

# 2025 SETBP1 Community Survey



77 # of SETBP1 families that completed the entire survey  
 8 # of SETBP1 families that completed most of the survey  
 85 **Total number of SETBP1 families included in these results**

## S1. All Individuals

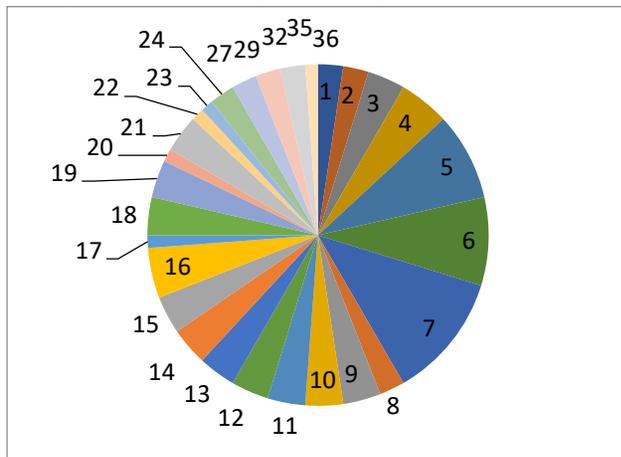
Q1 Who is completing this survey?

Role	#	%
<i>mother</i>	70	82%
father	8	9%
grandmother	2	2%
parent	5	6%
<b>Total</b>	<b>85</b>	<b>100%</b>

Q2 Is your child/young adult less than 17 years old?

	#	%
Yes	62	73%
No	23	27%
Total	85	

Q3 Enter your child/young adult's age.



Q4 Child's Age at SETBP1 Diagnosis

Age	#	%
3 - 6 months	1	1%
7 - 8 months	1	1%
9 - 12 months	3	4%
13 - 23 months	5	6%
2 years old	6	7%
3 years old	4	5%
4 years old	9	11%
5 years old	5	6%
6 years old	7	9%
7 years old	5	6%
8 years old	4	5%
9 years old	6	7%
10 years old	3	4%
11 years old	2	2%
12 years old	3	4%
13 years old	2	2%
14 - 18 years old	5	6%
19 - 25 years old	5	6%
25 + years old	6	7%
<b>Total</b>	<b>82</b>	<b>100%</b>
<b>Average</b>	<b>9 years old</b>	
<b>Median</b>	<b>6 years old</b>	

Q5 Which SETBP1 neurodevelopmental disorder does your child have?

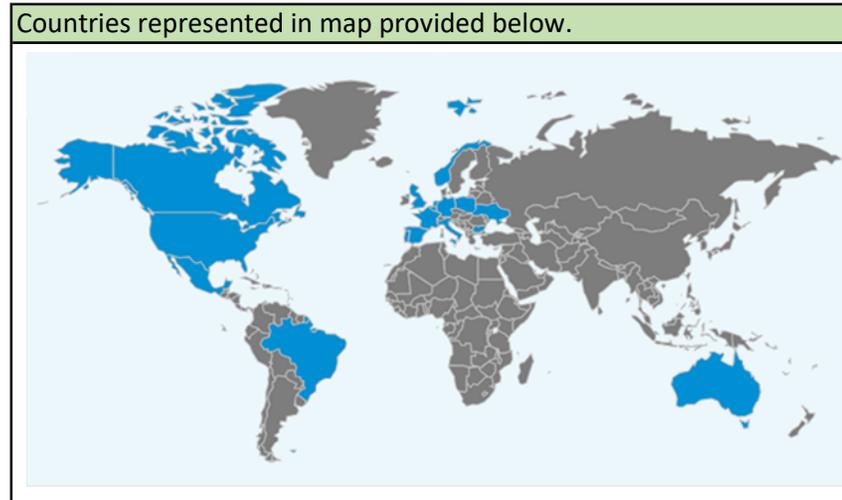
Type	#	%
SETBP1-HD ( <i>loss-of-function variant or deletion</i> )	59	69%
SETBP1 related disorders (missense variant)	17	20%
Unsure	4	5%
18q12.3 Deletion (large deletion including SETBP1 and other genes)	5	6%
Total	85	100%

Q6 Please select which best describes your child/young adult.

