

2024 SETBP1 Community Survey Results



61 # of SETBP1 families that completed the entire survey
 7 # of SETBP1 families that completed most of the survey
 68 **Total number of SETBP1 families included in these results**

S1. All Individuals

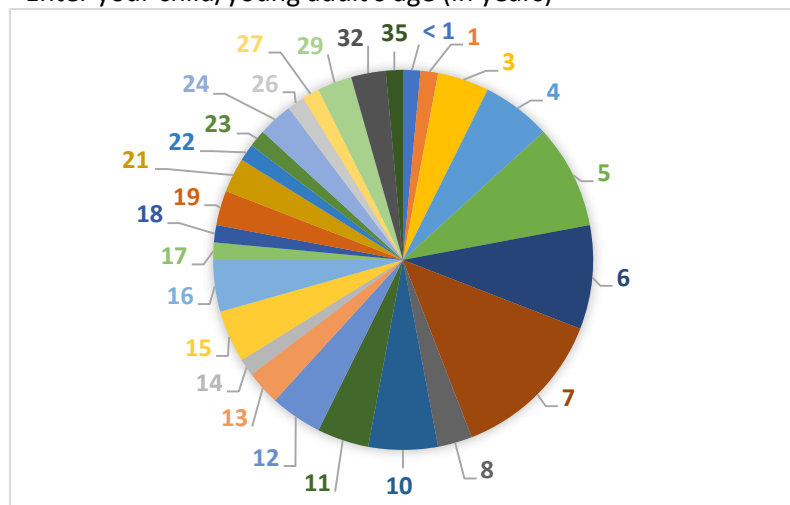
Q1 Who is completing this survey?

Role	#	%
<i>mother</i>	56	82%
father	4	6%
grandmother	2	3%
parent	6	9%
Total	68	100%

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

	#	%
Yes	51	75%
No	17	25%
Total	68	

Q3 Enter your child/young adult's age (in years)



Q4 Child's Age at SETBP1 Diagnosis

Age	#	%
3 - 6 months	1	2%
9 - 12 months	2	3%
13 - 23 months	4	6%
2 years old	4	6%
3 years old	4	6%
<i>4 years old</i>	9	14%
5 years old	4	6%
6 years old	5	8%
7 years old	4	6%
8 years old	2	3%
9 years old	4	6%
10 years old	4	6%
11 years old	2	3%
12 years old	2	3%
13 years old	1	2%
14 - 18 years old	4	6%
19 - 25 years old	4	6%
25 + years old	5	8%
Total	65	100%
Mean/Average		9 years old
Median		6 years old

Q5 Which SETBP1 neurodevelopmental disorder does your child have?

Type	#	%
SETBP1-HD (<i>loss-of-function variant or deletion</i>)	43	63%
SETBP1 related disorders (missense variant)	14	21%
Unsure	7	10%
18q12.3 Deletion (large deletion including SETBP1 and other genes)	4	6%
Total	68	0%

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

	#	%
Female	32	47%
Male	36	53%

Q7 Select your country.

Country	#	%
United States	34	50%
Australia	2	3%
Belgium	1	1%
Bolivia	1	1%
Brazil	1	1%
Bulgaria	1	1%
France	3	4%
Germany	2	3%
Italy	1	1%
Netherlands	9	13%
Norway	1	1%
Poland	1	1%
Portugal	1	1%
Puerto Rico	1	1%
Spain	1	1%
United Kingdom	8	12%



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 or select N/A:

note: question was only asked from caregivers with children <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?*
Skill Acquired							
Average age	10 months	22 months	33 months	45 months	62 months	51 months	50 months
Median age	9 months	22 months	24 months	48 months	60 months	48 months	48 months
Age range for skill acquisition	5 - 24 months	1 - 4 1/2 years	9 months - 10 years	15 months - 7 years	2-10 years	1 - 7 years	1 1/2 - 7 years
Total	49	47	40	35	24	36	27
Skill Not Yet Acquired							
Current		<1 - 1 years	3-7 years	3 - 15 years	3 - 15 years	3 - 7 years	2 - 12 years
Total	0	2	7	15	24	11	22
Total Responses	49	49	47	50	48	47	49

