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دانشگاه علوم پزشکی شهید شاپور اهواز

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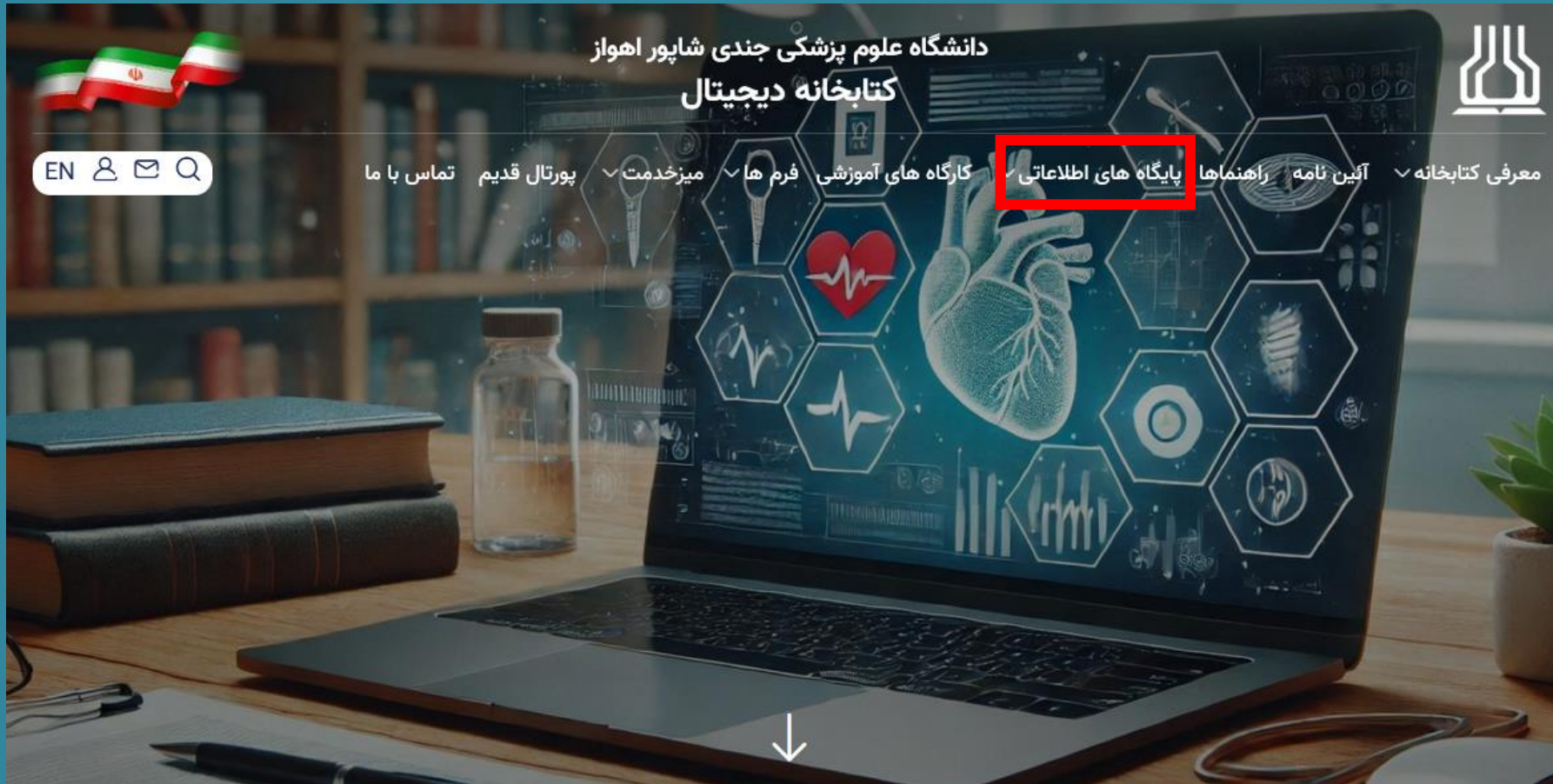
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# صفحة نتائج

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**TGFBF1 Variants Can Associate with Non-Syndromic Congenital Heart Disease without Aortopathy**  
Alaamery, Manal; Nour Albeshier; Alhabshan, Fahad; Barnett, Phil; Kabbani, Mohamed Salim; **et al.** **Journal of Cardiovascular Development and Disease; Basel** Vol. 10, Iss. 11, (2023): 455.

