9 May 2008

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DanceSalad

How a Genetic Man Is Actually a Woman

The Sox9 gene

We know that boys have XY sex chromosomes, while girls have XX. However, in some cases, newborn girls can be XY. A new research published in the Nature journal clarifies why some XY embryos, instead of being born as males, develop ovaries and evolve as girls: it's because of a gene called Sox9, involved in formation of the testes, and other two control genes. "There are a surprisingly large number of cases where this process goes wrong. This phenomenon could effect up to one in every 20,000 genetic males. Maybe one could treat some of these sex reversal or intersex cases after birth by manipulating whether Sox9 is active or not. This is all speculation but it's possible," said Robin Lovell-Badge, a biologist at London's MRC National Institute for Medical Research. When Sox9 is activated in a genetically female embryo (XX), male gonads will start to develop; if the gene cannot be activated in a male embryo (XY), the cells that form the gonads will develop ovaries. These phenomena were studied by the research team in genetically engineered mice. In this way, the researchers found that Sox9 was under the control of two other genes. When these are mutated (not active), Sox9 remains deactivated and the embryo grows ovaries. One of these genes is located on the Y chromosome, present only in males. "Scientists must now determine how Sox9's activity actually leads to the creation of testes," said Richard R. Behringer, a geneticist at the University of Texas's M.D. Anderson Cancer Center in Houston. This discovery is most likely to apply to humans, too, and could be useful in detecting "male" embryos prone to develop into girls. "That's important, because those people are at higher risk for ovarian tumors. With further analysis, scientists may determine ways to reassign gender later in life, perhaps for cases of sex reversal or perhaps even for individuals who want to undergo sex changes," said Lovell-Badge.