

WHITEPAPER

VarSome as Human Genomics Community

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Introduction

As a researcher or medical professional, you may encounter a rare variant which you have never seen before. Collecting information about such a variant may take you a lot of time without any guarantee that you will find what you need. However, if there was another clinician or researcher, anywhere in the world, who has already dealt with the same variant in the past and if there was an easy way to get in touch, well, everything would be much easier. And that's what VarSome as a Human Genomics Community is all about!

VarSome gives every user the opportunity to contribute and share the knowledge on genomic variants for the benefit of the whole community. VarSome offers a massive cross-referenced database of more than 50 integrated public genomics data sets, and we encourage you to further enrich it through your contributions in the form of variant classifications, comments, and publication links. We believe that VarSome with its global community of over 200'000 health care professionals and researchers sharing the knowledge with each other will have a very positive impact on the outcomes of research and diagnostic work of each member of its community, and will improve diagnostic yield of NGS tests and, in turn, improve lives of many patients.



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VarSome's Big Data

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Variant Classifications and Comments

On the free VarSome available at varsome.com you can classify any variant according to the 5-tier classification defined by American College for Molecular Genetics (Richards et al. 2015) and/or start a discussion on any variant of your interest. When classifying a variant you are encouraged to associate it with a phenotype or disease according to the HPO terms. Your classification and/or comment will become public along with your name (i.e. screen name you choose) and the organization you belong to (optional), allowing you to build your reputation as well as the reputation of your organization in the global VarSome community.



Community contributions (1)

Classification: **Likely Pathogenic**

Niemann-Pick Disease

by [e\[redacted\]](#), 09-Feb-2019 [Reply](#)

[Classify this variant](#) or [Link a publication](#) or [Comment on this variant](#)

With VarSome Clinical, the professional editions of the free VarSome for clinicians and researchers alike, you can even set up custom variant classifications, which compared to the free VarSome remain associated only with your account unless you explicitly decide to share it with the global VarSome community. This way, together with sample cross-referencing (see below) you can build over time your private database of samples and variants. In addition to sharing your knowledge privately only with the members of your own team, with VarSome Clinical you can share the data at single variant level with your partner institutions on the condition that both parties consent to data sharing in writing.



BROCHURE

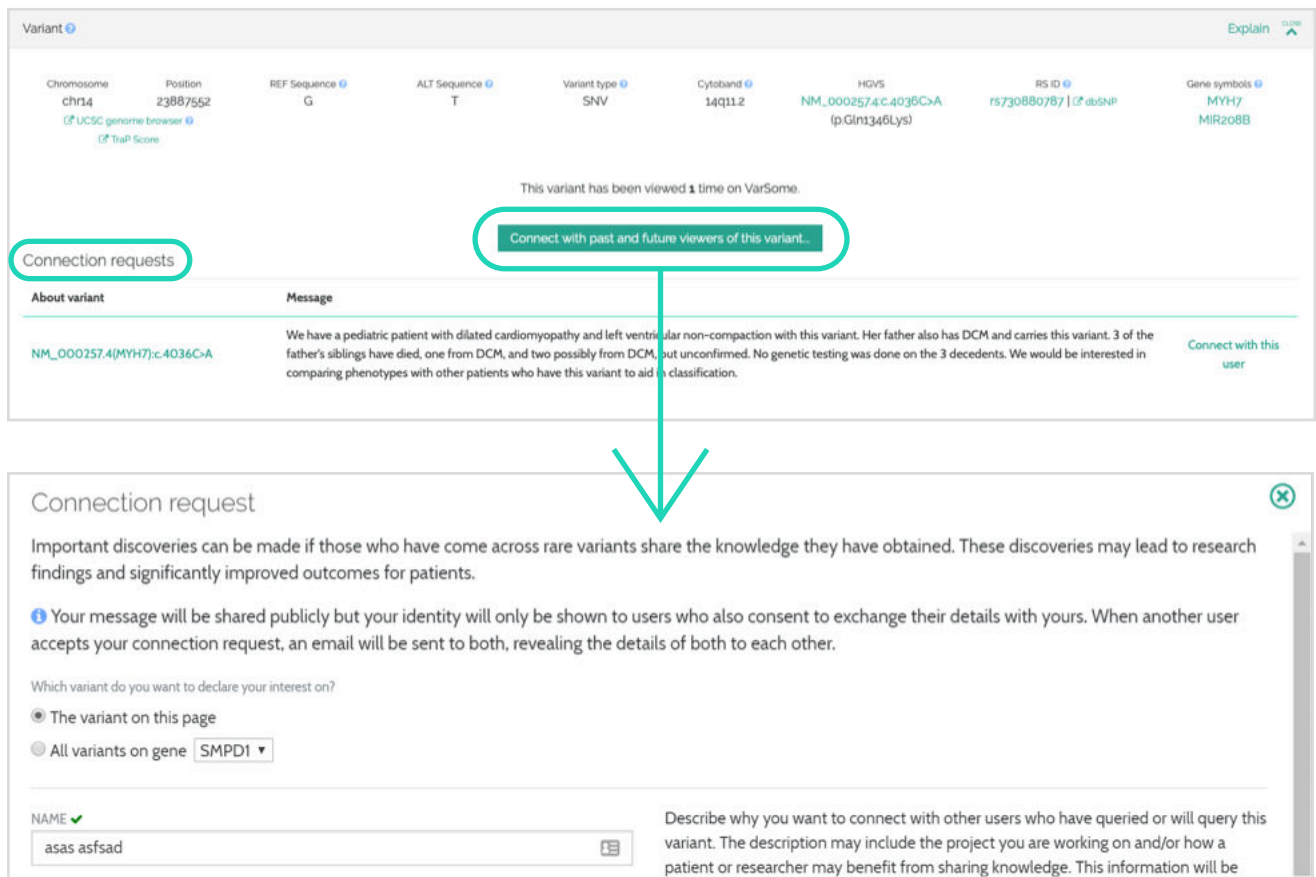
Introducing VarSome
Clinical

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Connect with past and future viewers of the variant

