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فاطمه رفیعی نسب کارشناس اشتراک مایکاه بای کتابخانه مرکزی دانشگاه

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آذر 1402

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Proquestیک سرویس الکترونیکی به هم پیوسته است و محتوای آن را، پایان نامه ها، سخنرانی ها، مقالات،

نشریات علمی و گزارش ها تشکیل می دهند.

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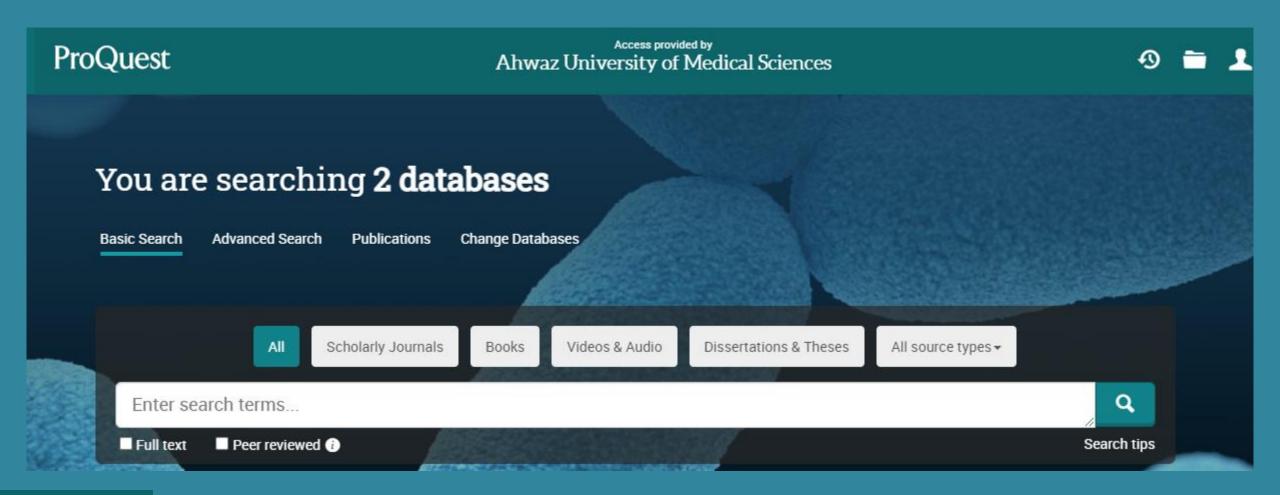
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دسترسی به Proquest



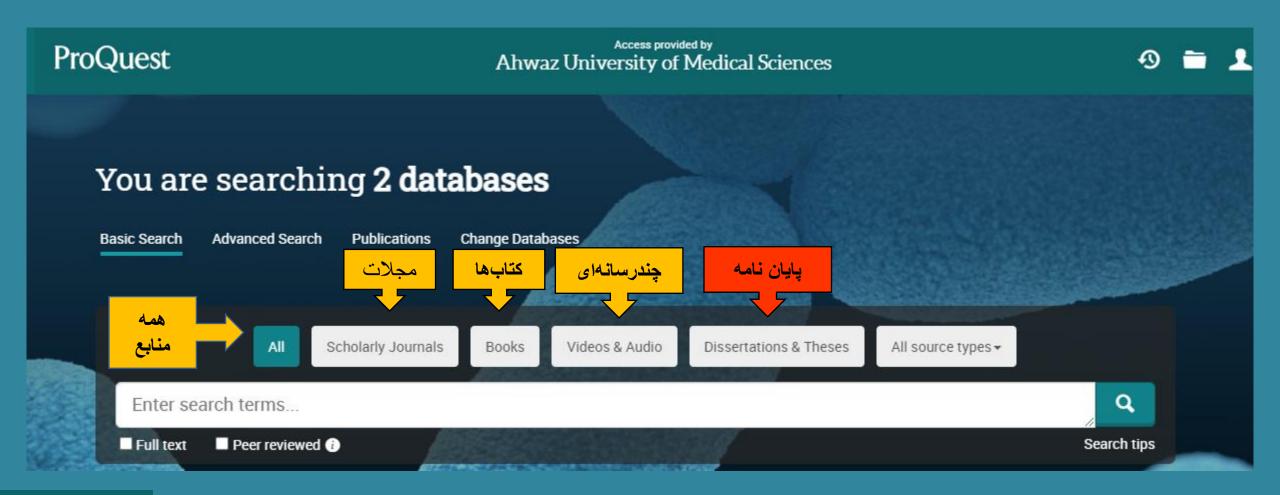
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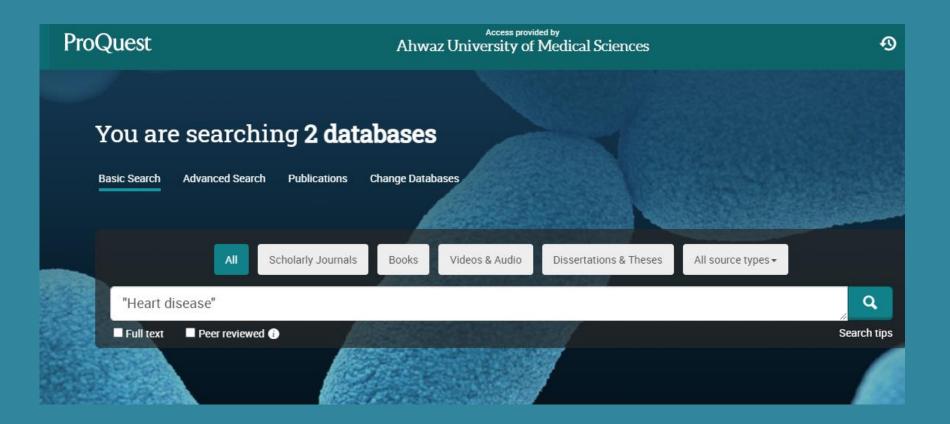


