New opportunities to improve the diagnosis of children with a rare disease

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After initial research, the Commission identified numerous barriers to diagnosis of rare diseases and divided those key barriers into three themes or “Tracks.”

The Commission then developed technology-enabled recommendations across each Track that aim to address those barriers and accelerate the time to diagnosis.

These recommendations are supported by global policy recommendations.
## Technology pilots

The Global Commission is supporting pilot projects, which aim to provide a proof-of-concept for distinct initiatives that lend themselves to near-term scale.

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Multifactorial machine learning to recognize symptom patterns

- The Global Commission is leveraging the power of machine learning. This collection of digital technologies integrates with EMR systems to support primary care physicians and pediatricians by recognizing symptom patterns that they may not immediately associate with a rare disease, expediting patient diagnosis.

- Three areas of focus: (1) medical records data, (2) patient-reported data, and (3) integration with genomics
  - Current pilot with Hospital La Paz de Madrid in Madrid, Spain