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Pedigree analysis supports correlation between an AXIN2 variant and polyposis/colorectal cancer

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Abstract

We present a patient with a history of colonic polyposis and family history significant for colon polyps and colorectal cancer (CRC). The patient and the family also had a history of bone loss of the jaw and early tooth loss, consistent with oligodontia. Genetic testing revealed the patient to have a previously unpublished variant of unknown significance (VUS) in the AXIN2 gene. These clinical findings have been demonstrated previously in only two other families, both of which exhibited oligodontia, colorectal neoplasia (polyps and cancer) and a heterozygous mutation in AXIN2 [1,2]. The AXIN2 protein is component of the Wnt pathway, which is known to be vital for organism development and cellular

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Pedigree analysis supports a correlation between an AXIN2 variant and polyposis

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2013年5月1日 - Colorectal cancer (CRC) in densely affected families without Lynch ... on 40 affected cases from 16 multi-case pedigrees to identify novel loci. .... of the variant with additional CRC- and polyp-affected and unaffected ..... Linkage analysis in a large Swedish family supports the presence of a .... External link.

[PDF] Chris Dvorak - Tulane University

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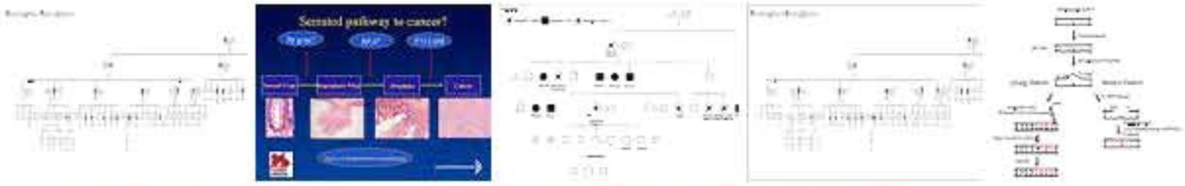

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作者：L Lammi - 2004 - 被引用次数：530 - 相关文章

2004年3月23日 - Our findings introduce a new gene for hereditary colorectal cancer and suggest ... the adenomatous polyposis coli gene) and Axin1 or its homolog Axin2, which .... For a genomewide search with linkage analysis, tooth agenesis was .... the connection between them and congenital malformations in humans.

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2004年3月23日 - Germline loss-of-function mutations in APC cause familial adenomatous polyposis (FAP [MIM 175100]), comprising ~10% of hereditary colorectal cancer (Groden et al.; Kinzler et al.). It is interesting that familial colorectal polyposis is often accompanied by extracolonic neoplasia, especially cysts, osteomas ...

Low frequency of AXIN2 mutations and high frequency of MUTYH ...

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