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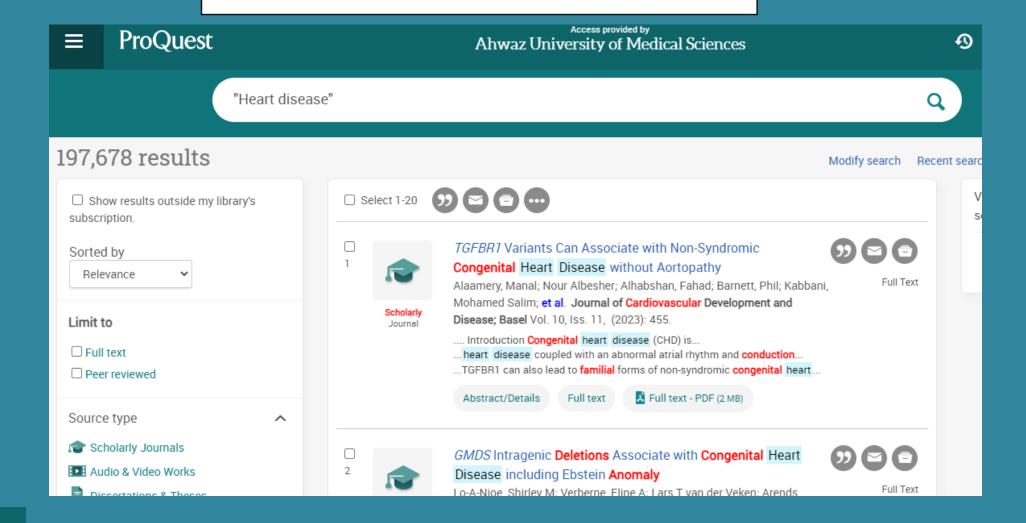


جستجو و مشاهده نتایج

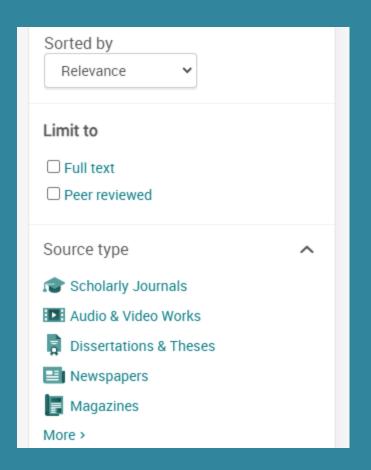
ابتدا کلید واژه و یا عبارت مورد نظر را داخل جعبه جستجو وارد نموده با انتخاب گزینه Searchفرآیند جستجو آغاز می گردد.

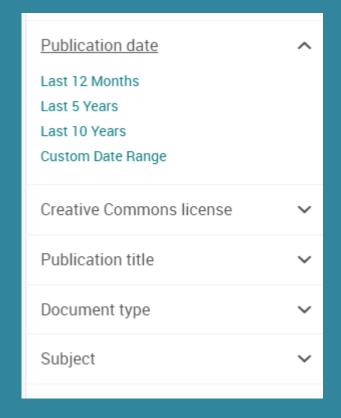


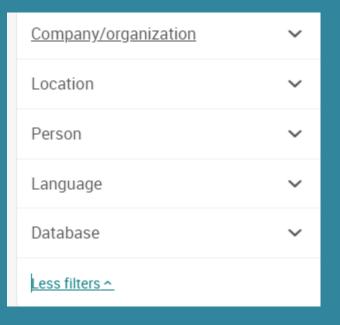
صفحه نتايج



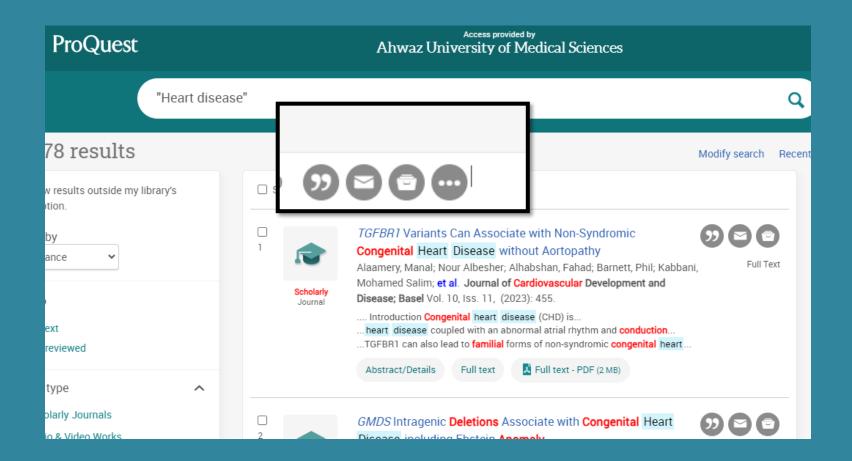
فیلترهای پایگاه







خروجی های پایگاه



مشاهده متن كامل مدارك

دسترسی به متن کامل مقاله













8 Full Text | Scholarly Journal

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

Alaamery, Manal; Nour Albesher; 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