* children 2 years of age and younger were not included in the totals for not acquiring skills

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

	#	%
non-verbal	6	9%
non-verbal (uses an augmentative device to communicate)	2	3%
non-verbal (uses sign language to help communicate)	6	9%
speaks in one-two word phrases or sentences	12	18%
speaks in simple sentences	23	34%
speaks in long, complex sentences	12	18%
other	7	10%
Total	68	



Q10 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 Concern for Specific Age Groups		
	No	Currently	Previously	< 5 years	School-Age	> 17 years
Anxiety	21	40	7	4	22	14
	31%	59%	10%	44%	52%	82%
Depressed mood	54	11	3	3	5	3
	79%	16%	4%	33%	12%	18%
Mood Swings	19	43	6	6	23	14
	28%	63%	9%	67%	55%	82%
Aggressive outbursts	22	35	11	4	23	8
	32%	51%	16%	44%	55%	47%
Temper Tantrums	19	35	14	5	24	6
	28%	51%	21%	56%	57%	35%
Self-injury, such as hitting self, biting self, scratching self	34	17	17	2	12	3
	50%	25%	25%	22%	29%	18%
Absent or delayed onset of language to communicate	5	37	26	8	23	6
	7%	54%	38%	89%	55%	35%
Repeating words or phrases over and over again	20	41	7	6	24	11
	29%	60%	10%	67%	57%	65%
Difficulties getting on with other people of similar age	26	38	4	4	25	9
	38%	56%	6%	44%	60%	53%
Immature development of social skills	13	51	4	4	35	12
	19%	75%	6%	44%	83%	71%
Repetitive behaviors, such as doing the same thing over and over again	20	39	9	7	18	14
	29%	57%	13%	78%	43%	82%
Very rigid or inflexible about how to do things or not liking change in routines	23	38	7	4	19	15
	34%	56%	10%	44%	45%	88%
Overactivity/hyperactivity such as being constantly on the go	22	35	11	5	23	7
	32%	51%	16%	56%	55%	41%
Difficulty paying attention or concentrating	2	65	1	7	41	17
	3%	96%	1%	78%	98%	100%
Restlessness or fidgetiness, such as wriggling or squirming	21	37	10	6	25	6
	31%	54%	15%	67%	60%	35%
Total Responses	68 (100%)			9	42	17



Q10 Continued

Statement	All Ages			Current Concern for Specific Age Groups		
	No	Currently	Previously	< 5 years	School-Age	> 17 years
Impulsivity, such as butting in, not waiting turn	8	53	7	7	35	11
	12%	78%	10%	78%	83%	65%
Difficulties with eating, such as eating too much, too little, or swallowing difficulties (before age 1)	31	18	19	5	8	5
	46%	26%	28%	56%	19%	29%
Difficulties with eating, such as eating too much, too little, unusual things, or swallowing difficulties	33	27	8	5	14	8
	49%	40%	12%	56%	33%	47%
Difficulties with stuffing too much food in their mouth (after age 1)	34	26	8	3	18	5
	50%	38%	12%	33%	43%	29%
Sleep difficulties, such as with falling asleep or waking	35	27	6	4	14	9
	51%	40%	9%	44%	33%	53%
Total Responses	68 (100%)			9	42	17



Q11 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 Concern for Specific Age Groups		
	No	Currently	Previously	< 5 years	School-Age	> 17 years
Blood Disorder	64	1	1	0	0	1
	97%	2%	2%	0%	0%	6%
Bone Disorder	61	5	0	1	1	3
	92%	8%	0%	11%	3%	18%
Congenital Disorder	44	20	2	1	12	7
	67%	30%	3%	11%	30%	41%
Epilepsy/Seizures (other than febrile)	54	8	4	2	4	2
	82%	12%	6%	22%	10%	12%
Febrile Seizures	48	6	12	2	3	1
	73%	9%	18%	22%	8%	6%
Gait/Movement Disorder	35	20	11	4	8	8
	53%	30%	17%	44%	20%	47%
Total Responses	68 (100%)			9	42	17



