Screening during IVF for inherited diseases can greatly reduce costs of care

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Credit: AI-generated image (disclaimer)

For prospective parents who are carriers of many inherited diseases, using in vitro fertilization along with genetic testing would significantly lower health care expenditures, according to researchers at Stanford Medicine.

Preimplantation genetic diagnostic testing during IVF, or PGD-IVF, is now being used to screen for single-gene defect conditions such as cystic fibrosis, sickle cell disease and Tay-Sachs disease, along with nearly 400 others.

The problem is that the high cost of IVF—and the lack of coverage by all but one state Medicaid program, that of New York—makes it unavailable to millions of people at risk. The majority of private employer health benefit plans also do not cover IVF.

"We have the capability to prevent these diseases with advances in prenatal genetic diagnostic testing combined with IVF," said Kevin Schulman, MD, director of the Clinical Excellence Research Center at Stanford Medicine. "But many benefit designs, especially for public programs such as Medicaid, have not kept pace with these advances."

Although there is a reluctance to add IVF as a benefit to address infertility alone, PGD-IVF would offer insurance organizations a cost-saving intervention when compared with the costs of lifetime care for patients with a single-gene-defect disease.

Stanford researchers set out to demonstrate what that cost savings would be. A paper based on their findings was published in the June 13 issue of *Health Affairs Forefront.*

"We conducted our study to see if it would be in the economic interest of these programs to include PGD-IVF in their benefit," said Schulman, a professor of medicine. "We showed that it was."

The costs of sickle-cell disease

The researchers compared the costs of the PGD-IVF intervention versus the costs of care for patients with the most common single-gene-defect

disease: sickle cell disease.

Affecting near 100,000 Americans a year, sickle cell disease affects 1 out of every 365 Black American births. Despite improvements in therapies, a sickle cell disease diagnosis means a lifetime of constant care and high costs: monthly blood transfusions, penicillin, opioids and hospitalizations. The researchers calculated that these interventions result in an annual cost of between $6,636 and $63,436 more than a person without sickle cell disease, and average lifetime costs of $602,000, with estimates as high as $1.7 million.

The only cure is bone marrow transplant, known as hematopoietic stem cell transplantation, but that comes with a risk of severe infections, seizures and infertility. And the cost is high: between $150,000 and $250,000 per patient. Even more promising gene therapies under development to treat sickle cell disease are expected to have a price tag of more than $1 million.

On the other hand, the estimated cost of PGD-IVF to prevent the disease is $15,000 to $25,000.

"We tried to show there's an economic argument to investing early on, by avoiding incidence of the disease," said Aadit Shah, a Stanford medical student who coauthored the paper with Schulman and fellow medical student Wasan Kumar.

Medicaid picks up the bill for half of all sickle cell disease patients, so the public bears much of the financial burden for a lifetime of costly treatment for more than 55,000 new patients a year. In this respect, the researchers point out, the lack of Medicaid coverage for PGD-IVF leads to health inequity. If PGD-IVF is used to screen out sickle cell disease, there would still be a significant number of people receiving care, but the number of new cases would drop dramatically.

The new research may help health officials make a more informed decision about whether to add coverage for PGD-IVF for single-gene-defect diseases. "In the wake of the COVID-19 pandemic and a renewed discussion about health equity," the paper reads, "adding PGD-IVF as a Medicaid benefit seems to have a compelling clinical and economic justification."

More information: Wasan M. Kumar et al, Benefit Determination For Single Gene Defect Diseases: A Paradigm Shift, *Health Affairs Forefront* (2023). DOI: 10.1377/forefront.20230608.910810

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