

# Double identities lie behind chromosome disorders

July 8 2007

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Chromosome disorders in sex cells cause infertility, miscarriage and irregular numbers of chromosomes (aneuploidy) in neonates. A new study from Karolinska Institutet published in the scientific journal *Nature Genetics* shows how chromosome disorders can arise when sex cells are formed.

Sex cells contain a control station for monitoring the mechanism that ensures that the correct numbers of chromosomes are distributed during cell division. Scientists have now shown that there is an alternative distribution mechanism in female sex cells that cause chromosome disorders. Aberrant chromosomes orientate themselves like normal chromosomes, and this ability to adopt double identities protects them from detection by the control centre.

“We believe that this new fundamental mechanism can help to explain why chromosome disorders are so common in female sex cells,” says Professor Christer Höög, leader of the study.

The research might eventually lead to new medical treatments able to reduce the risk of foetal damage.

Over 0.3 per cent of children are born with some kind of chromosome disorder. Most develop Downs Syndrome, or obtain the wrong number of sex chromosomes and develop Turner’s or Klinefelter’s syndrome. Turner’s syndrome only occurs in females and is caused when one of the two X chromosomes is missing. Girls with Turner’s have arrested

development and if no treatment is given do not enter puberty. Klinefelter's syndrome affects males, who receive an extra X chromosome. Symptoms include concentration difficulties, poor motor skills and infertility.

Publication: "Bi-orientation of achiasmatic chromosomes in mammalian MI oocytes contributes to aneuploidy", Anna Kouznetsova, Lisa Lister, Magnus Nordenskjöld, Mary Herbert, Christer Höög, *Nature Genetics*, 8 July 2007

Source: Karolinska Institutet

Citation: Double identities lie behind chromosome disorders (2007, July 8) retrieved 9 April 2024 from <https://medicalxpress.com/news/2007-07-identities-chromosome-disorders.html>

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