

**Poster presentation at the American Academy of Neurology Conference
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Neoplasms in Myotonic Dystrophy type 1

Background

Myotonic dystrophy type 1 (DM1) is the most common adult muscular dystrophy and causes progressive muscle weakness, myotonia, and multi-system complications. DM1 is the result of a specific genetic mutation referred to as a DNA expansion or an increase in the amount of DNA that is normally located on a chromosome. The mutation causes the cells in muscle and other tissue to function incorrectly. The extent to which this mutation affects the body is not entirely understood. For example, there is limited information on how the genetics of DM1 may potentially influence various types of benign (non-cancerous) and malignant (cancerous) tumors.

Specific types of tumors have been shown to be associated with DM1. The most common tumors linked to DM1 are small, non-cancerous skin lumps. These small, hard lumps are called pilomatrixomas and are commonly found on the neck and forehead. Clinicians have also noted an increase in the number of other benign and malignant tumors in DM1, but there is limited information on the overall prevalence of cancer in DM1. To date, there have been only a few research studies published to support these observations.

Methods/Results

We reviewed and summarized reports about tumors in DM1 that have been reported in medical literature. We also analyzed the data in the Registry regarding patient reported tumors.

Conclusion

DM1 patients may have an increased frequency of pilomatrixomas. Several tumors observed in DM1 patients have also been reported in patients with other genetic disorders. These tumors may have a shared pathophysiology. Future research in this area is necessary to determine if DM1 patients do have an increased incidence of tumors and to identify the specific cellular pathways that may influence tumor growth.