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“Cognitive and Behavior Skills in Children with Glut1 Deficiency Syndrome”

The GLUT1 gene is responsible for making the glucose protein. The job of the Glut1 protein is to transport glucose into the brain. This ultimately affects the structure and the function of the brain, resulting in cognition and behavior.

Seizures affect function of brain and the outcome can result in a change in cognition and behavior.

Intellectual function in Glut-1 DS patients:

Normal population average IQ is 100. Glut-1 DS kids have a wide range of functioning, but tend to fall below the average IQ.

Verbal skills in Glut-1 DS patients:

Communication skills- receptive language (*understanding what someone else has said*) is stronger than

expressive language (*expressing self through speech*).

Poor articulation/dysfluent speech (*speech is uneven*)

Non verbal skills:

We can test these skills which don't require the patient to speak

Tests include: drawing, matching, peg placing and fine motor skills.

Most kids fall below the average range, but some children are above the normal functioning.

Cognitive processing styles in general:

1) Sequential or local processing-absorbing information bit by bit (see the

trees before the forest). 2) Simultaneous or global processing-synthesize information (see the forest before the trees).

Glut-1 patients do better with sequential processing tasks

In higher functioning Glut-1 DS kids there is a primary reliance on sequential processing

Adaptive behavior (how does a person function in the real world?)

A person might do poorly on an IQ test but might be able to function well in the real world.

Adaptive behavior and IQ are positively correlated- if you have more IQ you will have more adaptive behavior.

Social skills are considered part of adaptive behaviors. Social skills are relative strengths in the Glut-1 patients. Glut1 DS parents report that their children are warm and affectionate, playful, empathetic, humorous, considerate, interested in others, and well liked.

Behavioral Issues

Children with developmental delay tend to have a higher incidence of behavior problems overall. However, if we look at the data about the Glut-1 DS children, we do not see any increased frequency in unusual behaviors in the kids compared to children without Glut-1 DS. Furthermore, behaviors are not associated with neurological score (i.e. if a Glut-1 DS child has a worse neurological score than another Glut-1 DS child, he/she is no more likely to have behavioral problems). It should not be assumed that behavioral issues that are seen in the Glut-1 DS patients are caused by the Glut-1 DS. In general, behavior in Glut-1 DS patients is within normal limits.

Glut-1 DS patients may be at risk for adjustment difficulties. This could be even more so with higher functioning more insightful children. What we have seen seems to be normal. It doesn't seem to be related to Glut-1 DS.

In Glut-1 DS, IQ is associated with neurological function and it is positively correlated. This means that if your neurological functioning score is higher you are more likely to have a higher IQ also (lower IQ scores are associated with lower neurological function).

Cognitive performance is not associated with age or gender in Glut-1 DS. We can make no strong statements regarding IQ and mutation type.

Performance across time

Developmental gains are made with time.

Relative functioning compared to peers seems to be stable over time. There is no evidence of progressive decline over time.

Cognitive skills tend to be the same over time.

Adaptive behavior tends to stay the same over time.

Behavior skills stay the same over time.

What are the implications of these findings?

1. Skills vary between Glut1 DS children.
2. Your child is unique.
3. Because of communication difficulties children can be judged as less intelligently than they are. Thus, targeting speech skills is important.
4. Visual search skills should be used in training these kids. For example try tests such as “spot the difference in this picture”.
5. Go over your child’s homework.
6. Since fine motor skills are weak, physical therapy and occupational therapy can help. The use of technology to help communicate could be of benefit.

7. Use teaching strategies that rely on sequential of step-wise approach (give bit by bit).
8. Rely on rote memorization and repetition.
9. Break large assignments into manageable steps. Consider using organization strategies similar to those recommended for kids with ADHD .
10. Give kids a little extra time/help when trying to pull together the whole picture.
11. Play with abstract puzzles where they have to pull together the sum of the parts to make a whole picture.
12. Work on adaptive behavior skills- give them chores at home and hold them to those standards.
13. Social skills are strengths- encourage kids to be active in school groups and all social groups.
14. There is no aberrant behavior associated with Glut1 DS. There may be inattention and adjustment concerns related to living with a developmental disability.