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# Genetic alteration linked with disorders of sex determination

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A variety of genetic factors are involved in sex determination. If something goes wrong with one of these factors, people who have a chromosome set that predicts they will be of one sex may develop as the other sex or have traits on the spectrum between the two sexes. There can be emotional and social stress associated with disorders of sex determination (DSD), and in many cases, infertility is an additional problem. Several genetic alterations that cause DSDs have been identified, and work continues in an effort to elucidate the cause in other individuals. Research published by Cell Press on December 2nd in the *American Journal of Human Genetics* reveals new information about how sex determination can go awry.

"The prevalence of DSDs is about one in a thousand—so fairly prevalent in a spectrum of birth defects that affect 2% of the population," explains the senior study author, Dr. Harry Ostrer from the New York University School of Medicine.

Some of the people participating in the study were women who had the chromosome set that predicted they would be males, and others were males who had problems with sex development. One of the families involved was first reported in the New England Journal of Medicine 40 years ago.

Dr. Ostrer, Alex Pearlman, and Johnny Loke at the NYU School of Medicine and collaborators Andy Greenfield from MRC Mammalian Genetics Unit in England, Andrew Sinclair and Stefan White at the

Murdoch Children's Research Institute in Australia, and Cedric LeCaignec and Albert David from the Centre Hospitalier Universitaire de Nante in France examined whether disruption of MAP3K1 could be responsible for the DSDs in their patients.

Several of the patients were found to have an alteration in their MAP3K1 gene, which is a member of the MAP kinase signaling pathway. "Work has shown that the MAP kinase pathway is involved in the balance of expression between genes involved in directing the undifferentiated gonad to become a testis or an ovary. Thus, we think that we've found an important switch in the 'battle of the sexes,'" explains Dr. Ostrer.

"The discovery is reassuring to the patients and their families that there is a cause and that their DSD was not serendipity," Dr. Ostrer continues. It also contributes to better management of the DSD because patients with an alteration in MAP3K1 can develop life-threatening tumors. "Removal of the gonads early in life is important for preventing cancer" Dr. Ostrer says.

Provided by Cell Press

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