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Sommario/riassunto	Creatine (Cr) transporter deficiency (CCDS1) is a very rare and severe condition due to impaired energetic metabolism. In this work we showed for the first time the following facts: this diseases is a progressive neurodegenerative disorder in which a set of maladaptive compensatory mechanisms leads to a progressive damage of brain functions; cell energy metabolism and mitochondria seem strongly involved in the pathogenesis and they could represent useful potential targets for therapeutic interventions; inflammation seems to play an important part in this progressive damage, and this observation can pave the way to treatment strategies; neural circuits disruption involving inhibitory systems could give a huge contribute to many of the clinical aspects observed in patients, as epilepsy and cognitive impairment, since the excitatory/inhibitory balance is fundamental for the normal function of neural circuits. Factors outside the CNS are important in the pathogenesis of at least some aspects of the disorder, since the conditional KO model show difference in the timing of onset of some cognitive defects and in the presence of stereotypies.