... Introduction **Congenital heart disease** (CHD) is...  
... **heart disease** coupled with an abnormal atrial rhythm and **conduction**...  
...TGFBF1 can also lead to **familial** forms of non-syndromic **congenital heart**...

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**GMDS Intragenic Deletions Associate with Congenital Heart Disease including Ebstein Anomaly**  
Lo-A-Nioe, Shirley M; Verberne, Eline A; Lars T van der Veken; Arends

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**TGFBR1 Variants Can Associate with Non-Syndromic Congenital Heart Disease without Aortopathy**

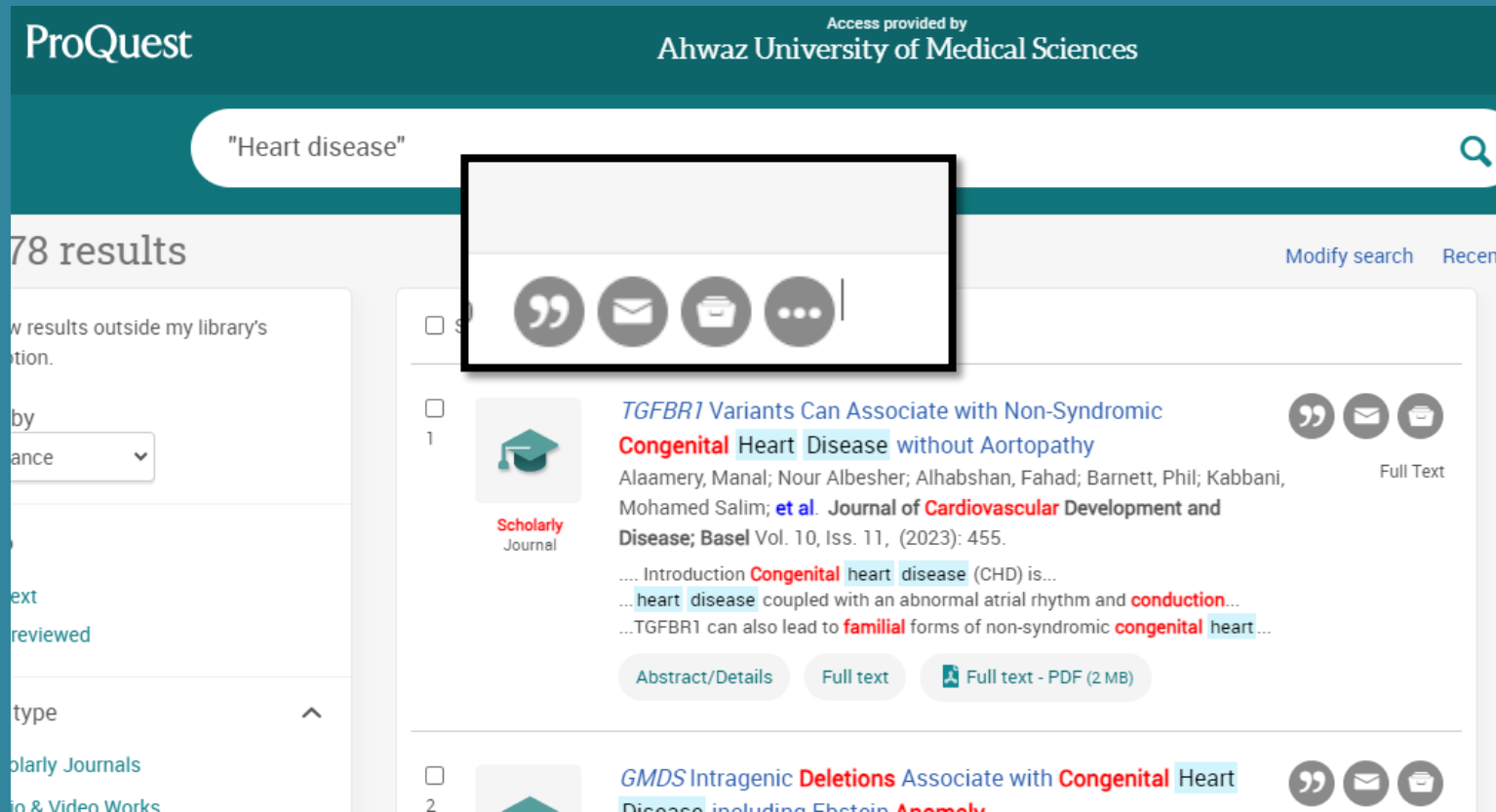
Alaamery, Manal; Nour Albeshier; Alhabshan, Fahad; Barnett, Phil; Kabbani, Mohamed Salim; et al. *Journal of Cardiovascular Development and Disease*; Basel Vol. 10, Iss. 11, (2023): 455.

