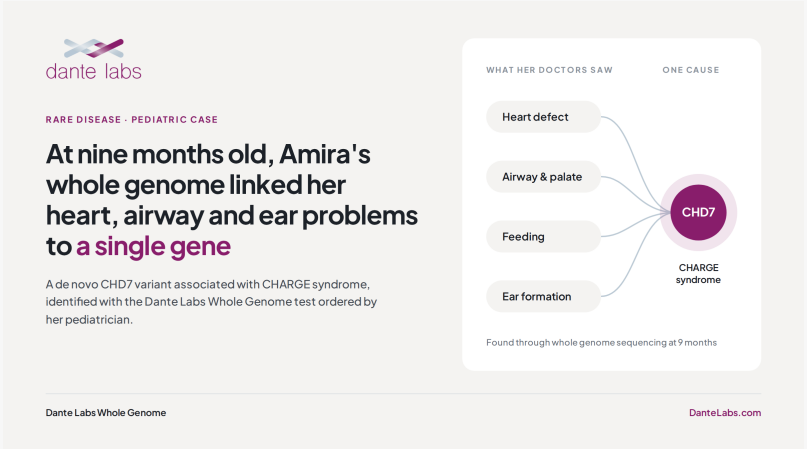
Nine Months Old, a Heart Defect and No Explanation, Until a Dante Labs Whole Genome Identified Amira's CHARGE Syndrome

A de novo pathogenic variant in the CHD7 gene, found by Dante Labs Genome gave an infant with heart, airway, feeding and ear complications a clear care pathway

NEW YORK, NY, UNITED STATES, September 29, 2026 / EINPresswire.com/ -- Amira was only nine months old when her parents found themselves trying to make sense of a list of medical problems that no single explanation seemed to connect, from a congenital heart defect and constant drooling to a palate that could not properly seal off her nasal airway, ears that had formed differently, and a difficult start to life marked by low oxygen levels at birth and an unusually weak cry. Dante Omics AI announced today that a Dante Labs Whole Genome test, ordered by Amira's pediatrician, through DanteLabs.com, identified a pathogenic variant in the CHD7 gene associated with CHARGE syndrome, a rare developmental disorder that affects several organ systems at once and can be life-threatening in infancy when its features are not recognized and managed together.



Dante Labs Genome for Rare Diseases



Dante Labs Genome for Rare Diseases - Wide

Each of Amira's symptoms could, on its own, have sent her family to a different specialist, from cardiology and ear, nose and throat surgery to feeding and hearing clinics, and managing them one at a time carried the risk that the thread connecting them would stay

> The answer was in Amira's DNA. The Dante Omics technology identified the answer and provided a clear clinical pathway with specialists."
>
> *Andrea Riposati*

hidden for months or years. Dr. Aman recognized that the combination of findings pointed toward an underlying genetic cause and chose to look at Amira's complete genome in a single test, rather than ordering a series of targeted panels that each examine a limited set of genes and can leave families waiting through one inconclusive result after another.

Amira's DNA was collected with a non-invasive kit designed to be suitable even for newborns and sequenced through the Dante Labs Whole Genome test, which reads the full genome at clinical-grade 30X coverage, including the intronic regions that sit between the protein-coding sections of each gene. The data was analyzed by Dante Omics platform. The analysis identified a heterozygous splice donor variant in intron 27 of CHD7, a change that disrupts the way the gene's instructions are assembled, and the evidence supported a pathogenic classification that aligned closely with Amira's clinical features, giving Dr. Aman a genetic explanation that tied together what had previously looked like separate problems.

When Amira's parents chose to be sequenced as well, neither of them carried the variant, which indicates that it arose de novo, as most CHD7 variants do, rather than being inherited from either parent. For the family, that result carried its own significance, because it answered the question of where Amira's condition came from and gave their clinicians the information needed to counsel them about the chance of recurrence in future pregnancies.

CHARGE syndrome is estimated to affect approximately 1 in 10,000 newborns, and its name is an acronym for several of its characteristic features, namely coloboma of the eye, heart defects, atresia of the choanae (a narrowing or blockage of the nasal passages), growth retardation, genital abnormalities and ear abnormalities. Because the pattern of features varies considerably from one child to another, many infants are identified only after a long sequence of specialist visits, which is why a genetic finding early in life allows a care team to anticipate concerns that have not yet appeared, including vision, hearing, growth and hormonal development, instead of discovering them one by one.
With a name for her condition, Amira's care team can now move from addressing each symptom in isolation to a coordinated plan built around the known course of CHARGE syndrome, and her parents, who spent the first months of her life trying to understand why so many things were going wrong at once, finally have an answer they can build on together with her doctors.

"Physicians need real answers and advanced tools to enable personalized medicine. The Dante Labs Whole Genome test keeps suppoting tens of thousands of individuals and their doctors," said Andrea Riposati, CEO of Dante Omics AI. "Amira's pediatrician asked the right question. Her genome and technology had the answers. We built Dante Omics so that any family, anywhere, living inside an unanswered medical question can reach that answer while it can still shape a

child's care."

Dante Labs supports people, pediatricians and hospitals with advanced genomics solutions. Individuals, families and physicians can order the Dante Labs Whole Genome test at DanteLabs.com.

Amira's name and Dr. Aman’s names have been changed to protect Amira’s and her family's privacy.

Key Facts
1) Patient: Amira, a nine-month-old girl with congenital heart disease, sialorrhea (excessive drooling), velopharyngeal incompetence affecting her palate and airway, ear malformations, and hypoxia at birth.
2) Test used: the Dante Labs Whole Genome test, ordered by her pediatrician through DanteLabs.com.
3) Finding: a heterozygous splice donor variant in intron 27 of the CHD7 gene, classified as pathogenic and associated with CHARGE syndrome.
4) Inheritance: both parents were sequenced and neither carries the variant, which indicates a de novo origin, as in most CHD7 cases.
5) Condition: CHARGE syndrome is a rare multi-system disorder affecting approximately 1 in 10,000 newborns, named for coloboma, heart defects, choanal atresia, growth retardation, genital abnormalities and ear abnormalities.
6) Outcome: a single genetic explanation for symptoms across several organ systems, allowing her care team to coordinate specialist follow-up around the known course of the condition.

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