

ESCO2 Gene

Subjects: **Genetics & Heredity**

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Establishment of sister chromatid cohesion N-acetyltransferase 2

genes

1. Normal Function

The *ESCO2* gene provides instructions for making a protein that is important for proper chromosome separation during cell division. Before cells divide, they must copy all of their chromosomes. The copied DNA from each chromosome is arranged into two identical structures, called sister chromatids. The *ESCO2* protein plays an important role in establishing the glue that holds the sister chromatids together until the chromosomes are ready to separate.

2. Health Conditions Related to Genetic Changes

2.1 Roberts Syndrome

At least 30 *ESCO2* gene mutations have been found to cause Roberts syndrome, which is characterized by limb and facial abnormalities and slow growth before and after birth. These mutations prevent the cell from producing any functional *ESCO2* protein. Some mutations change single protein building blocks (amino acids), while others result in an abnormally short protein. The absence of functional *ESCO2* protein causes some of the glue between sister chromatids to be missing around the chromosome's constriction point (centromere). In Roberts syndrome, cells respond to abnormal sister chromatid attachment by delaying cell division. Delayed cell division can be a signal that the cell should undergo self-destruction. The signs and symptoms of Roberts syndrome may be due to the loss of cells from various tissues during early development.

Researchers originally suspected that the varying severity of Roberts syndrome was caused by different types of mutations in the *ESCO2* gene. They predicted that people with the mild form of the disorder would have mutations that reduced the activity of the *ESCO2* protein, while those with the severe form would have mutations that completely eliminated the protein's function. However, all known mutations in the *ESCO2* gene prevent the production of any functional *ESCO2* protein. The underlying cause of the variation in disease severity remains unknown. Researchers suspect that other genetic and environmental factors may be involved.

3. Other Names for This Gene

- EFO2
- ESCO2_HUMAN
- establishment of cohesion 1 homolog 2
- establishment of cohesion 1 homolog 2 (*S. cerevisiae*)

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