Adaptation of Goal Attainment Scaling for Prader-Willi Syndrome: Development of a goal inventory for standardized implementation

ARDEA OUTCOMES

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▶ Background

- Prader-Willi Syndrome (PWS) is a complex neuroendocrine disorder with notable heterogeneity in disease manifestation and symptom progression. To better understand the impact of treatments for children with PWS, we proposed to develop a draft goal inventory for individuals with PWS.
- Goal Attainment Scaling (GAS) is a personalized endpoint that quantifies the impact of an intervention on individualized goals. Clinicians, patients, and/or care providers collaboratively identify goals that are meaningful to the patient and develop unique scales for each identified goal.
- These semi-standardized goal inventories:
  1. Maximize the psychometric properties of the scales
  2. Streamline the goal scale development process, and
  3. Provide a shared language between the patient and the clinician.

How Does Goal Attainment Scaling Works?

Identify Goals
Clinician to facilitate interview for subject or caregiver to identify goals

Build GAS Scales
Set the 5-point goal attainment scale for each identified goal

Measure Attainment
Rate during follow-up whether the goals have been attained

The Goal Attainment Scaling Process

1. Build a semi-standardized goal inventory
2. Train clinicians on GAS method
3. Conduct the goal setting visit
4. Assess goal attainment at follow up

▶ Methods

Five clinicians in the US with expertise in PWS participated in focus groups or an interview, and using semi-structured qualitative interview questions, the expert clinicians identified symptoms, challenges, and issues for people living with PWS and their families.

Interviews were recorded and transcribed; symptoms, challenges, and issues were then identified and coded using qualitative analysis software (NVivo 12).

▶ Results

IMPACT ON THE PERSON LIVING WITH PWS
Ability to work or go to school
Requiring supervision
Difficulty managing money
Future planning
Independence
Relationships
Vulnerability
Not understanding boundaries
Low self-esteem/feeling different
Social isolation
Susceptible to trauma
Being treated differently
Obsession with animals or babies
Treatments and therapies (speech, braces, etc.)
Other clinical diagnoses (depression, psychosis, etc.)

IMPACT ON THE FAMILY
Financial burden
Parental and sibling stress and anxiety
Dealing with unwanted behaviors
Safety concerns and worry
Providing routine; visual schedules, transitional cues
Need for constant supervision
Food security/locking food
Accessing care
Caregiver burden and burnout
Long-term care planning
Lack of available treatments and care protocols

- Age 0-2 years - low muscle tone, lack of interest in feeding (failure to thrive), sleep disorders, and global developmental delay.
- Age 3-8 years - weight gain, hip dysplasia, scoliosis, developmental delay, sleep disorders, and social-emotional and behavioral issues.
- Age 9 to adulthood - behavioral issues, hyperphagia, endocrine system issues, impacts on musculoskeletal functioning, and difficulties with sleep.

- Family impacts noted were parental and sibling stress and anxiety, the need for constant supervision, caregiver burnout, and the continual worry regarding safety.

DRAFT OF THE PWS GOAL INVENTORY

AGE 0-2	AGE 3-8	AGE 9-12	AGE 13-17	AGE 18+
Narcolepsy	Narcolepsy	Narcolepsy	Narcolepsy	Narcolepsy
Cataplexy	Cataplexy	Cataplexy	Cataplexy	Cataplexy
Trouble sleeping or restless sleep	Trouble sleeping or restless sleep	Trouble sleeping or restless sleep	Trouble sleeping or restless sleep	Trouble sleeping or restless sleep
Sleep apnea	Sleep apnea	Sleep apnea	Sleep apnea	Sleep apnea
Excessive sleepiness	Excessive sleepiness	Excessive sleepiness	Excessive sleepiness	Excessive sleepiness
Scoliosis	Scoliosis	Scoliosis	Scoliosis	Scoliosis
Hip dysplasia	Hip dysplasia			
Weight gain or fluctuations, obesity	Weight gain or fluctuations, obesity	Weight gain or fluctuations, obesity	Weight gain or fluctuations, obesity	Weight gain or fluctuations, obesity
		Low bone mineral density or osteoporosis	Low bone mineral density or osteoporosis	Low bone mineral density or osteoporosis
Low muscle tone				
Difficulty breathing				
		Constipation	Constipation	Constipation
	Hyperphagia and food seeking	Hyperphagia and food seeking	Hyperphagia and food seeking	hyperphagia and food seeking
Failure to thrive				
Lack of interest in eating or feeding				
Feeding tubes, over or under eating				
		Pubertal issues	Pubertal issues	Pubertal issues
		Hypogonadism	Hypogonadism	Hypogonadism
Undescended testicles	Undescended testicles			
Blood sugar high or low				
	Blurred perception of reality	Blurred perception of reality	Blurred perception of reality	Blurred perception of reality
Speech issues	Speech issues			
Delayed fine motor skills				
Decreased gross motor skills				
Global developmental delay				
	Bed wetting			
Language delay	Language delay			
Verbal apraxia	Verbal apraxia	Verbal apraxia	Verbal apraxia	Verbal apraxia
		Breaking the law	Breaking the law	Breaking the law
		Social withdrawal	Social withdrawal	Social withdrawal
	Inattentive	Food related behavior problems	Food related behavior problems	Food related behavior problems
Affectionate		Aggression	Food related behavior problems	Food related behavior problems
		Interrupting	Interrupting	Interrupting
		Impulsive	Impulsive	Impulsive
	Stealing	Stealing	Stealing	Stealing
	Skin and rectal picking	Skin and rectal picking	Skin and rectal picking	Skin and rectal picking
	Lying or making up stories	Lying or making up stories	Lying or making up stories	Lying or making up stories
		Attention seeking behavior	Attention seeking behavior	Attention seeking behavior
	Obsessive or hoarding behavior	Obsessive or hoarding behavior	Obsessive or hoarding behavior	Obsessive or hoarding behavior
	Repetitive behavior		Repetitive behavior	
	Stubborness, tantrums, outbursts	Stubborness, tantrums, outbursts	Stubborness, tantrums, outbursts	Stubborness, tantrums, outbursts
	Rigidity or inflexibility	Rigidity or inflexibility	Rigidity or inflexibility	Rigidity or inflexibility
	Needs routine	Needs routine	Needs routine	Needs routine

