

The use of preimplantation genetic diagnosis (PGD) is expanding rapidly. Yet, because of the absence of data collection systems and the fragmented regulatory system, important quality assurance and data collection mechanisms currently are unavailable for PGD. PGD is much more technically difficult than other forms of genetic testing, yet even the general laboratory quality standards under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) are not currently being applied to PGD labs. CLIA defines a "clinical laboratory" as a laboratory that examines materials "derived from the human body" in order to provide "information for the diagnosis, prevention, or treatment of any disease or impairment of, or the assessment of, human beings." Though recommended by a government advisory committee, the Center for Medicaid and Medicare Services (CMS) has not made embryo laboratories subject to CLIA regulations. Some within CMS worry that including PGD under CLIA would require CMS to take the position that an embryo meets the legal definition of a human being. It is unclear whether this concern is well founded since neither the agency nor any court has had occasion to formally address it. Nonetheless, the net result is that most PGD laboratories appear to be operating without CLIA certification.

In addition to gaps in oversight of PGD laboratories, there also are gaps in the data collection and monitoring of PGD. While the U.S. federal government currently collects data on IVF, there is no systematic collection of information on PGD. Thus, the number of PGDs performed, the type of tests being used, and the outcomes are largely unknown. In the absence of these data, it is impossible to define fully the risks associated with the procedure. This information is critical for patients to make informed decisions, for providers to improve the quality of care, and for policymakers to accurately assess whether PGD requires further oversight.

In the absence of comprehensive data, it is difficult to know the frequency and impact of misdiagnosis. But errors in PGD can be devastating. For example, a family with a child affected by Fanconi anemia, a rare but deadly blood disease, recently decided to have PGD in order to have a baby that would be a genetic match to their sick child and thus be able to provide a matched bone marrow or stem cell transplant from umbilical cord blood. During the PGD, doctors told the mother that they were transferring two embryos into her womb that would be a genetic match and would not have the Fanconi anemia genetic mutation. Nine months later twins were born; both have been diagnosed with Fanconi anemia. In a series of interviews with PGD patients conducted by the Center, three of seven women interviewed who became pregnant following PGD had had a misdiagnosis.

The Center has noted a dearth of data on PGD and in earlier work presented options for instituting national reporting requirements for PGD and studies on the health outcomes following PGD. Similarly, in its report *Reproduction and Responsibility*, the President's

Council on Bioethics noted the need "to develop a clearer sense of the uses to which genetic screening and selection are being put and the degree and frequency of use. Such basic information is for the moment difficult to come by, and we may not have the kind of understanding of the status quo that would be required to make further judgments regarding regulation." The Council concluded by recommending the development of specific reporting requirements for PGD.

The Center and others have long called for well-controlled and well-designed clinical studies looking at PGD's efficacy and safety. We have spearheaded an effort with the American Society of Reproductive Medicine, the Society for Assisted Reproductive Technology, and the PGD International Society to establish a U.S. PGD database. This database, now nearing fruition, will permit robust research and enable studies that can track outcomes over time.

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