Study uncovers cystic fibrosis screening limitations before or during early pregnancy

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New research from Australia's national science agency, CSIRO, has found that reproductive carrier screening tests, used before or in early pregnancy to identify inherited genetic conditions, potentially miss up to

10 percent of cystic fibrosis (CF) causing variants in Australia.

The study, which involved an analysis of data from 3958 people with CF between 1970 and 2020, also found that East Asian people are disproportionately affected by this testing limitation.

CF is an inherited disorder that causes severe damage to the lungs, digestive system, and other organs in the body. Reproductive carrier screening tests are important as the CF-causing gene is passed down from both parents who usually do not have the disease themselves.

Conducted by CSIRO scientists in partnership with Pathology Queensland and Australian gene testing lab Genepath, the research was recently published in *Prenatal Diagnosis*, and provides key insights to assist in improving reproductive carrier screening.

Letitia Sng, CSIRO research scientist and co-lead author on the study, said European and Australian guidelines for CF reproductive carrier screening recommend testing a small number of high frequency CF-causing variants.

"Our research found that the current approach of limiting reproductive carrier screening tests to a small number of high frequency CF variants impacts the effectiveness of screening," Dr. Sng said.

"This reduced effectiveness disproportionately affects people of East Asian ethnicity, indicating a potential failure of the current recommended testing protocols to include variants arising in certain ethnicities.

"This may be a result of the changing demographics of the Australian population over the last 50 years, with a significant increase in immigration to Australia from non-European countries such as China

and India," she said.

Bennett Shum from Genepath said along with findings about the failure of current screening to detect certain CF variants, they were also able to confirm that in Australia, 1 in 34 individuals carry the gene for CF (known as CTFR), making carrier screening useful for people looking to have babies.

Dr. Sng said given the increasing cost-effectiveness and comprehensiveness of CFTR gene analysis techniques, their results point to a need for the revision of reproductive carrier screening approaches in Europe and Australia to account for multi-ethnic populations.

"More research is needed into existing inequalities in current screening recommendations in CF and other genetic diseases to ensure the accuracy of carrier screening for all," Dr. Sng said.

More information: Bennett O. V. Shum et al, The inequity of targeted cystic fibrosis reproductive carrier screening tests in Australia, *Prenatal Diagnosis* (2022). DOI: 10.1002/pd.6285

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