Choosing conditions, genes, and variants: Four guiding principles

Genomics England's Newborn Genomes Programme is delivering a research study to explore the benefits, challenges, and practicalities of sequencing the genomes of newborn babies.

A key task for the Programme is to establish which conditions we should look for, and feed back to the parents of newborns who take part in the study. We've worked with patients, parents, members of the public, healthcare professionals, and researchers to establish four principles to guide this decision.

The genetic condition and variant(s) will be included in our Programme if...

Principle A

There is strong evidence that the genetic variant(s) causes the condition and can be reliably detected.

To satisfy this principle: Where appropriate, there may be a confirmatory test that can establish whether or not the child has the condition

Principle B

A high proportion of individuals who have the genetic variant(s) would be expected to have symptoms that would have a debilitating impact on quality of life if left undiagnosed.

To satisfy this principle: Symptoms should have a debilitating impact. The impact should include considerations such as the testimony of patients and families affected including social and environmental factors, and QALYs where available.

Principle C

Early or pre-symptomatic intervention for the condition has been shown to lead to substantially improved outcomes in children, compared to intervention after the onset of symptoms.

To satisfy this principle:

- The intervention would normally be initiated in early childhood (by age 5).
- The intervention available could either cure, delay, or modify the course of the condition.

Principle D

Conditions screened for are only those for which the interventions are equitably accessible for all.

To satisfy this principle: Agreement from relevant NHS clinical, commissioning and other regulatory bodies is required.

We'll be cautious and conservative in our initial approach, to ensure our processes work and that families receive safe and effective care. We also know that we won't be able to find every child with each condition we look for, and this is something we'll explore further through our research study. For updates on how our work is progressing, or if you have any questions or suggestions, please contact: **ge-newborns@genomicsengland.co.uk**