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Saving children's lives by newborn screening for metabolic disorders

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Newborn screening for metabolic diseases was initially introduced in the 1960's with a program for the early diagnosis of phenylketonuria, a severe neurodegenerative disorder of phenylalanine metabolism which if diagnosed early can have excellent outcomes. Guidelines for the introduction of additional conditions to newborn screen required that the condition was sufficiently common in the population to merit screening, that it was treatable and that the cost of diagnosis was not prohibitive. Additional conditions added to the screen included congenital hypothyroidism and congenital adrenal hyperplasia. The recognition of medium-chain acyl-CoA dehydrogenase deficiency coupled to the advent of tandem mass spectrometry as a diagnostic tool allowed for the inclusion of many more conditions into screening programs with as many as 50 disorders now being screened for on a single blood spot. Some of the additional disorders do not fit the original criteria for inclusion as they are very rare and treatments are not fully established. However, recent data is demonstrating improved outcomes for many of these conditions. Presymptomatic diagnosis by newborn screening is also an excellent mechanism for determination of disease frequency in the population and has been used to identify the most common conditions prevailing in specific populations. This presentation will discuss the history, current state and future of newborn screening for metabolic diseases and report on clinical outcomes measures of some conditions identified by the screening process.