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Case report on genetic diagnosis of fatal disorder in embryos before pregnancy

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Genetic testing of embryos for a fatal inherited neurodegenerative disorder allowed a woman to selectively implant two mutation-free embryos and conceive healthy twins, what researchers call the first case of in vitro fertilization (IVF) with preimplantation genetic diagnosis (PGD) to prevent genetic prion disease in children, according to a case report in *JAMA Neurology* by Alice Uflacker, M.D., of Duke University, Durham, N.C., and colleagues.

The 27-year-old woman is a carrier of the F198S mutation for Gerstmann-Sträussler-Sheinker syndrome (GSS), a fatal neurodegenerative disorder linked to abnormal prion protein folding. There is no known cure and the illness is fatal, according to the case background.

During IVF treatment, 12 of the 14 oocytes (egg cells) retrieved from the woman were fertilized and six mutation-free embryos were identified. The patient opted to have two embryos transferred and three remaining viable embryos frozen through cryopreservation.

The two embryos successfully implanted and the woman delivered twins by Cesarean section at 33 weeks and five days of gestation. By age 27 months, the twins had reached communication, social and emotional developmental milestones on schedule.

"IVF with PGD is a viable option for couples who wish to avoid passing the disease to their offspring. Neurologists should be aware of PGD to

be able to better consult at-risk families on their reproductive choices," the authors conclude.

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