BIMDG

British Inherited Metabolic Disease Group

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| **Contact Details Name:** |
| **Hospital** |
| **Telephone:** |

- …….. has **CITRIN DEFICIENCY**
  **(also called Citrullinaemia type 2)**

- Please read carefully. ASSESSMENT IS URGENT. Meticulous treatment is important as there is a high risk of serious complications.

- **Important note: The management of illness in CITRIN deficiency is quite different from other metabolic disorders. These patients have a special oral emergency regimen.**

- The major acute complications are encephalopathy and the patients are treated with a high protein, high fat, low carbohydrate diet. Patients may develop hypoglycaemia

- **Give normal saline 10 ml/kg** unless the peripheral circulation is poor or the patient is frankly shocked, and then give 20 ml/kg normal saline as a bolus immediately after the glucose. Repeat the saline bolus if the poor circulation persists as for a shocked non-metabolic patient.

- Restart the usual oral diet as soon as possible. If this is not possible, initially give normal saline IV for rehydration and maintenance fluids (ie daily volume = 100ml/kg for 1st 10kg then 50 ml/kg for next 10kg then 20ml/kg thereafter).

- **DO NOT GIVE GLUCOSE ORALLY OR INTRAVENOUSLY (except for proven and symptomatic hypoglycaemia)**

- **If proven hypoglycaemia and able to take oral fluids safely give glucose or glucose polymer glucose 10% 3ml/kg ( 300mg/kg) or if not able to take glucose orally give a single bolus of glucose intravenously of glucose 10% 2ml /kg (200mg/kg).**

- **Contact local metabolic unit for further advice about management as this is difficult.**


Last Revised January 2017