

What to do under a suspect of a defect of creatine metabolism or transport?

When the patient clinical picture (see [“When have a suspect ...”](#)) suggests a defect of synthesis or transport of creatine, we must perform the biochemical and/or neuroimaging investigations aimed to confirm or exclude one of these defects.

The first “screening” test includes measurement of creatine and guanidinoacetic acid in a random urine sample. If the result is abnormal, following the concentrations of these two metabolites it is possible to distinguish among the three genetic creatine defects: defect of AGAT, GAMT or of the creatine transporter CrT1.

This assay is available in all Laboratories included into the Workgroup (look at the [“services”](#) page for details).

Collect a random urine sample, freeze it at -20°C and sent to the lab in dry ice.

The lab must be contacted first.

At neuroimaging level, the MR spectroscopy is able to enhance a defect of creatine in brain , leading to the diagnosis, but without the possibility to distinguish among the three defects.

This investigation is available in some of Workgroup Centres (look at the [“services”](#) page for details)

Once the clinical suspect has been confirmed by biochemical or neuroimaging abnormalities, it is mandatory to proceed to a full investigation following the [diagnostic algorithm](#).