Q11 Continued

Statement	All Ages			Current Concern for Specific Age Groups		
	No	Currently	Previously	< 5 years	School-Age	> 17 years
Gastrointestinal Problems	33	19	14	2	10	7
	50%	29%	21%	22%	25%	41%
Hearing Issues	59	6	1	1	4	1
	89%	9%	2%	11%	10%	6%
Hypotonia (low muscle tone)	19	29	18	6	17	6
	29%	44%	27%	67%	43%	35%
Hypertonia (high muscle tone)	61	3	2	1	1	1
	92%	5%	3%	11%	3%	6%
Immune Problems	55	6	5	2	2	2
	83%	9%	8%	22%	5%	12%
Mitochondrial Disorder	64	2	0	0	1	1
	97%	3%	0%	0%	3%	6%
Skin Problems (other than acne)	42	15	9	1	7	7
	64%	23%	14%	11%	18%	41%
Sensory Processing Disorder	24	33	9	4	21	8
	36%	50%	14%	44%	53%	47%
Speech and/or Language Disorder	3	56	7	9	34	13
	5%	85%	11%	100%	85%	76%
Vision Problems (corrective eyewear and/or strabismus surgery)	29	31	6	3	18	10
	44%	47%	9%	33%	45%	59%
Vision Problems (not corrected with corrective eyewear and/or)	51	10	5	1	6	3
	77%	15%	8%	11%	15%	18%
Total Responses	68 (100%)			9	42	17



Q12 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
Attention Deficit Hyperactivity Disorder	2 22%	24 60%	7 44%	33 51%
Autism Spectrum Disorder (ASD), including autism,	1 11%	14 35%	8 50%	23 35%
Anxiety Disorder, including as panic, phobia, separation	1 11%	14 35%	6 38%	21 32%
Obsessive Compulsive Disorder (OCD)	0 0%	7 18%	4 25%	11 17%
Oppositional Defiant Disorder (ODD)	1 11%	7 18%	2 13%	10 15%
Depressive Disorder	4 6%	0 0%	1 3%	3 19%
Psychotic Disorder, including schizophrenia, bipolar	0 0%	2 5%	1 6%	3 5%

Q13 IQ Testing Results

	Number	Percentage
Normal Intellectual	1	2%
Borderline Intellectual	10	24%
Mild Intellectual	14	33%
Moderate Intellectual	9	21%
Severe Intellectual	5	12%
Profound Intellectual Disability (IQ <20)	2	5%
Developmental Delay	1	2%
Total	42	



Q14 Many people with SETBP1-HD and related disorders who are of school age will have difficulty in school. If your child/young adult is school-age or older, does your child/young adult have any difficulty with any of the following:

