Brazil-China Innovation Dialogue

Frontiers in Medicine and Public Policy Challenges II

How to implement precision healthcare in the SUS

Neonatal screening: How expanded should it be?

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Since neonatal screening has been implemented for phenylketonuria in the 60's of last century, it has been considered a very successful program and has continuously expanded both in the geographies covered and in the number of conditions screened for. Currently, most states of the USA, several countries in Europe and other regions screen for over 60 diseases. The question that frequently arises is: if we could do, should we do? In the beginning of the neonatal screening era, a document published by WHO (Wilson and Jüngner, 1968) established 10 principles that should be followed to consider a disease as a candidate for neonatal screening: 1) The condition sought should be an important health problem; 2) The natural history of the condition, including development from latent to declared disease, should be adequately understood; 3) There should be a recognizable latent or early symptomatic stage; 4) There should be a suitable test or examination; 5) The test should be acceptable to the population; 6) There should be an agreed policy on whom to treat as patients; 7) There should be an accepted treatment for patients with recognized disease; 8) Facilities for diagnosis and treatment should be available; 9) The cost of case-finding (including diagnosis and treatment of patients diagnosed) should be economically balanced in relation to possible expenditure on medical care as a whole; 10) Case-finding should be a continuing process and not a "once and for all" project. The recent advances in sequencing provide potential to include thousands of genetic diseases in these programs, as the use of genetic panels that include around 400 treatable diseases are being proposed as a platform to expand neonatal screening, and even the use of whole genome sequencing as a neonatal screening tool is also being considered. The fact of more expanded screening panels may be available to a portion of the population which subscribes private health insurance policies has an impact in the equity of health provision. The impact of neonatal identification in the public health system of a substantial number of conditions which are new to neonatal screening and still do not have the natural history completely understood and have disease modifying treatments that have high cost and/or sometimes questionable results should be discussed and the society should agree in a global policy applicable to all babies.