	#	%
Female	38	45%
Male	47	55%

Q7 Select your country.

Country	#	%
United States	40	47%
Australia	2	2%
Belgium	1	1%
Bolivia	1	1%
Brazil	1	1%
Bulgaria	1	1%
Canada	1	1%
France	6	7%
Germany	4	5%
Italy	1	1%
Mexico	1	1%
Netherlands	10	12%
Norway	1	1%
Poland	1	1%
Portugal	2	2%
Puerto Rico	1	1%
Spain	2	2%
United Kingdom	9	11%
Total	85	100%



Q8 Let's begin by talking about your child's development to get a sense of where they are at. Please write the age when your child accomplished the following, enter an X for Don't Remember or select Not Yet:

note: question was only asked from caregivers with children less than 17 years of age

	Sat without support	Walked without holding on	Used single words other than "mama" or "dada"	Used two words/short phrases*	Spoke in sentences*	Was toilet trained during the day*	Was toilet trained at night*
<b>Skill acquired</b>							
Average age	10 months	22 months	31 months	47 months	61 months	54 months	55 months
Current age range	5 months -2 years	1 -5 years	9 months -6 years	1 -9 years	2 -10 years	1 -8 years	2 -8 years
<b>Total Skill Acquired</b>	54	55	45	40	30	41	32
<b>Skill not yet acquired</b>							
Current age range	1 -1 years	<1-1 years	1 -9 years	2 -15 years	2 -15 years	1 -14 years	2 -14 years
<b>Total Skill Not Acquired</b>	2	3	11	19	29	20	26
<b>Total Responses</b>	56	58	56	59	59	61	58

\* children 1 years and younger were not included in the non-skill acquisition totals

Q9 What is your child/young adult's current level of Language:

	#	%
non-verbal	11	16%
minimally verbal (very limited spoken words, often inconsistently)	8	12%
speaks in one-two word phrases or sentences	18	26%
speaks in simple sentences	25	36%
speaks in long, complex sentences	18	26%
Other	1	1%
<b>Total</b>	<b>81</b>	

Please select any aides your child/young adult uses regularly to help them communicate?

	#
Augmentative and Alternative Communication Device - Non-Electronic (i.e., Picture Exchange Communication System, Communication Board, Alphabet Boards)	2
Augmentative and Alternative Communication Device - Electronic (i.e., TouchChat, Proloquo2Go, Dedicated Speech-Generating Devices)	6
Sign Language	12
Makaton (or similar sign/symbol language)	2
<b>Total</b>	<b>22</b>



Q11 Let’s talk about behaviors causing concern to you or other people. Has your child/young adult ever had difficulty with any of the following?

Statement	All Ages			Current for Specific Age Groups		
	No	Currently	Previously	< 5 years	School-Age	> 17 years
Anxiety	29	48	8	4	26	18
	34%	56%	9%	36%	51%	78%
Depressed mood	69	13	3	3	7	3
	81%	15%	4%	27%	14%	13%
Mood Swings	28	50	7	6	28	16
	33%	59%	8%	55%	55%	70%
Aggressive outbursts	30	42	13	5	26	11
	35%	49%	15%	45%	51%	48%
Temper Tantrums	22	47	16	8	23	11
	26%	55%	19%	73%	45%	48%
Self-injury, such as hitting self, biting self, scratching self	45	21	19	1	34	7
	53%	25%	22%	9%	67%	30%
Absent or delayed onset of language to communicate	6	43	36	10	29	3
	7%	51%	42%	91%	57%	13%
Repeating words or phrases over and over again	28	46	11	5	24	13
	33%	54%	13%	45%	47%	57%
Difficulties getting on with other people of similar age	37	45	3	3	25	14
	44%	53%	4%	27%	49%	61%
Immature development of social skills	16	65	4	4	20	18
	19%	76%	5%	36%	39%	78%
Repetitive behaviors, such as doing the same thing over and over again	30	46	9	6	24	16
	35%	54%	11%	55%	47%	70%
Very rigid or inflexible about how to do things or not liking change in routines	29	49	7	3	26	20
	34%	58%	8%	27%	51%	87%
Overactivity/hyperactivity, such as being constantly on the go	25	46	14	6	31	9
	29%	54%	16%	55%	61%	39%
Difficulty paying attention or concentrating	1	83	1	10	50	23
	1%	98%	1%	91%	98%	100%
Restlessness or fidgetiness, such as wriggling or squirming	24	51	10	7	34	10
	28%	60%	12%	64%	67%	43%
Impulsivity, such as butting in, not waiting turn	13	64	8	7	42	15
	15%	75%	9%	64%	82%	65%
Sleep difficulties, such as with falling asleep or waking	43	30	12	6	15	9
	51%	35%	14%	55%	29%	39%
Total Responses	85 (100%)			11	51	23



