Propionic Acidemia Genetics Part 2 (Prenatal Diagnosis)

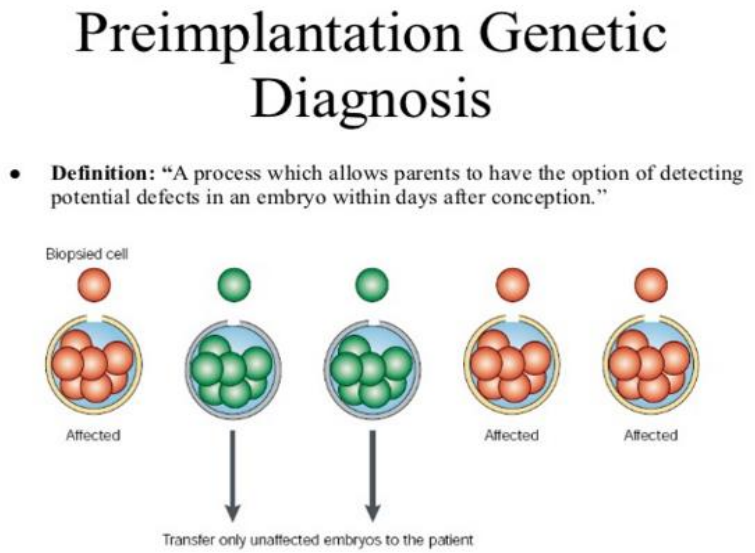
Prenatal diagnosis is a way for your doctor to determine if your baby has a certain condition, such as Propionic Acidemia, before the baby is born. In order to detect Propionic Acidemia in a baby, the laboratory needs to know the specific genetic mutations causing Propionic Acidemia in a family member. It can also test for more common genetic disorders, such as Down syndrome, which can occur in any pregnancy. Lastly, prenatal diagnosis can tell you the gender of the baby if that is information you want during your pregnancy. There are two traditional ways to undergo prenatal diagnosis.

The first method involves analyzing the DNA in cells from the placenta. The cells are gathered from a procedure called **chorionic villus sampling or CVS**. A CVS can be done two ways, one involves inserting a catheter through the cervix and the other is performed by inserting a thin needle through the abdomen. Both methods obtain a small sample of the placenta to perform the genetic test. A CVS can be performed from 10-13 weeks into the pregnancy. Results take approximately 7-10 days depending on the laboratory. There is a small risk of miscarriage associated with the procedure which varies from center to center, but generally <1% (<1/100).

The second method is called **amniocentesis** and analyzes the baby's DNA from cells present in the amniotic fluid (normal fluid that surrounds the baby in the womb). The procedure is performed by inserting a thin needle through the abdomen to obtain a sample of the amniotic fluid. An amniocentesis can be performed after 15 weeks into a pregnancy. Results take approximately 10-14 days to come back depending on the laboratory. There is a small risk of miscarriage associated with the procedure which varies from center to center but generally <0.5% (<1/200).

Both CVS and amniocentesis have the same degree of accuracy for genetic testing. The main difference between the two procedures is timing of when testing takes place during pregnancy and the difference in miscarriage risk, with risk associated with amniocentesis less than the risk associated with CVS.

There is a newer technology used for prenatal diagnosis called **pre-implantation genetic diagnosis (PGD).** Instead of testing an ongoing pregnancy, PGD works by testing DNA from embryos before they are implanted into the uterus. The embryos are created through standard **in vitro fertilization (IVF)**, the process where an egg is fertilized by a sperm in a laboratory instead of in the woman's uterus. At an early embryo state, 1 – 2 cells are safely taken from the embryo and used to analyze the DNA for the genetic disorder. The embryos without the genetic disorder are then implanted into the mother's uterus.



If you are interested in learning more about traditional prenatal diagnosis or preimplantation genetic diagnosis, we recommend that you meet with a genetic counselor to discuss these methods in greater detail.

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