MSH2 variant: MSH6-defective DNA mismatch repair

Arora, S., Gillespie, ME.

European Bioinformatics Institute, New York University Langone Medical Center, Ontario Institute for Cancer Research, Oregon Health and Science University.

The contents of this document may be freely copied and distributed in any media, provided the authors, plus the institutions, are credited, as stated under the terms of Creative Commons Attribution 4.0 International (CC BY 4.0) License. For more information see our license.

22/11/2021
Introduction

Reactome is open-source, open access, manually curated and peer-reviewed pathway database. Pathway annotations are authored by expert biologists, in collaboration with Reactome editorial staff and cross-referenced to many bioinformatics databases. A system of evidence tracking ensures that all assertions are backed up by the primary literature. Reactome is used by clinicians, geneticists, genomics researchers, and molecular biologists to interpret the results of high-throughput experimental studies, by bioinformaticians seeking to develop novel algorithms for mining knowledge from genomic studies, and by systems biologists building predictive models of normal and disease variant pathways.

The development of Reactome is supported by grants from the US National Institutes of Health (P41 HG003751), University of Toronto (CFREF Medicine by Design), European Union (EU STRP, EMI-CD), and the European Molecular Biology Laboratory (EBI Industry program).

Literature references


Reactome database release: 78

This document contains 1 reaction (see Table of Contents)
MSH2 variant: MSH6-defective DNA mismatch repair

Stable identifier: R-HSA-5577244

Type: transition

Compartments: nucleoplasm

Diseases: colorectal cancer

MSH2 is homologous to the E. coli MutS gene and is involved in DNA mismatch repair (MMR) (Fishel et al., 1994).

Evidence to support a role for the mismatch repair genes human mutS homolog 2 (hMSH2) in the etiology of colorectal cancer has come from linkage analysis, segregation studies, and molecular biologic analysis. More recently, carriers of potentially pathogenic mutations in the hMSH2 genes have consistently been shown to be at a greatly increased risk of developing colorectal cancer compared with the general population.

Two variants are described here MSH2 ARG406TER and MSH2 GLN601TER. Both variants disrupt the formation of the MSH2:MSH6 complex. The MSH2 ARG406TER occurred in a kindred with hereditary nonpolyposis colorectal cancer. The variant contains a CGA-to-TGA transition in codon 406, resulting in change of arginine to a stop (Leach et al., 1993).

The MSH2 GLN601TER variant occurs in a kindred with characteristics of the Muir-Torre syndrome (Kolodner et al., 1994).

Literature references


Editions

<table>
<thead>
<tr>
<th>Date</th>
<th>Action</th>
<th>Author</th>
</tr>
</thead>
<tbody>
<tr>
<td>2014-05-24</td>
<td>Authored</td>
<td>Gillespie, ME.</td>
</tr>
<tr>
<td>2016-11-01</td>
<td>Reviewed</td>
<td>Arora, S.</td>
</tr>
<tr>
<td>2017-02-28</td>
<td>Edited</td>
<td>Gillespie, ME.</td>
</tr>
</tbody>
</table>