ECM1 Gene

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Extracellular matrix protein 1

genes

1. Normal Function

The *ECM1* gene provides instructions for making a protein that is found in most tissues within the extracellular matrix, which is an intricate lattice that forms in the space between cells and provides structural support. The ECM1 protein can attach (bind) to numerous structural proteins and is involved in the growth and maturation (differentiation) of cells, including skin cells called keratinocytes. The protein may also regulate the formation of blood vessels (angiogenesis).

Four different versions (isoforms) of the ECM1 protein are produced from the *ECM1* gene. These isoforms vary in length and in the tissues where they are found. The most abundant and widespread version is known as ECM1a.

2. Health Conditions Related to Genetic Changes

2.1 Lipoid Proteinosis

At least 55 mutations in the *ECM1* gene have been found to cause lipoid proteinosis, a condition that results from numerous, small clumps (deposits) of proteins and other molecules that form in various tissues throughout the body. Affected individuals typically have a hoarse voice, skin abnormalities, and neurological and respiratory problems.

Typically, mutations that cause lipoid proteinosis occur in areas of the *ECM1* gene known as exon 6 and exon 7. One mutation that deletes a single DNA building block (nucleotide) from exon 6 of the *ECM1* gene (written 507delT) has been found in multiple individuals around the world. Another mutation that occurs in exon 7 of the gene is common in affected individuals in South Africa and results in a premature stop signal in the instructions for making the protein (written as Gln276Ter or Q276X). The *ECM1* gene mutations that cause lipoid proteinosis result in the production of a nonfunctional protein or no protein at all.

A lack of functional ECM1 protein reduces binding between ECM1 and other proteins, leading to an unstable extracellular matrix. Without adequate support from the extracellular matrix, cells in the skin and other tissues are

weakened. However, the cause of the deposits in skin and other tissues is not clear. The unstable extracellular matrix may cause neighboring cells to overproduce proteins and other materials. It is possible that, as these excess substances accumulate in tissues, they create the deposits characteristic of lipoid proteinosis.

3. Other Names for This Gene

- secretory component p85
- URBWD

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