosis		\circ		
Gastrointestinal problems / Chronic constipation	\circ	\circ	\circ	
Osteoporosis / Weak bones	0	0	0	0
Seizures	\bigcirc	\bigcirc		
Sleep problems				
Breathing problems				
Excessive hunger (hyperphagia)	\bigcirc	0	\circ	
Communication / Speech delays or difficulties	\bigcirc		\circ	
Difficult behavior		\bigcirc		
Anxiety				
Eye problems (ex. myopia, esotropia, strabismus, ptosis, cortical vision impairment)			0	

	It is not a worry	I worry about it a little bit	I worry about it a lot	I worry about this the most
ther (as listed above in uestion #20)	\bigcirc	\circ	\circ	\circ
* 25. Overall, how wo member with SYS so		mpact of SYS on the day	to-day life of second	immediate family
Little or no impact				
Mild - causes occas	sional disruptions or m	nanageable challenges in norm	al daily living	
Moderate – causes	regular disruptions or	occasional difficult challenges	in normal daily living	
Severe – causes fre	equent and severe dis	ruptions in normal daily living		
•	•	YS has on the second im tunities, independent livir	•	per with SYS to reach
Little or no impact				
Mild – has caused s	small changes to long	term goals		
Moderate – has cau	used some changes to	o long term goals		
Severe – has cause	ed major changes to lo	ong term goals		
(Check all that apply) Growth Hormone)	Spe	eech therapy	
СРАР		Occ	cupational therapy	
G-tube or NG tube		ABA	A therapy	
Scoliosis brace		GIr	medications (reflux, consti	pation)
Splints, bracing or f	foot orthoses for contra	actures Seiz	zure medications	
Tonsillectomy and/o	or adenoidectomy	Anx	iety medications	
Physical therapy		Nor	ne of the above	
Other (please specify)				
1				

PAP		Treatment is not effective	Treatment helps somewhat	Treatment is very helpful	Treatment is helpful but side effects are significant	Not receiving this treatment
-tube or NG tube coliosis brace plints, bracing or foot thoses for contractures consillectomy and/or denoidectomy hysical therapy peech therapy coupational therapy BA therapy (Applied ehavior Analysis) I medications (reflux, constipation) eizure medications nxiety medications ther (as reported in	rowth Hormone					
coliosis brace	PAP					
plints, bracing or foot rthoses for contractures consillectomy and/or denoidectomy hysical therapy peech therapy peech therapy BA therapy (Applied ehavior Analysis) Il medications (reflux, constipation) eizure medications nxiety medications ther (as reported in	i-tube or NG tube					
Inthoses for contractures Instillectomy and/or denoidectomy Industrial therapy Institute therapy Institu	coliosis brace					
thysical therapy peech therapy peech therapy pecupational therapy BA therapy (Applied ehavior Analysis) If medications (reflux, constipation) eizure medications nxiety medications ther (as reported in	plints, bracing or foot rthoses for contractures	0	0	\circ	0	
peech therapy Cocupational therapy BA therapy (Applied ehavior Analysis) Il medications (reflux, onstipation) eizure medications nxiety medications ther (as reported in	onsillectomy and/or denoidectomy	\bigcirc		\bigcirc	\bigcirc	\bigcirc
BA therapy (Applied ehavior Analysis) If medications (reflux, onstipation) eizure medications nxiety medications ther (as reported in	hysical therapy					
BA therapy (Applied ehavior Analysis) If medications (reflux, onstipation) eizure medications nxiety medications other (as reported in	peech therapy			\bigcirc		
ehavior Analysis) If medications (reflux, onstipation) eizure medications nxiety medications other (as reported in	ccupational therapy					
eizure medications nxiety medications other (as reported in	BA therapy (Applied ehavior Analysis)	\bigcirc	\circ	\circ	\circ	\bigcirc
nxiety medications O O O O O O O O O O O O O O O O O O O	I medications (reflux, onstipation)	0	0	\circ	0	0
other (as reported in	eizure medications			\bigcirc	\circ	\circ
	nxiety medications					
	other (as reported in uestion #24)					