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**Name of Journal:** *World Journal of Clinical Cases*

**Manuscript NO:** 57159

**Manuscript Type:** CASE REPORT

**A new mutation in *EPCAM* for congenital tufting enteropathy: A case report**

Zhou YQ *et al.* new mutation in *EPCAM* for congenital tufting enteropathy

Yan-Qiong Zhou, Guo-Sheng Wu, Yuan-Mei Kong, Xiao-Yuan Zhang, Chun-Lin Wang



A new mutation in EPCAM for Congenital Tufting Enteropathy: A ca



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## Novel Mutations in EPCAM Cause Congenital Tufting Enteropathy

<https://pubmed.ncbi.nlm.nih.gov/27875355>

Novel Mutations in EPCAM Cause Congenital Tufting Enteropathy J Clin Gastroenterol. 2018 Jan;52 ... Congenital tufting enteropathy (CTE) is a rare autosomal recessive form of intractable diarrhea of infancy. Patients develop chronic diarrhea within days after birth, leading to severe malabsorption and significant mortality. ... was subjected to ...

## Tufting Enteropathy with EpCAM Mutations in Two Siblings

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Sep 24, 2010 · Since tufting enteropathy was first described in 1994, 7 several cases including a Korean infant have been reported. 6, 8-10 The prevalence of tufting enteropathy is estimated at around 1/50,000-100,000 live births in Western Europe. 2 Recently, mutations in the epithelial cell adhesion molecule (EpCAM) gene have been identified as responsible for tufting enteropathy. ...

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Author: Jae Sung Ko, Jeong Kee Seo, Jeong Ok ...

Publish Year: 2010

## EPCAM mutation update: Variants associated with congenital ...

<https://pubmed.ncbi.nlm.nih.gov/30461124>

Biallelic mutations in EPCAM cause congenital tufting enteropathy (CTE), which is a rare chronic diarrheal disorder presenting in infancy. Monoallelic deletions of the 3' end of EPCAM that silence the downstream gene, MSH2, cause a form of Lynch syndrome, which is a cancer predisposition syndrome associated with loss of DNA mismatch repair.

## (PDF) Tufting enteropathy with EpCAM mutation: Case report

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Tufting enteropathy (TE), also known as intestinal epithelial dysplasia (IED), is a rare congenital



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**Cited by:** 6**Author:** Wenjuan Tang, Taosheng Huang, Zhongyao...**Publish Year:** 2016

## [\(PDF\) Tufting enteropathy with EpCAM mutation: Case report](https://www.researchgate.net/publication/274548180...)

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**Cited by:** 6**Author:** Sagar J. Pathak, Sagar J. Pathak, James L....**Publish Year:** 2019

## [Tufting enteropathy with EpCAM mutation: case report](http://www.scielo.br/scielo.php?script=sci_arttext&pid=S1676-24442014000300234)

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New mutation in EPCAM for congenital tufting enteropathy: A case



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Tufting enteropathy with EpCAM mutation: case report inflammatory cells within the lamina propria. Intraepithelial lymphocytes are normal in number (1, 5-7, 10, 11, 13, 17, 20). In contrast with the literature, in our report, the epithelial tufts were found in the terminal ileum and right colon instead of ...

## [Tufting enteropathy with EpCAM mutation: case report](https://www.scielo.br/scielo.php?script=sci_arttext&pid=S1676-24442014000300234)

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<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2956358/>

Tufting enteropathy (TE), also known as intestinal epithelial dysplasia (IED), is a rare congenital

## Congenital tufting enteropathy

Inherited Disorder

Congenital tufting enteropathy is an inherited disorder of the small intestine that presents with intractable diarrhea in young children.



Data from: Wikipedia

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