Genetics

Preimplantation genetic diagnosis

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Preimplantation genetic diagnosis (PGD) involves producing fertilized oocytes in vitro, which are then cultured until they reach the 8-cell blastomere stage. A single cell is removed from the embryo and tested for a specific genetic disease or chromosome abnormality. The embryos that have negative results for the disease are transferred to the woman's uterus. If the embryo implants and becomes a viable pregnancy, it is expected to be unaffected by the tested disease.

Bottom line. A couple who wishes to avoid both prenatal testing and making decisions about terminating an affected pregnancy but who also wishes to avoid having a child with a specific genetic disease might be interested in PGD; they should receive genetic counseling beforehand. Patients are responsible for covering the substantial costs of the in vitro fertilization and PGD procedures, and they should be aware of the possible need for multiple cycles and the pregnancy success rate of 30% to 45%.

The complete *Gene Messenger—Preimplantation Genetic Diagnosis* by the GenetiKit research team is available on **CFPlus.*** Past Gene Messenger articles can be accessed online at **www.cfp.ca.** On the home page, click on **Collections** in the left-hand menu, then click on **Genetics.**

Competing interests
None declared

The **GenetiKit** research team, a group of family physicians, genetic counselors and geneticists, designed the Gene Messenger series to provide practical information to help family physicians and their patients make informed choices about rapidly emerging genetic discoveries. The series is a collection of up-to-date, definitive, short reviews on genetics topics that have made headlines, and offers recommendations regarding referral for genetic services or testing.

Acknowledgment
Funding was provided by the Canadian Institutes of Health Research.

GENE MESSENGER

For more information on genetics topics, see **www.mtsinai.on.ca/FamMedGen/**

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*The Gene Messenger on preimplantation genetic diagnosis is available at **www.cfp.ca.** Go to the full text of this article online, then click on **CFPlus** in the menu at the top right-hand side of the page.

Dermacase

Can you identify this condition?

Jakub Sawicki Benjamin Barankin MD FRCPC

A 75-year-old healthy man presents with multiple grouped open comedones and yellow waxy papules on his cheeks. They are asymptomatic. The condition had slowly progressed over the past 10 years.

The most likely diagnosis is

1. Cutis rhomboidalis nuchae
2. Sebaceous hyperplasia
3. Comedonal acne
4. Favre-Racouchot syndrome
5. Chloracne

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