Cognitive and Behavior Skills in Children with Glut1 Deficiency

Goal 1. Determine characteristics associated with Glut1 Deficiency

Intellectual Function
There is a wide range across the group and the mean of the group is shifted down from general population about 2 standard deviations.

Language Ability
- Communication difficulties are common: Poor articulation, dysfluent speech
- Receptive language skills are stronger than expressive skills
- Children understand more than they may seem to judging by their speech limitations

Adaptive Behavior, as reported by parents on standardized questionnaire
- Adaptive behavior composite scores are shifted down from general population
- Social skills are strengths!

Behavior
- There is no evidence of any characteristic unusual behaviors associated with the disorder
- Poor attention is common
- Behavior concerns are not associated with neurological score, cognitive score, adaptive behavior or age
- Although some children may have problem behaviors, there was no consistency observed across the group

Processing Style
- Among higher functioning children there is a primary reliance on using a sequential information processing approach
- This differs from the general population who use both approaches comparably
- Holistic and spatial integration strategies are particularly impaired
Goal 2: To examine relationship of skills with different variables, such as neurological function, gender, age

Neurological performance
- Cognitive performance is associated with neurological score

Age and Gender
- There is no association of cognitive performance with age or gender

Goal 3: To examine effects of development over time

Cognitive skills
- Cognitive Skills remain stable across time

Adaptive behavior
- Adaptive behavior remains stable across time

Goal 4: To consider implications of findings on how to optimize quality of life

There is a wide range of cognitive function and as a group, scores are shifted down from general population
- Every Individual is unique!
- We present GROUP data, but each child needs a thorough clinical neuropsychological evaluation to determine his or her own individual strengths and weaknesses
- Data show what children are “at risk for” if they have Glut1 Deficiency diagnosis, but not all children will have every characteristic

Receptive language skills are stronger than expressive language skills
- Speech therapy is recommended!
- Because speech is dysfluent and poorly articulated, children may appear to be more impaired than they are. Speech therapy can help children express themselves with more ease.
- Children may be frustrated at times by the difficulties they face getting understood by those who don’t know them well
Visual attention to details is weak

- These are areas emphasized in academic school work but can be trained and improved
- Visual search puzzle-games like “spot the difference” in pictures, eye-spy, word search and “Where’s Waldo” help train this ability
- Help go over school assignments carefully, step-by-step to help teach your child to focus on details

Fine motor skills are weak

- Another areas emphasized in academic school work – writing and copying
- Physical therapy and occupation therapy can help with this
- Encouraging play with blocks and legos and small items as well as encouraging drawing is recommended
- For some, use of an assistive keyboard may be essential

There is a definite bias in cognitive processing style. Be aware of limitations

- Give extra help when trying to pull together the whole picture
- Talk about stories and what the point is (fables and folktales work well for this!)
- Play with abstract puzzles where the whole is greater than the sum of the parts – work on integrating visual information

Adaptive behavior scores are comparable to scores on cognitive tests

- These are crucial areas to learn
- Helping children be more independent in caring for themselves will instill greater self-confidence and help them achieve more

Social skills are strengths!!

- Children are delightful and full of charm and empathetic and socially outgoing.
- Their ability to make friends will serve them throughout life
- This is a remarkable strength and one that is not emphasized as much academically as other skills
- Encourage them to be active in school groups and all social settings
- Thus, praise and encouragement are likely the most useful tools in teaching children with Glut1 Deficiency
There are no aberrant behaviors associated with Glut1 Deficiency

- There may be inattention and adjustment concerns related to living with a developmental disability
  - Consider using behavioral interventions that work for children with Attention Deficit Hyperactivity Disorder

- The predominant behavior characteristic is how cued into others the children are

- Thus, praise and encouragement are likely the most useful tools in teaching children with Glut1 Deficiency

For those children on the diet, normal developmental gains are made over time. There is no evidence of decline!

- Even though the ketogenic diet may not be a “cure,” it does help keep children developing and there is no evidence of progressive worsening

- Attention is better maintained, allowing for the child to learn more

- Stay in good ketosis!!!