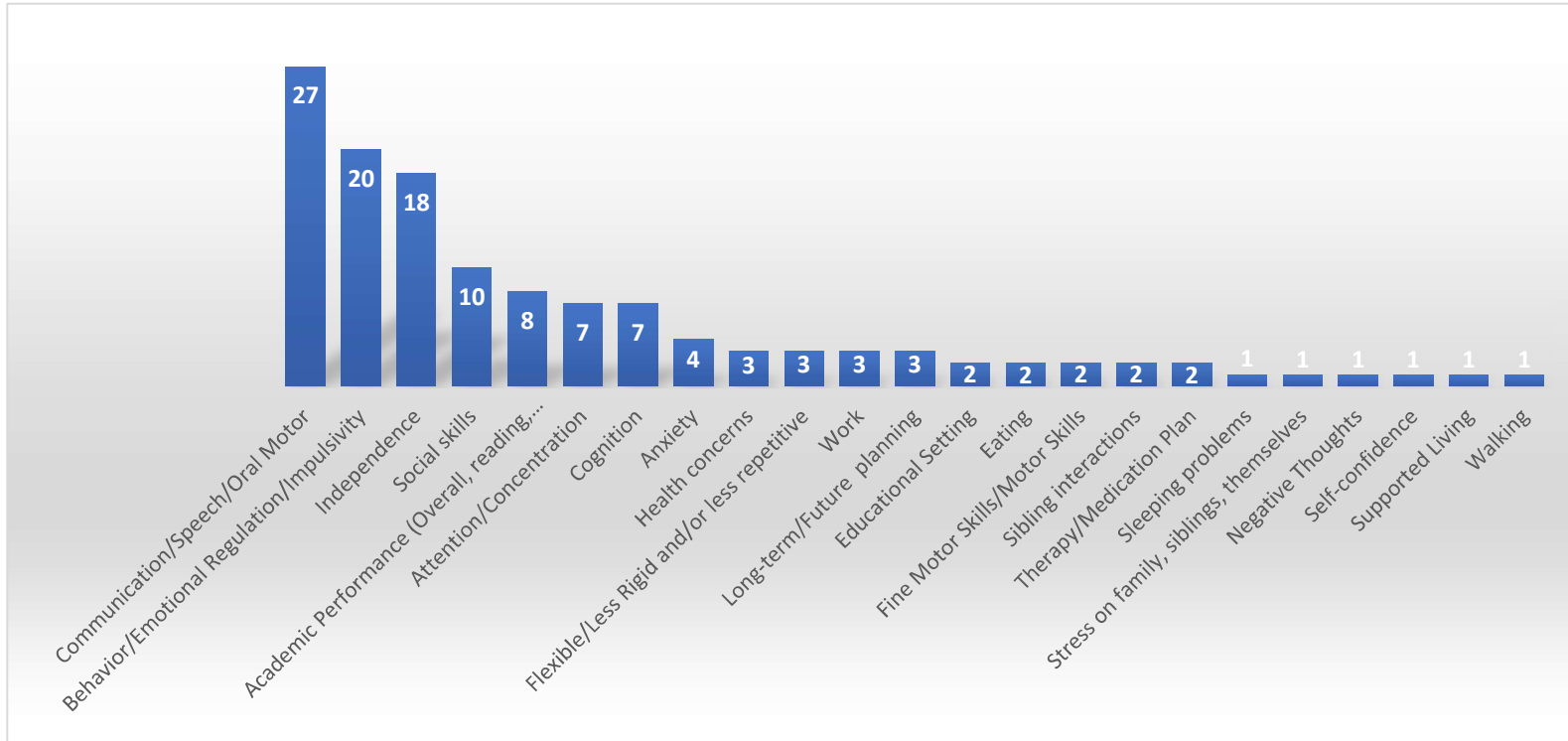
Statement	No
Reading	8
	13%
Writing	7
	11%
Spelling	9
	15%
Mathematics	1
	2%
Total Responses	61

Q15 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		<5 years	School-Age	> 17 years
	No	Yes	Yes	Yes	Yes
Memory, such as remembering things that	38	23	1	16	6
	62%	38%	20%	40%	38%
Attention, such as concentrating well, not getting distracted	3	58	5	38	15
	5%	95%	100%	95%	94%
Dual-tasking/ Multi-tasking, such as doing 2 tasks at the same time	5	56	4	38	14
	8%	92%	80%	95%	88%
Visuo-spatial tasks, such as solving puzzles or using building blocks	24	37	2	25	10
	39%	61%	40%	63%	63%
Executive skills, such as planning, organizing, flexible thinking	5	56	4	38	14
	8%	92%	80%	95%	88%
Getting disoriented, such as not knowing the date or where you are	35	26	0	22	4
	57%	43%	0%	55%	25%
Total Responses	61		5	40	16



Q16 Of all the concerns listed in this survey, what are your top 2-3 priorities to work on next?



