



**Name of Journal:** *World Journal of Gastroenterology*

**Manuscript NO:** 40292

**Manuscript Type:** CASE REPORT

**Novel MARS gene variants/phenotypes in interstitial lung and liver disease:  
A case report and review of the literature**

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Jian-She Wang

**Abstract**

Interstitial lung and liver disease (ILLD) is caused by biallelic mutations in the methionyl-tRNA synthetase (MARS) gene. To date, no genetic changes other than missense variants were reported in the literature. Here we report a five-month-old female infant with typical ILLD (failure to thrive, developmental delay, jaundice, diffuse interstitial lung disease, hepatomegaly

Match Overview

1	<b>Crossref</b> 29 words Hyun, Y.S., H.J. Park, S.-H. Heo, B.R. Yoon, S.H. Nam, S.-B. Kim, C.I. Park, B.-O. Choi, and K.W. Chung. "Rare variants i	1%
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### Mutations in methionyl-tRNA synthetase gene in a Chinese family with ...

<https://www.ncbi.nlm.nih.gov/pubmed/28148924> ▾ [翻译此页](#)

作者 : Y Sun - 2017 - 被引用次数 : 6 - [相关文章](#)

2017年2月2日 - ... in a Chinese family with **interstitial lung and liver disease**, postnatal growth failure and anemia. ... Methionyl-tRNA synthetase (**MARS**) catalyzes the ligation of ... Here we report a Chinese ILLD family with two affected boys ... They carry **novel** compound heterozygous **MARS** mutations (p. ... **Case Reports** ...

缺少字词 : variants

### "Eur J Med Genet"[ta] - PubMed Result - NCBI

<https://www.ncbi.nlm.nih.gov/sites/entrez?term...> - [翻译此页](#)

2: Al-Murshedi F, Meftah D, Scott P. Underdiagnoses resulting from **variant** ... Boduroglu K. A **novel** NKX3-2 mutation associated with perinatal lethal **phenotype** of ... Newbury-Ecob R, Tobias ES; DDD **Study**, Joss S. Extending the clinical and ... **interstitial lung and liver disease** and dominant Charcot-Marie-Tooth disease.

### Whole-exome sequencing reveals a novel missense mutation in the ...

<https://onlinelibrary.wiley.com/doi/full/10.1111/jns.12264> - [翻译此页](#)

作者 : L Sagi-Dain

2018年3月26日 - Arg737Trp mutation in the **MARS** gene (OMIM 156560). ... Family segregation analysis **study** revealed that the patient had inherited the **variant** from her ... Two common types, type I (encompassing 40%-50% of the **cases**) and type II .... with a rare type of **interstitial lung and liver disease** (MIM #615486), ...

### MARS variant associated with both recessive interstitial lung and liver ...

<https://europepmc.org/abstract/med/29655802> ▾ [翻译此页](#)

作者 : J Rips - 2018

2018年4月12日 - **MARS variant** associated with both recessive **interstitial lung and liver disease**



gene variants/phenotypes in interstitial lung and liver disease: A case report



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## Advances in understanding the genetic architecture of cleft lip and palate disorders

[PDF] uiowa.edu

EJ Leslie - 2012 - [ir.uiowa.edu](http://ir.uiowa.edu)

... Page 21. xv LIST OF GENE NAMES ... Subphenotypes are also important to consider in the design of genetic studies, where the power to detect an effect may be weakened ... (<http://www.ncbi.nlm.nih.gov/OMIM>). Identification of genes causing these syndromes ...

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### Interstitial Lung Disease in Childhood: Clinical and Genetic Aspects

<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4603523/> ▼ [翻译此页](#)

作者: H Kitazawa - 2015 - 被引用次数: 8 - [相关文章](#)

2015年10月11日 - Abstract. **Interstitial lung disease** (ILD) in childhood is a heterogeneous group of rare ..... The clinical **phenotype** is severe hypoxemic respiratory **failure** and pulmonary ..... **Report** of three **cases** and **review** of the **literature**. .... A large kindred of pulmonary fibrosis associated with a **novel** ABCA3 **gene variant**.

### MARS variant associated with both recessive interstitial lung and liver ...

<https://www.ncbi.nlm.nih.gov/pubmed/29655802> - [翻译此页](#)

作者: J Rips - 2018 - [相关文章](#)

2018年4月12日 - **MARS variant** associated with both recessive **interstitial lung** and **liver disease** and dominant Charcot-Marie-Tooth disease. Rips J(1) ... (4)Department of **Genetic** and Metabolic Diseases, Hadassah-Hebrew University Medical Center, ... We **report** a male infant of non-consanguineous origin, presenting with ...

缺少字词: **novel case review literature**

### OMIM Entry - # 615486 - INTERSTITIAL LUNG AND LIVER DISEASE ...

<https://www.omim.org/entry/615486> ▼ [翻译此页](#)

615486 - **INTERSTITIAL LUNG AND LIVER DISEASE**; ILLD - PULMONARY ALVEOLAR ...

INFANTILE **LIVER FAILURE** SYNDROME 2, FORMERLY; ILFS2, FORMERLY. **Phenotype-Gene** Relationships ..... is caused by homozygous or compound heterozygous mutation in the **MARS gene** (156560) on chromosome 12q13.

缺少字词: **novel case report**

### Rare recessive loss-of-function methionyl-tRNA synthetase mutations ...

<https://bmcmmedgenet.biomedcentral.com/articles/10.../1471-2350-14-106> ▼ [翻译此页](#)

作者: E van Meel - 2013 - 被引用次数: 25 - [相关文章](#)

2013年10月8日 - Methionyl-tRNA synthetase (**MARS**) catalyzes the ligation of ... DARS mutations have been identified in recessive neurologic **phenotypes** [2] ... In this **study** we identified loss-of-function mutations in the **gene** ... **Case** presentation ... hypothyroidism, **interstitial lung disease** and transfusion-dependent anemia