

## Anorectal Malformations and Genetic Factors

[Anorectal malformations](#) are currently considered to be "multifactorial," meaning that there are many unknown factors involved in causing these malformations. Most likely, there are also many genes involved, but they have not yet been identified.

### More Information

[Genetics Study](#)

Investigations of the genetic components of different diseases have tended to focus on conditions that have an already established hereditary link and are determined by a single gene. That makes these conditions relatively easy to study. Anorectal malformations do not belong to that group of conditions and discoveries about possible genetic cause of anorectal malformation will probably have to await further advances in genetic research.

Some preliminary work has already been done. In 1999, researchers involved with the human genome project at National Institutes of Health sought our input and questioned us about our experiences treating anorectal malformations.

We have treated patients who are identical twins, one who has an anorectal malformation and the other patient who does not. This means that perhaps there are other factors, such as the varying environmental of the mother's womb, which may contribute to producing anorectal malformations. We also know that a couple that already has one child with an anorectal malformation has a 1 in 100 chance of having another child with this condition, whereas for the general population, the chance of having a child with an anorectal malformation is 1 in 4,000.

In addition, a specific type of defect known as a perineal fistula is known to occur more frequently in some families. A perineal fistula is an abnormal anal opening that is situated towards the front, rather than the back of the body. We have personally worked with families that have two or three children with perineal fistulas. Our records for the year 2000 included eight families that had two children with this particular defect.

## Contact the Colorectal Center at Cincinnati Children's

For more information or to request an appointment for the Colorectal Center at Cincinnati Children's Hospital Medical Center, please [contact us](#).