Meet Luca,* an infant admitted to the NICU at birth

Luca was experiencing respiratory distress and hypoglycemia at birth.

He was immediately transferred to the NICU for monitoring and ongoing care for respiratory distress. Upon initial examination, microcephaly and mild hypotonia were noted and a heart murmur was identified.

Follow up investigations included a brain ultrasound, chest x-ray, and echocardiogram. The brain ultrasound was normal, but the other tests identified an atrial septal defect.

Hospital A

Hospital A's genetic testing protocol uses rapid genome sequencing for critically ill babies in the NICU whose symptoms cannot be fully explained by prematurity, trauma, infection, or who meet the guidelines and recommendation criteria.

Hospital B

Hospital B follows internal protocols, with genetic testing ordered on a case-to-case basis in the NICU. Rapid genome sequencing can only be ordered after a genetics consult.

Hospital protocols can impact patient outcomes. A **stepwise testing approach** may result in a **significantly delayed diagnosis** for children with an underlying genetic condition.

* Fictionalized case studies for illustrative purposes only.

GeneDx

Hospital A

Given the findings of hypotonia, microcephaly, and atrial septal defect, his doctor suspected a genetic condition and ordered rapid genome sequencing the following day.

Results were delivered in 5 days, revealing a *KMT2D* de novo variant, which provided a diagnosis of Kabuki syndrome.

5 days

For Luca and his family, rapid genome sequencing provided an accurate and timely diagnosis.

Luca's early genetic diagnosis:

- Informed prognosis.
- Guided targeted evaluations for clinical symptoms associated with Kabuki Syndrome (i.e., specific growth charts).
- Implemented referrals for symptom monitoring (e.g., hearing loss, immune dysfunction, eye abnormalities) and early interventions, if needed.
- Provided the family with support and resources through the Kabuki Syndrome Foundation and parent groups.

Hospital B

After three days in the NICU, the hypoglycemia and respiratory distress resolved, and Luca was discharged.

He was referred for an outpatient visit with the cardiology clinic regarding his septal defect; the cardiologist monitored Luca through his early years.

1 year

Luca's family also reported feeding difficulties and a delay in his developmental milestones. His pediatrician monitored his slow yet steady development and noted that he started to fall off his growth curve at 1 year of age. Luca also had chronic ear infections and eventually developed hearing loss.

At age 4, his pediatrician ordered a microarray and *FMR1* testing for fragile X. These were both negative. His pediatrician referred Luca to the local genetics clinic given his atrial septal defect, microcephaly, short stature, hearing loss, and developmental delay. Luca had also been recently diagnosed with scoliosis and his doctor noted possible dysmorphic features.

5 years

At age 5, Luca had genome sequencing ordered by the genetics clinic. The results revealed a *KMT2D* de novo variant, which provided a diagnosis of Kabuki syndrome.

Transform patient care. Choose rapid genome as a first-line test for critically ill babies.

Hospital A's protocol of using rapid genome sequencing for critically ill babies who meet clinical criteria gave Luca answers in his first week of life—rather than years later, making all the difference for his family and his healthcare.

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