GLUT1 DEFICIENCY SYNDROME (GLUT1DS)

Pre-natal diagnosis

Often not feasible as most mutations are de novo and the mutation is not detectable with only karyotype

Clinical Presentation / Symptoms
If one parent has a known SLC2A1 mutation, there is a 50% probability of having a child with Glut1DS

Identifying patient needs
Understanding the risks and consequences of having children

Ideal outcome / support
Genetic counseling and pre-natal testing in at-risk families

Pre-symptomatic diagnosis

First months of life

Clinical Presentation / Symptoms
Normal children with no symptoms

Identifying patient needs
Identify asymptomatic patients in order to begin treatment before energetic deficit can permanently impair cerebral function

Ideal outcome / support
Universal perinatal screening

First symptoms

3 months – adolescence

Clinical Presentation / Symptoms
Developmental delay ranging from mild to severe epilepsy, movement disorder (constant, intermittent, or paroxysmal)
(in variable combination of degree and severity)

Identifying patient needs
Family needs reassurance and support in optimizing the care for the child, as well as guidance in management and the diagnostic journey

Ideal outcome / support
Early recognition of patients with mild phenotypes, and early intervention with rehabilitation and social support

Diagnosis

Earlier the better

Clinical Presentation / Symptoms
Cognitive impairment and/or epilepsy and/or movement disorder
Low glucose in cerebrospinal fluid upon spinal puncture
Genetic testing for confirmation

Identifying patient needs
Family must understand the importance of this diagnosis in that it allows the implementation of the ketogenic diet that often could be considered a disease modifying therapy which can improve the immediate and long-term outcomes for the patient

Ideal outcome / support
Provide adequate information on the disease, genetic counseling, and referral to a ketogenic diet therapy center and patient support group.
Explanation of implications of diagnosis

Treatment

From diagnosis then life-long

Clinical Presentation / Symptoms
Ketogenic Diet therapy is so fa the gold standard treatment

Identifying patient needs
Guidance in implementation of the ketogenic diet
Optimization of management
Adequate follow-up
Management of adverse effects and long-term risks of the treatment
Rehabilitation

Ideal outcome / support
Genetic counseling and pre-natal testing in at-risk families

Follow-up

life-long

Clinical Presentation / Symptoms
Clinical evaluations: neurological, dietological, endocrinological, neuropsychological, emotional, and behavioral (if applicable) evaluation
Instrumental evaluations: Blood tests and Complete Urinalysis; EEG, Calorimetry, Abdomen echo, supra aortic trunks echocolordoppler (annually), Computerized bone mineralometry (annually or biennal).
Telemedicine visits, which have been recently implemented in the clinical practice, could be considered a valuable option to be associated to on-site visits.

Identifying patient needs
Continuous fine-tuning of KDT and side effects management, Growth monitoring, Developmental and neuropsychological monitoring, Rehabilitation management

Ideal outcome / support
Optimal long-term maintenance of ketosis with limited adverse effects and long-term metabolic risk
Optimal growth and development outcomes
Optimal global outcomes for individual, family, society