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## The efficiency of adequate diagnostics in the treatment of metabolic crises

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Children with an acute organic aciduria in the neonatal period, as an acute metabolic crisis, need emergency specialized nursing and consultation by the clinical geneticist. The conduction of the selective screening has allowed revealing the following diseases among all children who are in intensive care units (centers for emergency medical care and disaster medicine): Glutaric aciduria type 1 (GA1) – six patients; Canavan disease – three patients; Alkaptonuria – four patients; maple syrup urine disease – five patients; 5-oxoproline aciduria (deficiency of the mitochondrial import of 2-aminoadipate) – one patient; disorders of mitochondrial oxidation of fatty acids – four patients; peroxisomal disorders – one patient; tetrahydrofolate reductase deficiency– one patient; carnitine deficiency – one patient. Three of patients with 5-oxoproline aciduria died. The rest of these patients survived. Medication that should be available in every accident emergency department of children's hospitals: biotin, folic acid, folinic acid, hydroxocobalamin, L-arginine-HCl, L-carnitine, pyridoxal phosphate, pyridine-HCl (Vitamin B6), Na-benzoate. The chances of survival rate of children will be increased due to the presence of the mentioned medication in every accident emergency department. The received experience has increased the survival rate of children, which for today is 88.5 percent.

### Biography

Olena Crechanina has completed her PhD from National Medical University, Department of General Medicine; and Postdoctoral studies from National Medical University, Department of Obstetrics and Gynecology. She is the General Director of Kharkiv Interregional Specialized Medical Genetic Center – Center of Rare (Orphan) Diseases.

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