Q17 Taking together all the difficulties discussed above, how much have these bothered, troubled or distressed you/your child or young adult/family? Select the value.

Statement	Not at all/0	1	2	3	4	5	6	7	8	9	10/Extremely	Overall
All Ages	3 6%	2 4%	1 2%	3 6%	5 9%	2 4%	2 4%	3 6%	13 24%	11 20%	9 17%	54 100%
< 5 years	2 40%	0 0%	0 0%	0 0%	1 20%	1 20%	1 20%	0 0%	0 0%	0 0%	0 0%	5 100%
School Age	1 3%	1 3%	1 3%	2 6%	4 11%	1 3%	0 0%	3 9%	8 23%	8 23%	6 17%	35 100%
> 17 years	0 0%	1 7%	0 0%	1 7%	0 0%	0 0%	1 7%	0 0%	5 36%	3 21%	3 21%	14 100%



S2. Young Adult Section

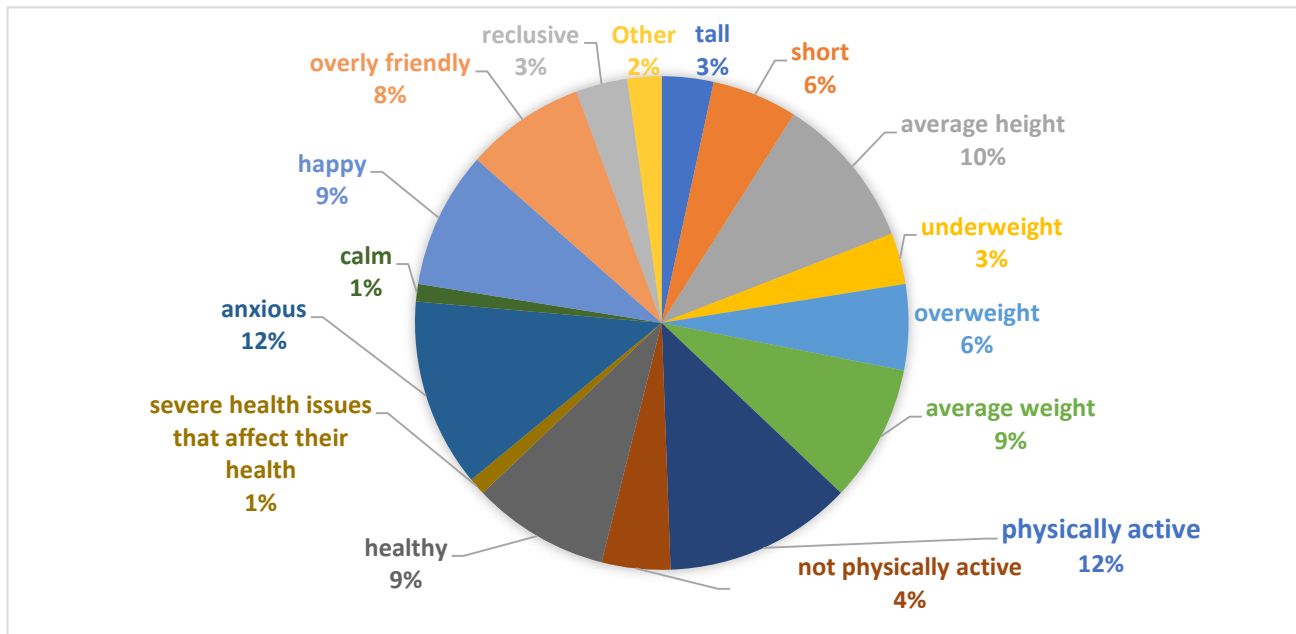
Q19 Has your young adult had any surgical procedures after the age of 16? If yes, please provide details for each surgery.