Full text

Full text - PDF

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

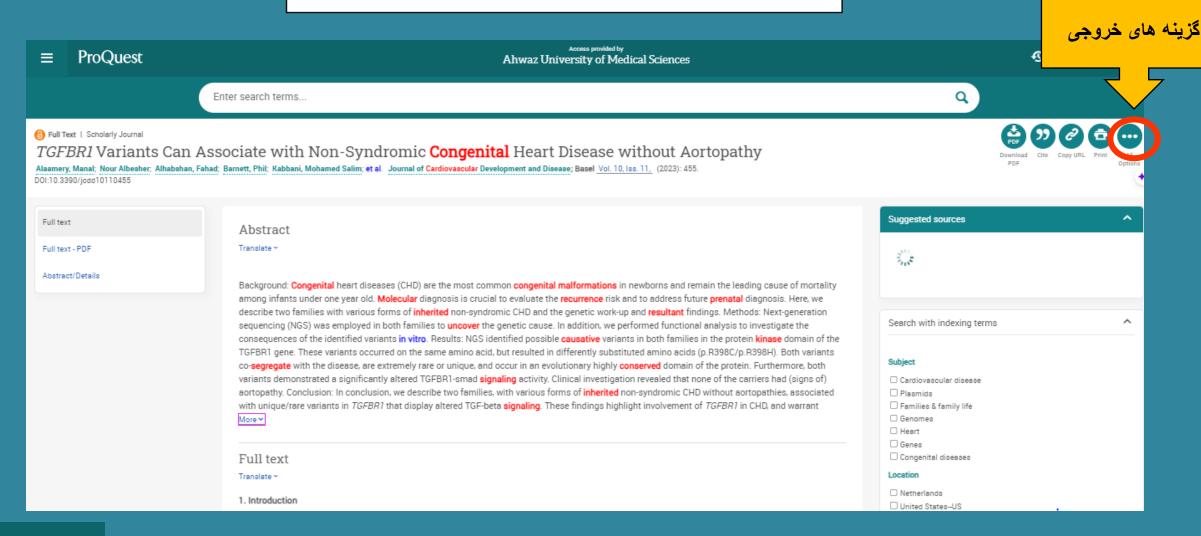
Genetic heterogeneity of cardiomyopathy and its correlation with patient care

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.

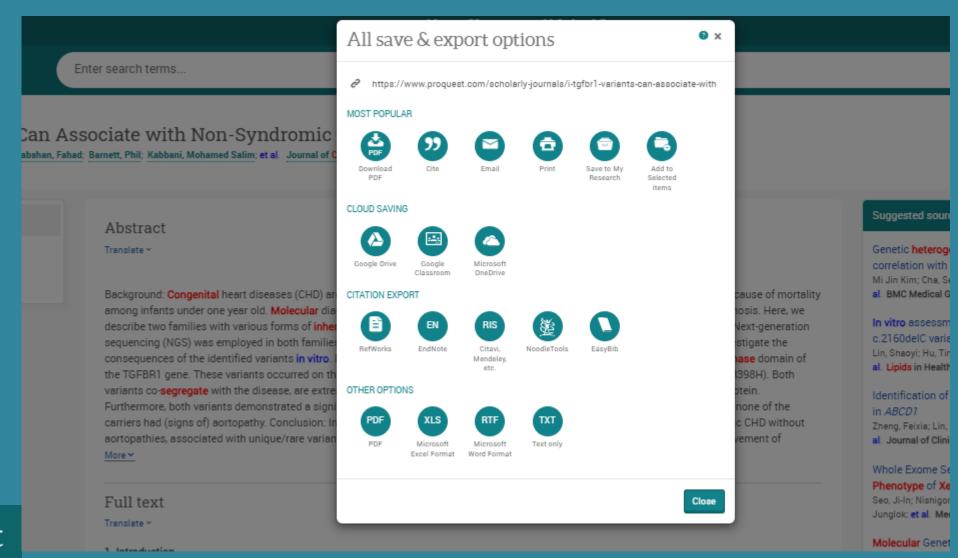
In vitro assessment of the pathogenicity of the LDLR c.2160delC variant in familial hyper

Lin, Shaoyi; Hu, Tingting; Wang, Kaihan; Wang, Jiagi; Zhu, Yunyun; et al. Lipids in Health and Disease;

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دسترسی به گزینه های خروجی و ذخیره



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