

PREVALENCE OF LYNCH SYNDROME MUTATIONS IN PATIENTS WITH COLORECTAL AND ENDOMETRIAL CANCER BASED ON DECADE OF DIAGNOSIS



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INTRODUCTION:

Lynch syndrome is an autosomal dominant genetic syndrome causing an increased risk primarily for colorectal and endometrial cancers in addition to other cancers.

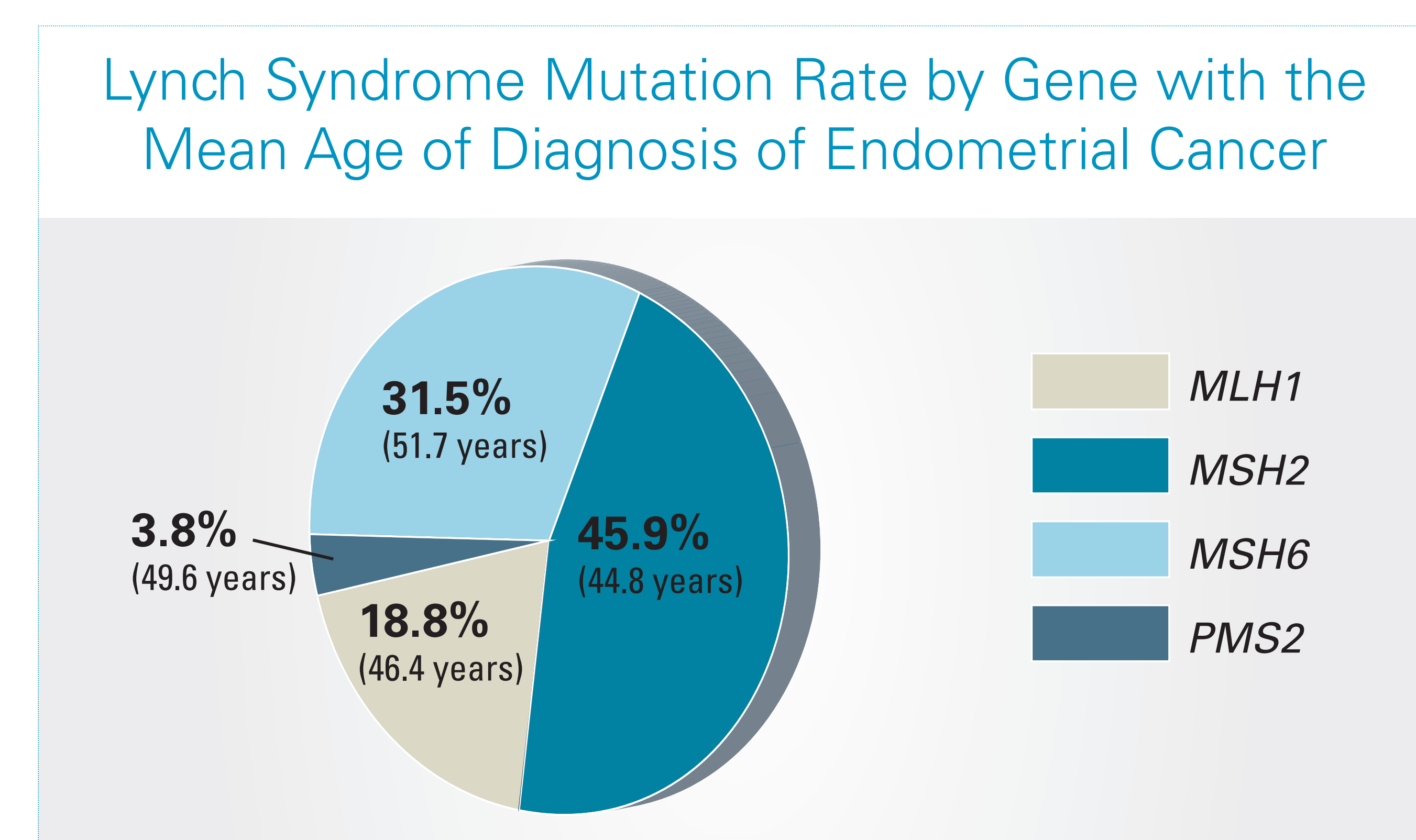