Q11 Let's talk about behaviors causing concern to you or other people. Has your child/young adult ever had difficulty with any of the following?

(Continued->)

Statement	All Ages		
	No	Currently	Previously
Difficulties with eating, such as eating too much, too little, or swallowing difficulties (before age 1)	37	21	27
	44%	25%	32%
Difficulties with eating, such as eating too much, too little, unusual things, or swallowing difficulties (after age 1)	37	34	14
	44%	40%	16%
Difficulties with stuffing too much food in their mouth (after age 1)	39	35	11
	46%	41%	13%
<b>Total Responses</b>	<b>85 (100%)</b>		
Loss of speech skills (after age 5)	24	1	0
	96%	4%	0%
Loss of motor skills (after age 5)	24	1	0
	96%	4%	0%
Hallucinations (seeing things that are not there)	24	1	0
	96%	4%	0%
<b>Total Responses</b>	<b>48 (100%)</b>		



Q12 Let's talk about conditions causing concern to you or to other people. Has your child/young adult ever had any of the following?

Statement	All Ages			Current for Specific Age Groups		
	No	Currently	Previously	< 5 years	School-Age	> 17 years
Blood Disorder	80	1	1	0	0	1
	98%	1%	1%	0%	0%	4%
Bone Disorder	75	7	0	1	2	4
	91%	9%	0%	9%	4%	17%
Congenital Disorder	59	21	2	2	10	9
	72%	26%	2%	18%	21%	39%
Epilepsy/Seizures (other than febrile)	68	10	4	3	5	2
	83%	12%	5%	27%	10%	9%
Febrile Seizures	60	6	16	2	3	1
	73%	7%	20%	18%	6%	4%
<b>Total Responses</b>	<b>82 (100%)</b>			<b>11</b>	<b>48</b>	<b>23</b>



Q12 Let's talk about conditions causing concern to you or to other people. Has your child/young adult ever had any of the following?

(Continued->)

Statement	All Ages			Current for Specific Age Groups		
	No	Currently	Previously	< 5 years	School-Age	> 17 years
Gait/Movement Disorder	23	38	21	4	9	10
	28%	46%	26%	36%	19%	43%
Gastrointestinal Problems	42	20	20	0	12	8
	51%	24%	24%	0%	25%	35%
Hearing Issues	74	7	1	2	3	2
	90%	9%	1%	18%	6%	9%
Hypotonia (low muscle tone)	23	38	21	7	22	9
	28%	46%	26%	64%	46%	39%
Hypertonia (high muscle tone)	76	3	3	1	1	1
	93%	4%	4%	9%	2%	4%
Immune Problems	66	9	7	2	5	2
	80%	11%	9%	18%	10%	9%
Mitochondrial Disorder	78	3	1	0	1	2
	95%	4%	1%	0%	2%	9%
Skin Problems (other than acne)	53	21	8	3	10	8
	65%	26%	10%	27%	21%	35%
Sensory Processing Disorder	30	41	11	4	24	13
	37%	50%	13%	36%	50%	57%
Speech and/or Language Disorder	6	68	8	10	40	18
	7%	83%	10%	91%	83%	78%
Total Responses	82 (100%)			11	48	23
Vision Problems (corrective eyewear and/or strabismus surgery fixed the issue)	35	35	9	3	20	12
	44%	44%	11%	30%	43%	55%
Vision Problems (not corrected with corrective eyewear and/or strabismus surgery)	66	9	4	1	5	3
	84%	11%	5%	10%	11%	14%
Total Responses	79 (100%)			10	47	22



Q13 Problem behaviors may add up to meet criteria for specific psychiatric disorders. Has your child/young adult ever received a diagnosis of:

