Some muscular dystrophy patients at increased risk for cancer

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People who have the most common type of adult muscular dystrophy also have a higher risk of getting cancer, according to a paper published today in the *Journal of the American Medical Association*.

The team found that patients who have myotonic muscular dystrophy are at increased risk primarily for four types of cancer: brain, ovary, colon, and the uterine lining known as the endometrium. The team also found a possible increased risk for some other types of cancer, including cancer of the eye, thyroid, pancreas, and other female reproductive organs.

Physicians estimate that approximately 40,000 Americans have myotonic dystrophy, an inherited disease that is marked by progressive muscle weakness. While the course of the disease varies from patient to patient, symptoms can include muscle stiffness, difficulty speaking and swallowing, problems walking, and in some patients, heart problems and cataracts.

Neurologists like study author Richard T. Moxley, M.D., of the University of Rochester Medical Center have long known that patients with myotonic dystrophy are at greater risk of a rare type of skin growth, and they know that skin cancer occurs more often as well in some families with the disease. Moxley teamed with cancer experts at the National Cancer Institute as well as scientists in Sweden and Denmark to study the link between muscular dystrophy and cancer more closely.

The team used detailed registries about the health of people with

myotonic dystrophy to look closely at the records of 1,658 patients. Among that group, 104 people developed cancer – twice the number of cases that would be expected in the overall population.

"Our findings suggest that patients with myotonic dystrophy need to be absolutely vigilant about cancer screening, particularly colon cancer screening," said Moxley, director of the University's Neuromuscular Disease Center and professor of Neurology. Moxley also heads the University's Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Center, one of six research centers funded by the National Institutes of Health.

Over the last 15 years, Moxley's colleague, Charles Thornton, M.D., has discovered how the genetic flaw at the root of myotonic muscular dystrophy – a genetic repeat, a kind of molecular stutter – actually causes the disease. He found that the flaw causes extra messenger RNA to accumulate in the nucleus of cells, making it difficult for a protein crucial for normal muscle growth to do its job.

Moxley notes that the genetic miscue that causes the disease is one that might also make cancer more likely to occur. The investigators say it's possible that the extra RNA may also hinder proteins involved in repairing DNA; a malfunction of DNA repair machinery is one way that cancer comes about. In addition, they note that the product made by the faulty gene is a close molecular relative of several cancer susceptibility genes.

The scientists say much more study is needed to understand the process. They plan to study the issue in more depth using a muscular dystrophy registry which includes about 1,600 patients with muscular dystrophy that has been compiled by Moxley and colleagues at Rochester, thanks to funding from the National Institutes of Health.

More information: JAMA. 2011;306[22]:2480-2486.