Editorial
Volume 15:12, 2021

Journal of Molecular and Genetic Medicine

ISSN: 1747-0862 Open Access

Editorial on Hereditary Multicentric Osteolysis

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Editorial

MONA stands for multicentric osteolysis, nodulosis, and arthropathy, a rare genetic condition marked by bone tissue loss (osteolysis), especially in the hands and feet. The disorder originally known as nodulosis-arthropathy-osteolysis (NAO) syndrome is now known as MONA. MONA stands for multicentric osteolysis, nodulosis, and arthropathy, a rare genetic condition marked by bone tissue loss (osteolysis), especially in the hands and feet. The disorder originally known as nodulosis-arthropathy-osteolysis (NAO) syndrome is now known as MONA. Torg syndrome is a related condition that may be included in MONA, while it is unclear whether Torg syndrome is a separate disorder caused by a mutation in a different gene. Bone loss begins in the hands and feet in most cases of MONA, causing pain and restricting movement. Joint issues (arthropathy) can develop in the elbows, shoulders, knees, hips, and spine as a result of bone abnormalities spreading to other parts of the body. MONA causes poor bone mineral density (osteopenia) and bone thinning (osteoporosis) across the skeleton in the majority of persons. Bones become brittle and more prone to fractures as a result of these anomalies. Short stature can also cause by bone defects.

Many affected individuals develop subcutaneous nodules, which are firm lumps of noncancerous tissue underneath the skin, especially on the soles of the feet. Skin abnormalities, such as patches of dark, thick, and leathery skin, are also seen in some of the affected people. Other features of MONA can include clouding of the clear front covering of the eye (corneal opacity), excess hair growth (hypertrichosis), and overgrowth of the gums, heart abnormalities, and distinctive facial features that are described as course. Mutations in the *MMP2* gene are the cause of MONA. This gene codes for matrix metallopeptidase 2, an enzyme whose major purpose is to cut (cleave) a protein termed type IV collagen. Basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues, contain a lot of type IV collagen. Matrix metallopeptidase 2 activities appears to be vital for a number of physiological activities, including bone remodelling, which is a natural process in which old bone is broken down and new bone is formed to replace it.

The *MMP2* gene mutations that cause MONA totally disable the matrix metallopeptidase 2 enzymes, preventing type IV collagen from being cleaved normally. It's unclear how a lack of enzyme function causes MONA's distinct characteristics. Researchers believe it alters the equilibrium of new bone formation and current bone breakdown during bone remodelling, resulting in gradual bone tissue loss. It's unclear how a lack of matrix metallopeptidase 2 causes other MONA symptoms such subcutaneous lumps and skin abnormalities. This disease is autosomal recessive, which means both copies of the gene in each cell have mutations. Each of the parents of a person with an autosomal recessive trait.

How to cite this article: Rita, Badigeru. "Editorial on Hereditary Multicentric Osteolysis." J Mol Genet Med 15 (2021): 528.

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Received 20 December 2021; **Accepted** 25 December 2021; **Published** 30 December 2021