COLOR KEY - from the top
Sleep disorders
Physical Body Impact
Respiratory Issues
Gastrointestinal Issues
Food and Nutrition
Endocrine System
Cognitive & Developmental Delay
Behavior

▶ Discussion & Conclusion

Critical to the drug development efforts, expert clinicians introduced a variety of other symptoms and manifestations of the disease that are not in the current diagnostic criteria but still impact the person living with PWS and their families.

The heterogeneity in symptom manifestations corroborates the value of GAS in capturing meaningful change following treatment, with the purpose of the inventory to help standardize GAS implementation in clinical development.

Patient reported symptoms are often underrepresented and thus limit opportunities to advance future management and treatment options. Qualitative research with patients and families is needed to further refine the inventory.

▶ Acknowledgements & References

Kiresuk, T. J., & Sherman, R. E. (1968). Goal attainment scaling: A general method for evaluating comprehensive community mental health programs. *Community mental health journal*, 4, 443-453.

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RUNNING FOR RESEARCH PRADER-WILLI SYNDROME

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Title: Adaptation of Goal Attainment Scaling for Prader Willi Syndrome (PWS): Development of a goal inventory for standardized implementation

Purpose: Goal Attainment Scaling (GAS) is a personalized endpoint that quantifies the impact of an intervention on individualized goals. Clinicians, in partnership with the patient and/or care provider, identify goals that are meaningful to the patient and develop unique scales for each identified goal. Semi-standardized goal inventories 1-) maximize the psychometric properties of the scales; 2-) streamline the goal scale development process; and 3-) provide a shared language between the patient and the clinician. Here we aimed to develop a goal inventory for individuals with Prader Willi Syndrome (PWS), a complex neuroendocrine disorder with notable heterogeneity in disease manifestation and symptom progression.

Background: Expert clinicians identified the symptoms, challenges and issues of individuals living with PWS and their caregivers through semi-structured qualitative interviews.

Methods: Five clinicians in the US with expertise in PWS participated in focus groups or an interview and were asked questions regarding the symptoms, challenges and issues for people living with PWS and their families, categorized by age (0-2; 3-8; 9-12; 13-17 and 18+). Interviews were recorded and transcribed; then symptoms, challenges and issues were identified and coded using qualitative analysis software (NVivo 12).

Results: For PWS patients aged 0-2, clinicians stated low muscle tone and other impacts on the musculoskeletal functioning and development, lack of interest in feeding (failure to thrive), sleep disorders, global developmental delay and effects on the endocrine system. For patients aged 3-8, they expressed weight gain, hip dysplasia, scoliosis, developmental delay, sleep disorders, social-emotional difficulties (e.g. trouble forming relationships and vulnerability) along with behavioral issues (e.g. tantrums, repetition, lying, stealing and skin picking). Clinicians identified similar symptoms from age 9 to adulthood including behavioral issues (e.g. tantrums, repetitive behaviors, and need for routine); as well as hyperphagia; endocrine system issues, impacts on the musculoskeletal functioning (e.g. scoliosis, osteoporosis, and mobility issues), GI concerns, and difficulties with sleep. Among family impacts,

parental and sibling stress and anxiety, the need for constant supervision, caregiver burnout and the continual worry regarding safety were noted.

Conclusions: Critical to the drug development efforts, expert clinicians introduced a variety of other symptoms and manifestations of the disease that are not in the current diagnostic criteria, but still impact the person living with PWS and their families. The heterogeneity in symptom manifestations corroborate the value of GAS in capturing meaningful change following treatment, with the goal inventory standardizing its implementation to enhance drug development efforts.