MMP2 Gene

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matrix metallopeptidase 2

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1. Introduction

The *MMP2* gene provides instructions for making an enzyme called matrix metallopeptidase 2. This enzyme is produced in cells throughout the body and becomes part of the extracellular matrix, which is an intricate lattice of proteins and other molecules that forms in the spaces between cells. One of the major known functions of matrix metallopeptidase 2 is to cut (cleave) a protein called type IV collagen. Type IV collagen is a major structural component of basement membranes, which are thin, sheet-like structures that separate and support cells as part of the extracellular matrix.

The activity of matrix metallopeptidase 2 appears to be important for a variety of body functions. These include the breakdown of the uterine lining (endometrium) during menstruation, formation and growth of new blood vessels, repair of damaged tissues, and inflammation. Matrix metallopeptidase 2 also plays a role in bone remodeling, which is a normal process in which old bone is broken down and new bone is created to replace it.

2. Health Conditions Related to Genetic Changes

2.1. Multicentric osteolysis, nodulosis, and arthropathy

At least eight mutations in the *MMP2* gene have been found to cause multicentric osteolysis, nodulosis, and arthropathy (MONA), a rare inherited bone disease that is characterized by the loss of bone tissue (osteolysis), particularly in the hands and feet, and related joint problems described as arthropathy. Each of the known *MMP2* gene mutations eliminates the function of the matrix metallopeptidase 2 enzyme, preventing the normal cleavage of type IV collagen. It is unclear how a loss of enzyme activity leads to the specific features of MONA. Researchers suspect that it somehow disrupts the balance of new bone creation and the breakdown of existing bone during bone remodeling, resulting in a progressive loss of bone tissue. How a shortage of matrix metallopeptidase 2 leads to other features of MONA, such as firm lumps under the skin (subcutaneous nodules) and skin abnormalities, is unknown.

3. Other Names for This Gene

- 72 kDa gelatinase
- 72 kDa type IV collagenase
- CLG4
- CLG4A
- collagenase type IV-A
- gelatinase A
- matrix metallopeptidase 2 (gelatinase A, 72kDa gelatinase, 72kDa type IV collagenase)
- matrix metalloproteinase-2
- matrix metalloproteinase-II
- MMP-2
- MMP-II
- MMP2_HUMAN
- neutrophil gelatinase
- TBE-1

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