The aim of mass neonatal screening from dry blood spot is to systemically and actively identify specific congenital disorders in all the newborns. Inborn conditions being screened are either without clinical symptoms in early postnatal period, or the symptoms are easily overlooked. The onset of symptoms is often past the critical time when clinical damage was reversible. Mass neonatal screening enables early diagnostics and intervention. Screening prevents morbidity associated with these conditions, especially concerning central nervous system. It avoids the manifestations of acute crises, which may leed to the death of patients.

Mass newborn screening has been a very successful preventive program to be initiated in the Czech Republic in 1975. Since 1.10. 2009 the neonatal screening in the Czech Republic encompasses these conditions: phenylketonuria/hyperphenylalaninemia, congenital hypothyroidism, congenital adrenal hyperplasia, cystic fibrosis, maple syrup urine disease, medium-chain acyl-CoA dehydrogenase deficiency, very long-chain acyl-CoA dehydrogenase deficiency, long-chain hydroxyacyl-CoA dehydrogenase deficiency, carnitine palmitoyl transferase deficiency I, cernitine palmitoyl transferase deficiency II, carnitine/acylcarnitine translocase deficiency, glutaric aciduria I and isovaleric acidemia.

Longitudinal follow-up of patients diagnosed by the methods of mass newborn screening has confirmed its extensive medical and economical benefit. Laboratory tests used for the screening have high values of sensitivity and specificity.