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Chromosome X unraveled

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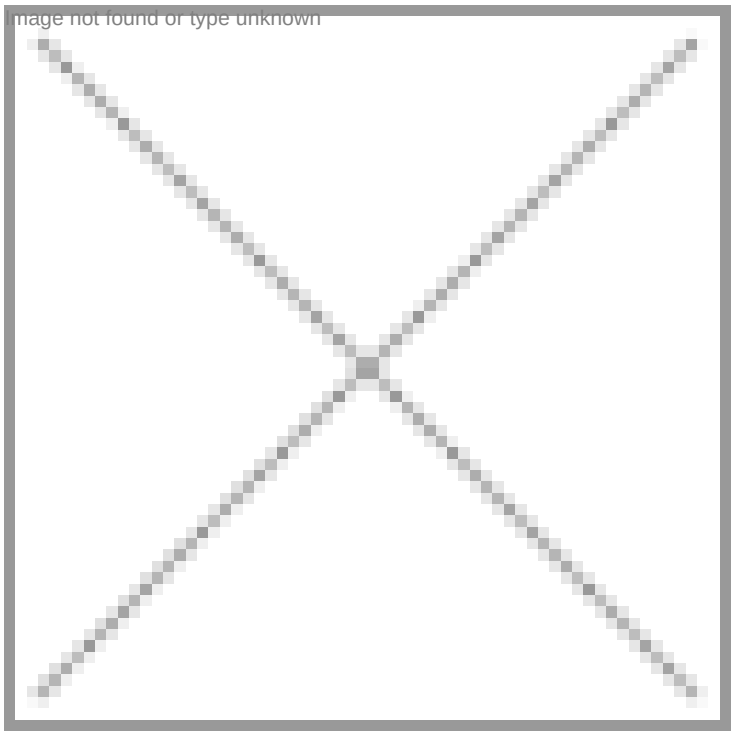

An international team led by the Wellcome Trust Sanger Institute, Cambridge, UK has succeeded in coming up with complete analysis of the X chromosome.

"From studying such genes, we can get remarkable insight into disease processes. We have also gained a deep insight into the evolution and biology of the whole chromosome. We can see the way evolution has shaped the chromosomes that determine our gender to give them their unique properties", said Mark Ross, project leader at the Wellcome Trust Sanger Institute.

In humans and other mammals, sexual identity is governed by a pair of chromosomes known as X and Y. Every female has two X chromosomes, inherited from both parents, while all males have one X from the mother and one Y chromosome from the father. As a result, any defects in genes on the X chromosome are often apparent in males because the Y chromosome does not carry corresponding genes to compensate. Women who carry such defective genes are usually protected by their backup copy of the X.

The X chromosome was originally named X for "unknown", remaining an oddity that has puzzled geneticists for centuries. The complete sequencing has enabled the researchers to construct a remarkably detailed portrait of the X's past. The study has shown how both the chromosomes, X and Y evolved from an "ordinary" pair of identical chromosomes. After the X and Y seceded, mutations in genes on Y made it the male determining chromosome. Over time Y has disintegrated to a shadow of its former self.

More than 300 diseases have been mapped to the X chromosome, by far the highest proportion of any chromosome, including Duchenne Muscular Dystrophy and Haemophilia, in which the blood fails to clot properly.

The primary sequence of this chromosome is an advanced starting point for exploring the mysteries of evolution and development. As Professor Allan Bradley, Director, Wellcome Trust Sanger Institute, remarked, "We often describe the results of sequencing as a 'catalogue of human genes'. The results of projects such as the finished X chromosome are so much more than that. They are the forces that will drive biomedical advance in the UK and around the world." n