BillionToOne UNITY Screen's Unique Approach to Cystic Fibrosis Prenatal Screening Allows More Treatment Options for Expecting Families

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Largest study of CF prenatal screening to date showed UNITY Fetal Risk™ Screen identifies all CF cases in study population

MENLO PARK, Calif., Sept. 15, 2025 /PRNewswire/ -- **BillionToOne**, Inc., a molecular diagnostics company with a mission to create powerful and accurate tests that are accessible to all, today announced a new publication demonstrating the superior performance of its UNITY Fetal Risk™ Screen in identifying fetuses at high risk for cystic fibrosis (CF). The comprehensive retrospective study, published in the peer-reviewed *Journal of Cystic Fibrosis* titled, **Routine cell-free DNA prenatal screening identifies pregnancies at high risk for cystic fibrosis that may benefit from fetal therapy**[1], analyzed over 100,000 consecutive general-risk pregnant patients and represents the largest validation of cell-free DNA screening for CF to date. The study demonstrated 100% sensitivity in identifying high risk CF pregnancies and found that 95% of pregnancies identified as high risk carried CF variants eligible for *CFTR* modulator therapy, i.e., drugs that help correct the protein defect that causes CF, providing expecting families with more treatment options.

The study identified 2,587 individuals who were carriers of cystic fibrosis, encompassing 296 distinct *CFTR* gene variants, not limited to the commonly recognized F508Del mutation. UNITY demonstrated superior detection capabilities by identifying a broad range of genetic variations, including both homozygous and compound heterozygous cases, without requiring a paternal sample, which is often difficult to obtain in the prenatal setting.

UNITY achieved exceptional clinical performance with 100% sensitivity, meaning no CF-affected pregnancies were missed by the screening. In addition, all pregnancies assigned the highest risk category (9-in-10 chance) with available outcome data were confirmed to be affected through neonatal outcomes.

Of particular clinical significance, 95% of pregnancies identified as high-risk (1-in-4 chance or

greater) carried CF variants eligible for *CFTR* modulator therapy, breakthrough therapies that can modify the course of cystic fibrosis. With testing available as early as 9 weeks of gestation and median TAT of 8.7 days, UNITY may enable timely confirmation and potential initiation of emerging *in utero CFTR* protein modulator medicines that show promise in case reports for improving neonatal outcomes.

"The ability to identify CF-affected pregnancies early in gestation, particularly those eligible for CFTR modulator therapy, represents a transformative advancement," said Dr. Aaron Trimble, M.D., pulmonologist and one of the authors of the study. "This could enable access to promising *in utero* treatments and fundamentally change outcomes for babies with cystic fibrosis."

The research demonstrated UNITY's advantage over traditional sequential carrier screening methods, which require testing both parents and often result in incomplete risk assessments due to logistical challenges. UNITY's single-sample approach uses the pregnant individual's blood, eliminating barriers associated with partner availability. The test provides personalized quantitative fetal risk assessments ranging from 1-in-5,000 to 9-in-10, compared to the fixed population risk to a maximum 1-in-4 risk estimate from traditional screening.

BillionToOne's proprietary Quantitative Counting Templates™ (QCT™) technology enables precis quantification of fetal cell-free DNA variants. This allows UNITY to determine fetal genotypes and provide accurate risk predictions for autosomal recessive conditions like cystic fibrosis. This technology addresses the long-standing challenge of detecting recessive genetic conditions through non-invasive prenatal testing, opening new possibilities for early intervention and treatment.

"This landmark study validates our approach to comprehensive CF screening without requiring a partner sample, addressing a critical gap in traditional carrier screening where more than half of partners never complete testing when the pregnant mother is identified as a carrier," said Dr. Haywood Brown, Chief Medical Officer, Prenatal at BillionToOne. "Most importantly, this gives families early insights they need to prepare for their child's care and explore new therapeutic options that could significantly improve their child's quality of life when CF is detected."

About BillionToOne

Headquartered in Menlo Park, California, BillionToOne is a precision diagnostics company on a mission to make molecular diagnostics more powerful, efficient, and accessible for all. The company's patented Quantitative Counting Templates™ (QCT™) molecular counting platform is th only multiplex technology that can accurately count DNA molecules at the single-molecule level. For more information, visit **www.billiontoone.com**.

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[1] J. Wynn et al., Journal of Cystic Fibrosis, **https://doi.org/10.1016/j.jcf.2025.08.004**

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