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Manuscript NO: 36206

Manuscript Type: Case Report

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 Genetic testing revealed the patient to have a previously unpublished variant of unknown significance (VUS) in the AXIN2 gene.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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
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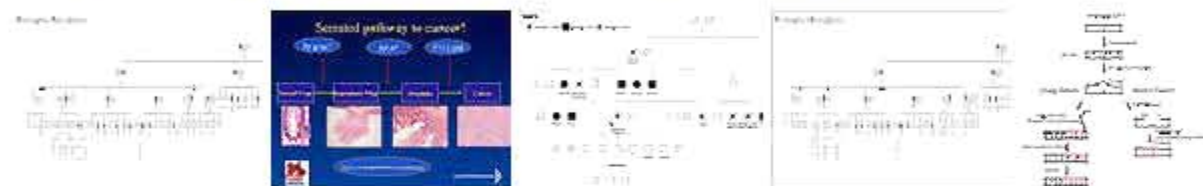
2013年5月1日 - Colorectal cancer (CRC) in densely affected families without Lynch ... on 40 affected cases from 16 multi-case pedigrees to identify novel loci. of the variant with additional CRC- and polyp-affected and unaffected Linkage analysis in a large Swedish family supports the presence of a External link.

Chris Dvorak - Tulane University

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Mutations in AXIN2 Cause Familial Tooth Agenesis and Predispose to ...

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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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Colon cancer is a well-known feature of familial polyposis coli. ... Pedigree analysis showed best fit with autosomal dominant inheritance. ... Likelihood analysis strongly supported dominant inheritance of a susceptibility to ... by treatment with the demethylating agent 5-aza-C. A striking correlation between genetic instability ...

Familial Colorectal Cancer Type X (FCCTX) and the correlation with ...

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2017年10月18日 - In this review, we focus on the genes and their variants identified in ... Recent studies describe the correlation between FCCTX and genes ... Among patients with clinical characteristics of hereditary non-polyposis colorectal cancer (CRCs), ... When samples were analyzed for copy-number alterations, they ...

AXIN1 and AXIN2 Variants in Gastrointestinal Cancers - NCBI - NIH

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作者: SM Mazzoni - 2014 - 被引用次数: 31 - 相关文章

2014年9月16日 - Mutations in the APC (adenomatous polyposis coli) gene, which encodes a ... Keywords: Wnt signaling, beta-catenin, colon cancer, hepatocellular carcinoma, gastric cancer The connection between AXIN2 exon 7 frameshifts and Further efforts may support the role of AXIN2 variant alleles in cancer ...

缺少字词: pedigree