"It's been a frustrating process," says Professor Gaspar, who is also chief scientific officer at Orchard Therapeutics, which is designing gene therapies for SCID. "We need to get this implemented as soon as possible because this has the potential to save lives."

Need evidence to screen for more conditions

Hard evidence is difficult to come by with rare diseases, says Maximilian Zeyda, technical director of Austria's newborn screening lab, because data is intrinsically scarce. Austria screens for 28 diseases based largely on recommendations from a panel of specialist paediatricians.

"There is not so much hard scientific evidence and much more evidence by experience," he says, adding that newborn screening is an area where expert opinion should carry more weight.

Finding a balance is tricky though, says Dr Tarini. In the United States, the fragmented medical system means there's little information on cost effectiveness and public health impact for the various state genetic screening programmes, which makes informed debate difficult.

The bias should be towards including tests, she says, but going forward there's a need for robust data collection to enable educated reassessments. "This gives you the benefit of action with re-evaluation," says Dr Tarini. "We have not generally done that in a systematic way."

Genomics set to shake up newborn screening

The cost of unraveling someone's genetic code has fallen dramatically in recent years. This is opening up the tantalising prospect of screening babies for genetic diseases using DNA sequencing.

Robert Green, medical geneticist at Brigham and Women's Hospital and Harvard Medical School, is running the first US trial of newborn genomic sequencing. Half of the 325 babies in the programme have had their genomes sequenced and early results are promising.

Genetic predictions can be imprecise because many conditions are caused by complex gene interactions, which has raised concerns of unduly worrying parents. There are also ethical issues around detecting diseases that appear later in life.

“If we identify babies with predispositions to become ill in adult life, you remove their right not to know,” says Professor Jim Bonham, consultant at Sheffield Children’s Hospital.

But Dr Green says all medical diagnosis deals with risk factors. High blood pressure is only indicative of heart problems, it doesn’t guarantee them, for example. Both he and Professor Bonham agree that the challenge is finding better ways of conveying this information.

Also found in

#GENES #GENETICS #DISEASES #CHILDREN #DISEASE

SPECIAL REPORT