

# Biotinidase deficiency or holocarboxylase synthetase deficiency

Patient label

No risk of decompensation if treatments are taken compliantly.

## 1 PATHOPHYSIOLOGY

**There is NO RISK of coma or acute metabolic decompensation with this disorder, as long as the treatment is taken correctly and is well absorbed.**

Biotin (vitamin B8) is the cofactor for 4 carboxylase enzymes (pyruvate carboxylase, propionyl-CoA carboxylase, 3-methylcrotonyl-CoA carboxylase and acetyl-CoA carboxylase). If there is a disorder of intracellular biotin metabolism (recycling by biotinidase, or binding of biotin to carboxylases by the holocarboxylase synthetase), these enzymes are no longer able to function. This explains the clinical signs (coma with hypotonia, seizures, acidosis, skin lesions [often around orifices], alopecia, kerato-conjunctivitis, psychomotor retardation, optic atrophy, and central deafness), as well as the laboratory findings (lactic acidosis with ketosis, specific biochemical abnormalities revealed by organic acids urine analysis and acylcarnitines).

**Treatment is based on Biotin (vitamin B8): orally 5-10mg/day for biotinidase deficiency, 10-100mg/day for holocarboxylase synthetase deficiency. LIFELONG daily treatment.**

Once they are treated, patients no longer show any symptoms of their disease.

## 2 IN CASE OF HOSPITALISATION / INTERCURRENT DISEASE

If vomiting / refusal of food / diarrhoea / incorrect administration of biotin treatment: the treatment must not be stopped for several days. **Double the dose in case of gastrointestinal intolerance.** An IM form (Biotin 5mg/1mL) is available. The biotin content of IV polyvitamin solutions (soluvit, cernevit) is too low for these to be considered as alternative.

If the patient is consulting for another reason: ensure that the biotin treatment is being taken correctly and continue as normal, **without ever stopping it.** Manage the condition leading to A&E visit as for any other patient. There is no need for expert metabolic advice.

## 3 DRUG CONTRAINDICATIONS / GENERAL ADVICE:



**Contraindications:** none

- All vaccinations are recommended.

## 4 ACTION TO BE TAKEN BEFORE PLANNED GENERAL ANAESTHESIA

- Continue treatment with biotin
- If oral administration is not possible for several days, consider switching to IM administration.

Look up the emergency information on the G2M site



## NUMBERS AND MEDICAL SPECIALISTS

To be completed by each department

At night, only medical teams can call in emergency situations, and only if the emergency certificate has not been understood or if the clinical state or test results are worrying. As far as possible, make calls before nighttime.

Secretarial issues must be dealt with via the medical secretariat during the week, or by email addressed to the patient's metabolic medicine specialist.

Certificate issued on

Dr