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Pediatrics & Therapeutics

Short Communication

Lynch Syndrome and Colorectal Cancer in Pediatrics

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DESCRIPTION

A heterozygous change in one of the DNA Mismatch Repair (MMR) genes results in Lynch syndrome, an autosomal dominant disorder that predisposes people to colon tumors and other malignancies with an early start. Lynch syndrome is typically thought of as an adult-onset illness because childhood cancer manifestations are uncommon. Although colorectal cancer in children is exceedingly rare, genetic diseases like Lynch syndrome play a significant role in its development.

Hereditary Non-Polyposis Colon Cancer (HNPCC), also known as Lynch syndrome, is an autosomal dominant genetic disorder that predisposes people to colorectal cancers with early onset as well as a number of other extra-intestinal cancers like endometrial, ovarian, gastric, pancreatic, biliary, skin, and central nervous system tumors. A heterozygous germline pathogenic variation in one of the five known DNA Mismatch Repair (MMR) genes MLH1, MSH2, MSH6, PMS2, and EPCAM is responsible for Lynch syndrome. With a prevalence of roughly 1 in 300 in the adult population, Lynch syndrome was first identified by Dr. Henry Lynch in 1985 and is now known to be the most prevalent genetic cause of colorectal cancer [1,2]. The average age at which colon cancer is diagnosed in people with Lynch syndrome is 45 years old, and malignancy seldom manifests in youngsters.

Less than 2 in 1 million children in the United States are diagnosed with primary colorectal cancer, making it an extremely rare illness. However, colorectal cancer is typically discovered in late stages in children and has a terrible prognosis. Although pediatric patients rarely develop colorectal cancer, Lynch syndrome is a significant contributor to this and other pediatric patients' malignancies. As a result, it is crucial that healthcare professionals are aware of Lynch syndrome in order to pursue the proper genetic screening of affected and at-risk children as well as their families.

About 2-3% of colorectal cancers in adults are caused by Lynch syndrome, but its frequency in children is unclear, in part because pediatric colorectal malignancies are so uncommon [3]. According to current recommendations, screening colonoscopies should start in people with Lynch syndrome between the ages of 20 and 25, and subsequent colonoscopies should be performed every two years, unless the results indicate otherwise [4]. Screening should start two to five years before the earliest colorectal cancer diagnosed before age 25 in families with exceptionally early-onset colorectal malignancies. In families with known Lynch syndrome, genetic testing is routinely provided after the age of 18, while the ideal age for testing has proven debatable. Young children have been referred for genetic counseling in families with early-onset Lynch syndrome-associated cancers. This brings up some difficult moral questions about genetic testing on young children, such as the psychological effects of parents' and kids' health concerns related to cancer risk syndromes and worries about discrimination when adults apply for life or health insurance. Due to these factors, many people continue to be hesitant about genetic testing, even in families where Lynch syndrome has been definitively diagnosed.

CONCLUSION

Although colorectal cancer in children is still relatively uncommon, Lynch syndrome can play a significant role in its development as well as other pediatric cancers. In order to best promote early identification and enhance outcomes, impacted families must be thoroughly educated on the signs of colorectal cancer and encouraged to talk openly with their children about this diagnosis. Additionally, it is critical that tissues from these cases undergo IHC testing in order to screen for DNA MMR pathogenic mutations given the prevalence of Lynch syndrome in pediatric colorectal malignancies. It would be beneficial to conduct more research to see how frequently Lynch syndrome actually contributes to pediatric colorectal malignancies.

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Received: 06-Sep-2022; Manuscript No. PTCR-22-19824; **Editor assigned:** 08-Sep-2022; PreQC. No. PTCR-22-19824 (PQ); **Reviewed:** 22-Sep-2022; QC. No. PTCR-22-19824; **Revised:** 29-Sep-2022; Manuscript No. PTCR-22-19824 (R); **Published:** 07-Oct-2022, DOI: 10.35841/2161-0665.22.12.462.

Citation: Kankirawatana P (2022) Lynch Syndrome and Colorectal Cancer in Pediatrics. Pediatr Ther. 12.462.

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