PART I

POPULATION SCREENING: ISSUES, REALITIES AND POSSIBILITIES
A – Introduction: Expansion of Screening?

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All parents want their babies to be and remain in good health. They want—no, they expect—to be informed by the healthcare system about anything that conflicts with this expectation. Over the last 40 years, neonatal screening technology has evolved tremendously, and it is still making giant leaps. In the beginning, there were just simple tests for diseases like amino acidopathies, phenylketonuria being the classic example. The fact that the PKU phenotype may be the result of a variety of genotypes was discovered years later.

The success of the Guthrie bacterial inhibition assay using filter paper blood samples was the key to the expansion of screening programmes. At first, there was the addition of one other disease, congenital hypothyroidism (CH), which was depicted as being as simple as PKU but in fact has been shown to have various ethiologies. PKU and CH were and are still important because of their impact on lifelong suffering, if undetected, and their relatively easy means of treatment. For these reasons, virtually all neonatal screening programmes have started with those two diseases, and many still are limited to them. Advances in biochemical techniques in the 1970s and 1980s made it possible to detect many more diseases in the same blood spot material. Many of them are relatively easy to treat and simple to explain to parents and professionals.