Statement	< 5 years	School-Age	> 17 years	All Ages	SETBP1-RD
Attention Deficit Hyperactivity Disorder	1 9%	29 60%	9 41%	39 48%	6 38%
Autism Spectrum Disorder (ASD), including Anxiety Disorder, including as panic, phobia, separation anxiety disorder	1 9%	16 33%	9 41%	26 32%	9 56%
Bipolar	0 0%	0 0%	2 9%	2 2%	2 13%
Obsessive Compulsive Disorder (OCD)	0 0%	6 13%	4 18%	10 4%	0 0%
Oppositional Defiant Disorder (ODD)	0 0%	6 13%	2 9%	8 12%	0 0%
Depressive Disorder	0 0%	1 2%	2 9%	3 10%	1 6%
Psychotic Disorder, including schizophrenia	0 0%	2 4%	0 0%	2 2%	1 6%

Q14 IQ Testing Results

	Number	Percent	SETBP1-RD
Normal Intellectual	3	7%	0 \ 0%
Borderline Intellectual	9	20%	4 \ 40%
Mild Intellectual Disability (IQ 50-69)	14	<b>31%</b>	3 \ 30%
Moderate Intellectual	10	22%	1 \ 10%
Severe Intellectual	6	13%	1 \ 10%
Profound Intellectual Disability (IQ <20)	2	4%	1 \ 10%
Developmental Delay	1	2%	0 \ 0%
<b>Total</b>	<b>45</b>		

School-age children and older who do not have trouble Q15 in the following areas.

Statement	Number
Reading	9
	13%
Writing	9
	13%
Spelling	10
	14%
Mathematics	1
	1%
<b>Total Responses</b>	<b>72</b>



Q16 Many individuals with SETBP1-HD and related disorders will have some difficulties in some specific brain skills. Does your child/young adult have difficulty with any of the following:

Statement	All Ages		School-Age	> 17 years
	No	Yes	Yes	Yes
Memory, such as remembering things that have happened	45 65%	24 35%	8 40%	8 38%
Attention, such as concentrating well, not getting distracted	3 4%	66 96%	21 95%	21 94%
Dual-tasking/ Multi-tasking, such as doing 2 tasks at the same time	6 9%	63 91%	20 95%	20 88%
Visuo-spatial tasks, such as solving puzzles or using building blocks	24 35%	45 65%	16 63%	16 63%
Executive skills, such as planning, organizing, flexible thinking	5 7%	64 93%	20 95%	20 88%
Getting disoriented, such as not knowing the date or where you are	36 52%	33 48%	9 55%	9 25%
Total Responses	69		47	22



Q17 Of all the concerns listed in this survey, what are your top 2-3 priorities to work on next?



Q18

Taking together all the difficulties discussed above, how much have these bothered, troubled or distressed you/your child or young adult/family?

Statement	Not at all/0	1	2	3	4	5	6	7	8	9	10/Extremely
All Ages	3	2	2	3	6	6	1	4	15	13	11
	5%	3%	3%	5%	9%	9%	2%	6%	23%	20%	17%
< 5 years	2	0	0	0	1	0	0	0	0	0	0
	67%	0%	0%	0%	33%	0%	0%	0%	0%	0%	0%
School-Age	2	1	3	2	5	2	0	3	9	9	6
	5%	2%	7%	5%	12%	5%	0%	7%	21%	21%	14%
> 17 years	1	1	0	1	0	0	1	1	6	4	5
	5%	5%	0%	5%	0%	0%	5%	5%	30%	20%	25%



**S2. Young Adult Section**

Which symptoms are most impactful for your young adult

Q19 currently? Top 5 provided below.

Statement	Yes	Percent
Anxiety	10	43%
Learning Deficits	8	35%
Social Understanding and	7	30%
Aggressive, Self-Injurious Behavior, and/or Explosive Behavior	7	30%
Attention Issues	6	26%

Has your young adult had any new diagnoses after the

Q20 age of 16?

Number	Answer
17	no
1	tethered spinal cord and polycystic ovarian
1	osteoporosis gait ataxia
1	urine retention
1	autism & bipolar (SETBP1-RD)

Q21 Has your young adult had any surgical procedures after the age of 16? If yes, please provide details for each surgery.

