# TCOF1 Gene

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Treacle ribosome biogenesis factor 1: The TCOF1 gene provides instructions for making a protein called treacle.

Keywords: genes

## 1. Normal Function

The *TCOF1* gene provides instructions for making a protein called treacle. This protein is active during early embryonic development in structures that become bones and other tissues of the face, and it appears to play a critical role in the formation of these structures.

Studies suggest that treacle is involved in the production of a molecule called ribosomal RNA (rRNA), a chemical cousin of DNA. Ribosomal RNA helps assemble protein building blocks (amino acids) into functioning proteins, which is essential for the normal functioning and survival of cells. Treacle is active in the nucleolus, which is a small region inside the nucleus where rRNA is produced.

## 2. Health Conditions Related to Genetic Changes

### 3. Treacher Collins syndrome

About 200 mutations in the *TCOF1* gene have been identified in people with Treacher Collins syndrome, a condition that affects the development of bones and other tissues of the face. Most of these mutations insert or delete a small number of DNA building blocks (base pairs) in the *TCOF1* gene, which leads to a reduction in the amount of functional treacle in cells. As a result, the production of rRNA is reduced, which likely triggers the self-destruction (apoptosis) of certain cells involved in the early development of facial bones and tissues. Researchers believe that this abnormal cell death may lead to the specific problems with facial development found in Treacher Collins syndrome. However, it is unclear why the effects of a reduction in rRNA are limited to facial development.

### Coloboma

## 3. Other Names for This Gene

- TCOF_HUMAN
- TCS
- Treacher Collins syndrome protein
- Treacher Collins-Franceschetti syndrome 1
- treacle

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