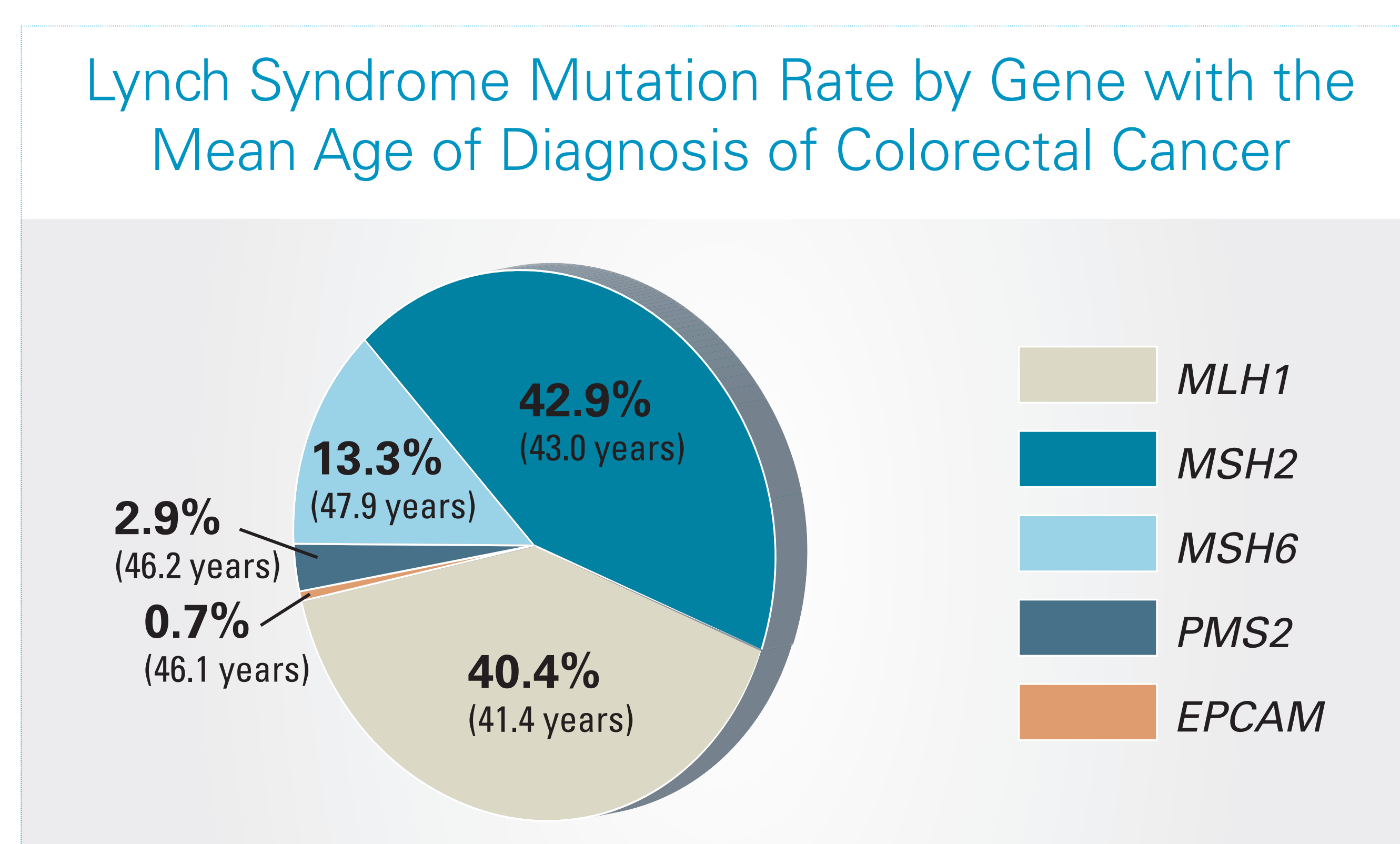
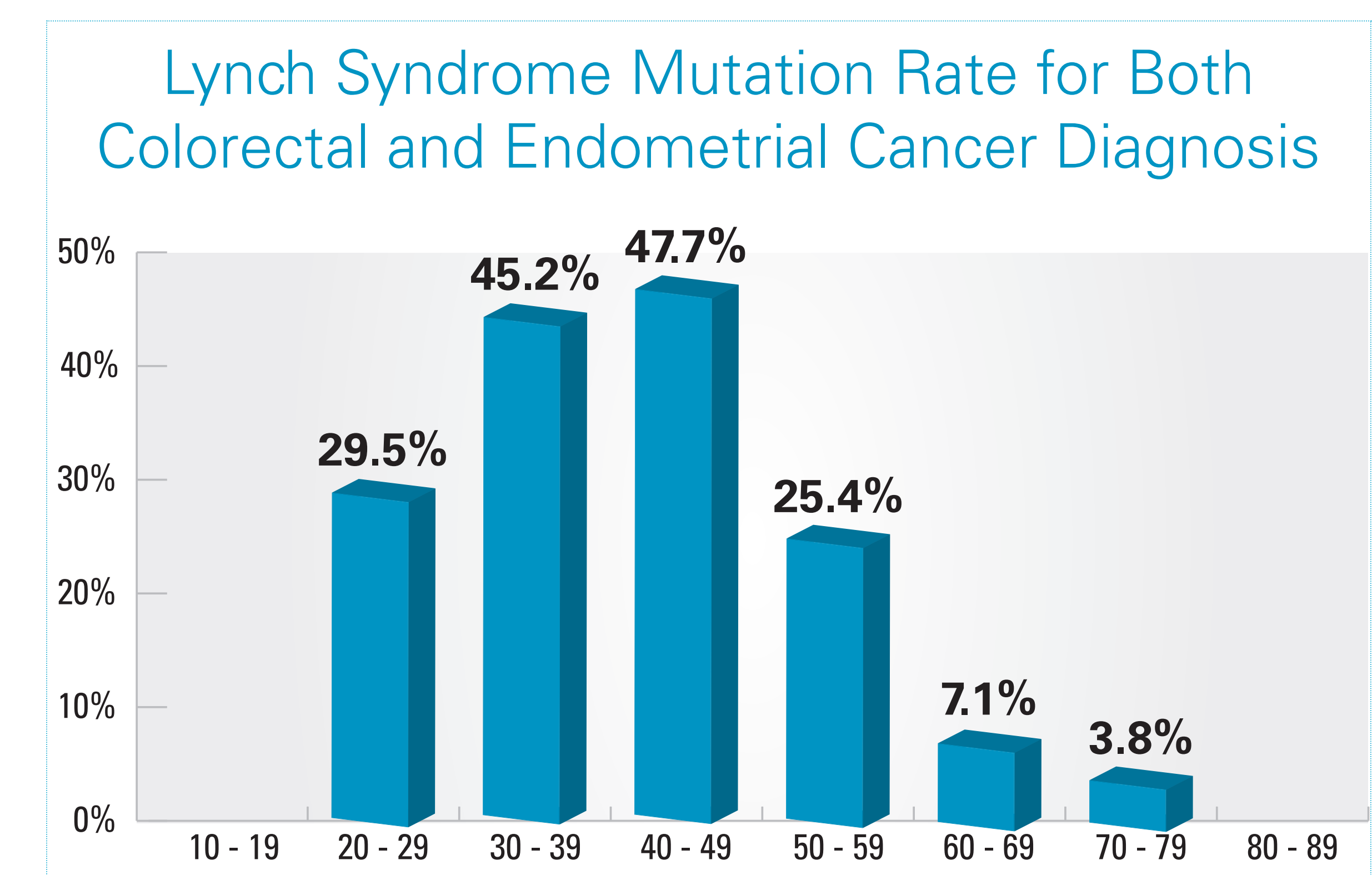
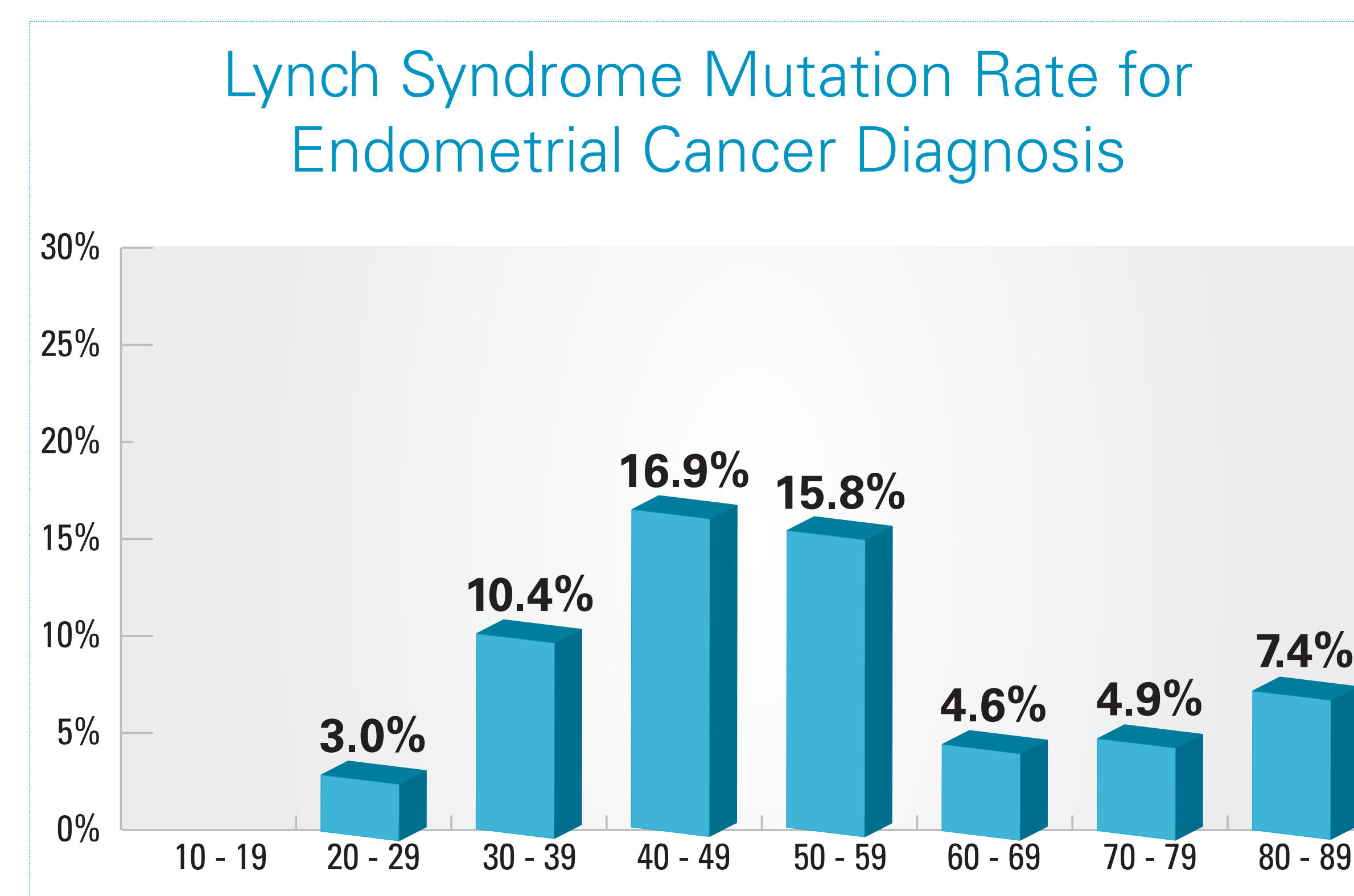
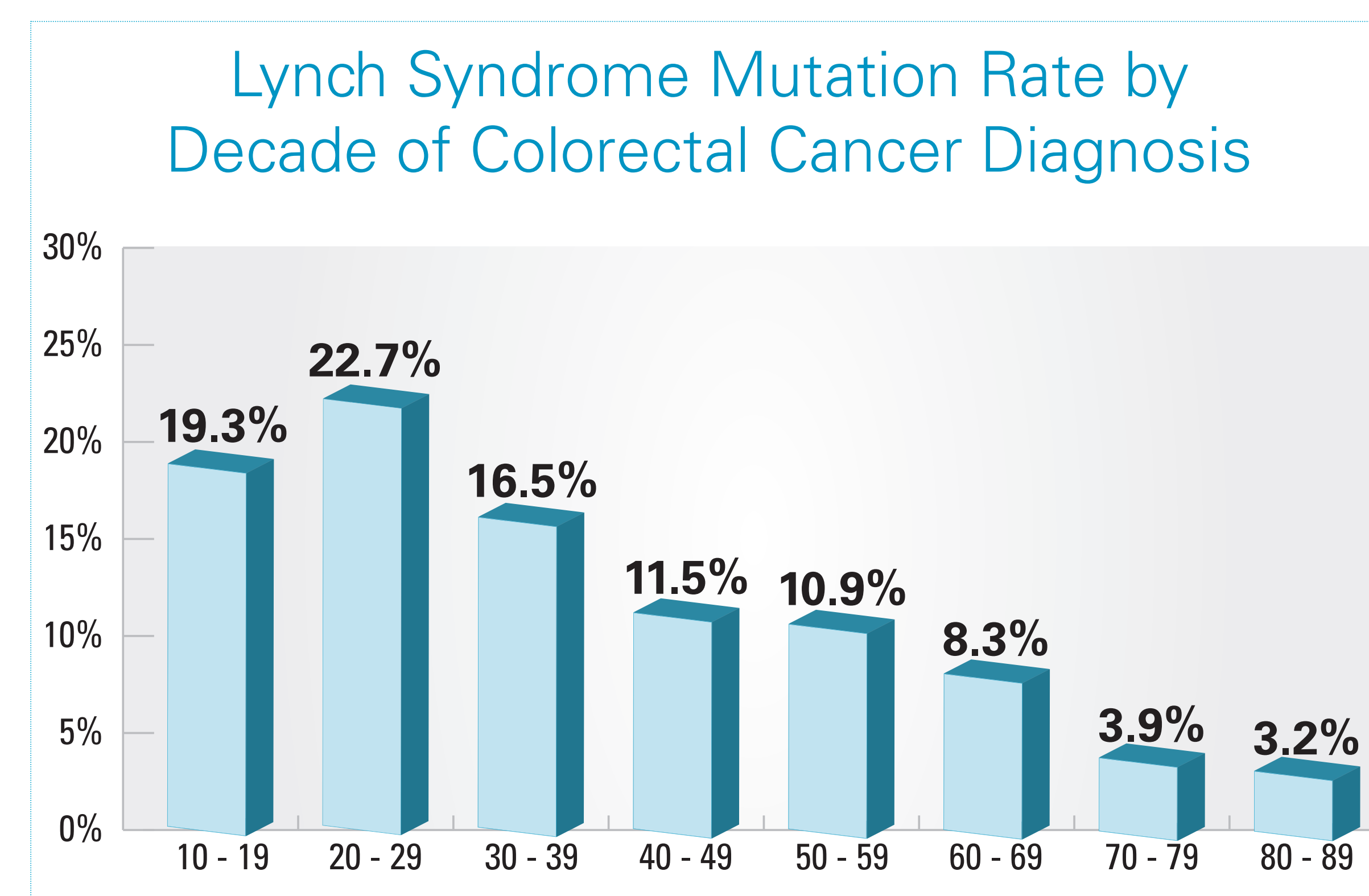
To understand more about the phenotype of Lynch syndrome patients, we queried patients with a history of colorectal cancer (CRC) and/or endometrial cancer (EC) who underwent Lynch syndrome testing from a genetic testing laboratory database, one of the largest databases of patients tested for Lynch syndrome.

METHODS:

Data was obtained from over 20,000 patients from September 2006 to October 2012 for individuals who had LS testing and were affected with CRC and/or EC, regardless of family history.

Patients who had single gene testing, presumably based upon tumor testing via immunohistochemistry, or mutation specific testing based upon a known mutation in the family history were excluded from the analysis.

RESULTS:



CONCLUSIONS:

- Patients diagnosed with CRC and/or EC in their 50s have a comparable Lynch syndrome mutation rate to patients diagnosed in their 40s.
- Testing criteria for Lynch syndrome should consider including patients diagnosed with CRC or EC less than age 60 regardless of family history.
- The high Lynch syndrome mutation rate among patients with both CRC and EC demonstrates the high risk of these patients developing a second cancer and highlights the critical importance of evaluating all CRC and EC patients at their initial diagnosis for Lynch syndrome to potentially decrease the risk of future cancers.