Number	Answer
14	no
4	dental / oral
2	eye (ptosis, cataract removed)
1	appendicostomy & appendicovesicostomy
1	gall bladder removal
1	forum magnum decompression for chiari malformation & detethering of spinal cord



Does your young adult work? If yes, please provide the jobs your

Q22 young adult has held after the age of 16.

Number	Answer
13	no
8	yes (carpentry, lawn care, county council disability adviser, maintenance, disability workshop assistant, restaurant server -6 years, inclusive employer, janitorial work)

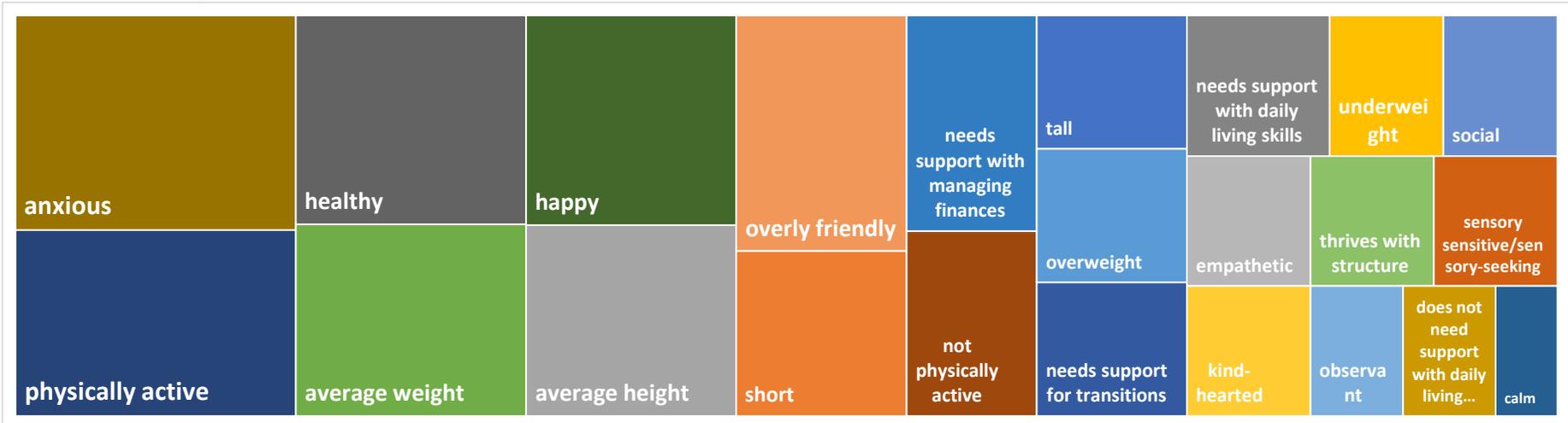
Has your young adult always lived with you? If no, please provide the type of living arrangements your young adult has lived in after the age of 16. If yes,

Q23 please enter yes.

Number	Answer
15	yes
5	no (residential placement or supportive/assisted living)

Please select all description words that apply currently to your young adult with a SETBP1 difference? Selections selected by more than 1

Q24 individual are provided below.



Q25 What is your biggest concern for your young adult with a SETBP1 difference?

Number	Answer
6	Independence
4	Concern for future after parent is not around
3	Overall well-being
2	Residential Placement
1	Friends
1	Future in general
1	Behavior
1	Deteriorating health



### S3. Other Data

Q26 Concerns from parents with SETBP1-HD/RD children/adults

"I am worried about helping him achieve his maximum independent functioning so that he may have a life of choice and purpose"

"I am unsure of what my expectations should be as he grows into adulthood. Will he be able to live independently? Will his issues worsen with age?"

"Worried about what life will look like when she's grown up"

"I wonder if he has Childhood Apraxia of Speech."

"Lonliness"

"What happens when my husband and I either die or can no longer take care of her?"

"I worry as she gets older and realizes she can't do the same things her peers are doing (driving, dating, working) with how that will affect her confidence and behavior."

Q27 What is working for your SETBP1-HD/RD children/adults?

Inclusive sports participation

Connecting with peers

Q28 Please add a few thoughts or words about what you love about your child/young adult or makes your child/young adult shine:



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