- 11 no
- 2 oral
- 1 eye (ptosis)
- 1 appendicostomy & appendicovesicostomy
- 1 gall bladder removal
- 1 forum magnum decompression for chiari malformation & detethering of spinal cord

Q20 Has your young adult had any new diagnoses after the age of 16 (other than SETBP1)? If yes, please provide the name of each diagnoses given after the age of 16.

- 13 no
- 1 tethered spinal cord and polycystic ovarian syndrome
- 1 osteoporosis gait ataxia
- 1 urine retention

Q21 Please select all description words that apply currently to your young adult with a SETBP1 difference?



Q22 Does your young adult work? If yes, please provide the jobs your young adult has held after the age of 16.

- 8 no
- yes (carpentry, lawn work, county council as adviser for people with disabilities, maintenance, workshop for disabled,
- 7 waiter for 6 years at a restaurant that employs individuals who may face challenges in finding work due to various



Q23 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, please enter yes.

3 no (residential placement or assisted living)

11 yes (although one will move to residential placement at 23)

Q24 What is your biggest concern for your young adult with a SETBP1 difference?

6 Concern for future after parent is not around

3 Independence

1 Work

1 Friends

1 Communication

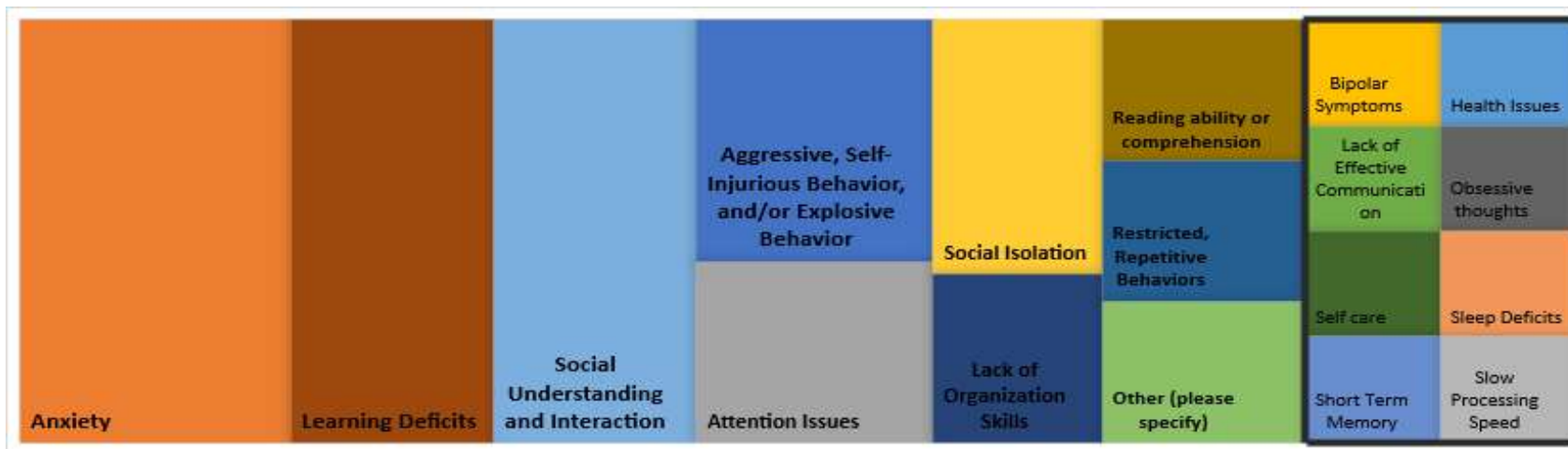
1 Safety

1 Future in general

1 Proper placement back into residential placement

1 Social inclusion

Q25 Which symptoms are most impactful for your young adult currently? Select up to 3.



Note: The black box contains concerns selected by ONLY 1 caregiver. Other options that no caregiver selected were: long term memory, hearing issues, motor control/coordination challenges, seizure control, sensory integration issues, and vision issues