To facilitate information sharing between unrelated VarSome users and organizations you can raise a request to be contacted with past and future viewers of a particular variant of interest. When doing so, you are asked to describe why you want to connect with other users. The description may include the project you are working on and/or how a patient or researcher may benefit from sharing knowledge with you. This information will be visible to all other VarSome users who visit the variant or gene page, so use this space to convince them to connect with you.

Your message will be shared publicly on varsome.com but your identity will only be shown to users who also consent to exchange their details with yours. When another user accepts your connection request, an email will be sent to both, revealing the details of both to each other.



The image shows a screenshot of the VarSome website interface. At the top, there's a 'Variant' header with a 'Close' button. Below it, a table displays variant details: Chromosome (chr14), Position (23887552), REF Sequence (G), ALT Sequence (T), Variant type (SNV), Cytoband (14q11.2), HGVS (NM_000257.4:C.4036C>A (p.Gln1346Lys)), RS ID (rs730880787), and Gene symbols (MYH7, MIR208B). A message states 'This variant has been viewed 1 time on VarSome.' Below this, a button labeled 'Connect with past and future viewers of this variant...' is highlighted with a red circle. To the left, a 'Connection requests' section is also highlighted. A red arrow points from the button to a 'Connection request' form. The form includes a title 'Connection request', a paragraph about sharing knowledge, a note about public sharing and consent, a question 'Which variant do you want to declare your interest on?' with radio buttons for 'The variant on this page' (selected) and 'All variants on gene' (with a dropdown menu showing 'SMPD1'), a 'NAME' field with a green checkmark and the text 'asas asfsad', and a text area for describing why you want to connect.

Variant [Close](#)

Chromosome	Position	REF Sequence	ALT Sequence	Variant type	Cytoband	HGVS	RS ID	Gene symbols
chr14	23887552	G	T	SNV	14q11.2	NM_000257.4:C.4036C>A (p.Gln1346Lys)	rs730880787 dbSNP	MYH7 MIR208B

[UCSC genome browser](#) [Ensembl](#) [TraP Score](#)

This variant has been viewed 1 time on VarSome.

[Connect with past and future viewers of this variant...](#)

Connection requests

About variant	Message	Connect with this user
NM_000257.4(MYH7):c.4036C>A	We have a pediatric patient with dilated cardiomyopathy and left ventricular non-compaction with this variant. Her father also has DCM and carries this variant. 3 of the father's siblings have died, one from DCM, and two possibly from DCM, but unconfirmed. No genetic testing was done on the 3 decedents. We would be interested in comparing phenotypes with other patients who have this variant to aid in classification.	Connect with this user

Connection request [Close](#)

Important discoveries can be made if those who have come across rare variants share the knowledge they have obtained. These discoveries may lead to research findings and significantly improved outcomes for patients.

i Your message will be shared publicly but your identity will only be shown to users who also consent to exchange their details with yours. When another user accepts your connection request, an email will be sent to both, revealing the details of both to each other.

Which variant do you want to declare your interest on?

☒ The variant on this page

☐ All variants on gene [SMPD1](#)

NAME [✓](#)

asas asfsad

Describe why you want to connect with other users who have queried or will query this variant. The description may include the project you are working on and/or how a patient or researcher may benefit from sharing knowledge. This information will be

Sample cross-referencing

Another powerful feature is sample cross-referencing, available only in VarSome Clinical, the CE-IVD-certified AND HIPSS-compliant fully-fledged platform for analysis of NGS data. VarSome Clinical cross-references all your samples, whether they are gene panels, exomes or genomes, and links all your samples through common variants. This, together with custom classifications and comments, allows you to build your own database of samples and variants. As mentioned before, you may even share data at single variant level with partner institution(s) on the condition that all parties consent in writing. With VarSome Clinical you can also import your own local database with allele frequencies and use it privately for variant annotation and classification of your samples.



Get involved!

As you can see, there are many ways how you can participate in the global VarSome community of 200'000 users. VarSome is the Human Genomics Community backed by the expertise of our multi-disciplinary team, and we would appreciate any feedback you may provide. Any suggestion is more than welcome! For example, we are working on a feature allowing you to subscribe to a variant or a gene, meaning whenever there is a new piece of information on a variant or gene available you will get an instant notification. What do you think, is this feature going to be useful for you?

Further learning resources



VarSome Clinical is a CE-IVD-certified and HIPAA-compliant platform allowing fast and accurate variant discovery, annotation, and interpretation of NGS data for whole genomes, exomes, and gene panels. VarSome Clinical helps molecular geneticists and clinicians reach faster and more accurate diagnoses and treatment decisions for genetic conditions.

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No delays, premium features & data resources! AACT, COSMIC, Polyphen-2, CADD, OncoKB, CKB, PharmaGKB, and more.

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