

# GLUT1 deficiency syndrome

Acute risks: seizures, paroxysmal dyskinesia.



## 1 FOR A&E CONSULTATIONS

- If the consultation is for a neurological reason, contact the patient's referring neurologist if necessary.
- If the reason for consultation is not neurological, manage the condition that prompted the visit to A&E or the hospitalisation as you would for any other patient, without requiring input from a specialist in metabolism.

## 2 PATHOPHYSIOLOGY

**This is a neurological disorder. There is no risk of metabolic disturbance.**

It is caused by a deficiency of the cerebral glucose transporter GLUT1 (glucose transporter protein type 1). Patients present with chronic neurological involvement characterised by one or more of the following: epilepsy, intellectual disability, spasticity, ataxia, dystonia, and dysarthria. They may also experience paroxysmal neurological episodes, often occurring before meals. The treatment (which should never be stopped) consists of:

- A ketogenic diet (more or less strict depending on the patient), low in carbohydrates and high in lipids (specific Ketocal formula for some patients). Provides an alternative energy source for the brain. Its effectiveness can be assessed by the presence of ketosis.
- Triheptanoin oil for some patients. Provides an alternative energy source for the brain.
- Antiepileptic drugs, depending on the patient, and symptomatic treatments (for dystonia, spasticity, etc.).

## 3 MANAGEMENT

### A- Treatments

- **Continue all usual treatments**
- **If there is no vomiting, maintain the usual ketogenic diet** orally or through continuous enteral feeding. If not provided by the parents, Ketocal can be easily reconstituted as follows:

Define the total intake according to age- and weight-based recommendations

Reconstitution	Ketocal	Maltodextrin	Water	Final volume	Energy intake
4/1	14 g	0	90 mL	100 mL	1.0 Kcal/mL
3/1	14 g	1 g	90 mL	100 mL	1.0 Kcal/mL
2/1	14 g	2 g	89 mL	100 mL	1.0 Kcal/mL
1/1	12 g	6 g	87 mL	100 mL	1.0 Kcal/mL

The Ketocal reconstitution determines the fat-to-carbohydrate ratio provided by the diet or enteral nutrition. The higher the ratio (4/1), the stricter the ketogenic diet. During acute decompensation, the ketogenic diet may be intensified (made stricter). Outside of decompensation, maintain the ratio of the patient's usual diet (as known by the patients or parents).

- **In cases of food intolerance:**
  - **Infuse 2.5% glucose** to avoid compromising ketosis (e.g. to prepare 1 litre of 2.5% glucose: mix or administer via a Y-Set a 500 mL bag of 0.9% NaCl and a 500 mL bag of 5% glucose solution at the same infusion rates + electrolyte supplementation based on blood electrolyte levels). **Daily volume:** as required, based on usual age-related intake and clinical status.
  - **Via Y-Set, if available, IV lipid infusion:** 1.5 g/kg/day or 0.3 mL/kg/h (Medialipids 20%, Smoflipids 20%, Ivelip 20%, etc.)

*If IV access is not possible → Nasogastric tube. Administer the above IV fluids through the tube at the same infusion rates via a Y-Set (better tolerated than enteral feeding).*

### B- Specific Management

- **If status epilepticus occurs:**
  - Administer antiepileptic treatment according to standard protocol (benzodiazepines as first-line treatment, phenytoin as second-line treatment).
  - Consult the referring neurologist about increasing the ketogenic diet (see table above).

### C- Other recommendations, applicable in all cases

- Avoid glucose loading
- Treat both the cause of decompensation (infection, etc.) and the symptoms

## 4 DRUG CONTRAINDICATIONS/GENERAL ADVICE:

- Some antiepileptic drugs that inhibit the GLUT1 transporter should be avoided: phenobarbital, diazepam, valproate
- All vaccinations are recommended.
- In case of surgery: No contraindication to anaesthetics. Maintain the ketogenic diet with an appropriate infusion (see above). Continue all usual treatments.

View the Emergency section of the G2M website



REFERRING DOCTORS AND CONTACT DETAILS

On-call telephone numbers for metabolic emergencies of:

At night, only the medical teams can call in emergency situations and only if the emergency certificate has not been understood or if the patient's clinical condition or test results are concerning. Where possible, calls should be made before night time. Secretarial issues must be dealt with via the medical secretariat during the week or by email addressed to the patient's referring metabolic doctor.

Certificate issued on

Dr