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**GMDS Intragenic Deletions Associate with Congenital Heart Disease including Ebstein Anomaly**



# مشاهده متن کامل مدارک

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کامل مقاله

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## TGFBR1 Variants Can Associate with Non-Syndromic Congenital Heart Disease without Aortopathy

Alaamery, Manal; Nour Albeshar; Alhabshan, Fahad; Barnett, Phil; Kabbani, Mohamed Salim; et al. Journal of Cardiovascular Development and Disease; Basel Vol. 10, Iss. 11, (2023): 455. DOI:10.3390/jcdd10110455

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Abstract/Details

### Abstract

Translate

Background: Congenital heart diseases (CHD) are the most common congenital malformations in newborns and remain the leading cause of mortality among infants under one year old. Molecular diagnosis is crucial to evaluate the recurrence risk and to address future prenatal diagnosis. Here, we describe two families with various forms of inherited non-syndromic CHD and the genetic work-up and resultant findings. Methods: Next-generation sequencing (NGS) was employed in both families to uncover the genetic cause. In addition, we performed functional analysis to investigate the consequences of the identified variants in vitro. Results: NGS identified possible causative variants in both families in the protein kinase domain of the TGFBR1 gene. These variants occurred on the same amino acid, but resulted in differently substituted amino acids (p.R398C/p.R398H). Both variants co-segregate with the disease, are

### Suggested sources

Genetic heterogeneity of cardiomyopathy and its correlation with patient care  
Mi Jin Kim; Cha, Seulgi; Baek, Jae Suk; Jeong Jin Yu; Seo, Go Hun; et al. BMC Medical Genomics; London Vol. 16, (2023) 1-10.

In vitro assessment of the pathogenicity of the LDLR c.2160delC variant in familial hyper...  
Lin, Shaoyi; Hu, Tingting; Wang, Kaihan; Wang, Jiaqi; Zhu, Yunyun; et al. Lipids in Health and Disease; London Vol. 22, (2023) 1-10.

# دسترسی به گزینه های خروجی و ذخیره

گزینه های خروجی

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## TGFBRI Variants Can Associate with Non-Syndromic Congenital Heart Disease without Aortopathy

Alaamery, Manal; Nour Albeshier, Alhabahan, Fahad; Barnett, Phil; Kabbani, Mohamed Salim; et al. Journal of Cardiovascular Development and Disease; Basel Vol. 10, Iss. 11, (2023): 455.  
DOI:10.3390/jcdd10110455

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### Abstract

Translate

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More

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1. Introduction

### Suggested sources

Search with indexing terms

#### Subject

- Cardiovascular disease
- Plasmids
- Families & family life
- Genomes
- Heart
- Genes
- Congenital diseases

#### Location

- Netherlands
- United States--US

# دسترسی به گزینه های خروجی و ذخیره

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## Can Associate with Non-Syndromic

abshah, Fahad; Barnett, Phil; Kabbani, Mohamed Salim; et al. Journal of C

### Abstract

Translate ▾

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1. Introduction

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**Suggested sources**

Genetic **heterog** correlation with Mi Jin Kim; Cha, S al. BMC Medical G

cause of mortality osis. Here, we Next-generation stigate the ase domain of (398H). Both ptein. none of the c CHD without ement of

**In vitro** assessm c.2160delC varia Lin, Shaoyi; Hu, Tir al. **Lipids in Health**

Identification of in **ABCD1** Zheng, Feixia; Lin, al. **Journal of Clini**

Whole Exome Se **Phenotype of Xe** Seo, Ji-In; Nishigor Junglok; et al. **Me**

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# منابع پیشنهادی مرتبط با مدرک بازیابی شده

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## TGFBRI Variants Can Associate with Non-Syndromic Congenital Heart Disease without Aortopathy

Alaamery, Manal; Nour Albeshar; Alhabshan, Fahad; Barnett, Phil; Kabbani, Mohamed Salim; et al. Journal of Cardiovascular Development and Disease; Basel Vol. 10, Iss. 11, (2023): 455. DOI:10.3390/jcdd10110455

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