

**UNIVERSIDADE FEDERAL DE VIÇOSA
CENTRO DE CIÊNCIAS AGRÁRIAS
DEPARTAMENTO DE ZOOTECNIA**

TUTORIAL

VarElect: The Next Generation Sequencing Phenotyper

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VarElect

Com uma quantidade muito grande de genes identificados associados com um(a) fenótipo/doença, o VarElect ajuda a reduzir essa lista buscando genes candidatos mais prováveis de estarem associados com a característica estudada.

Mais informações no site: <https://varelect.genecards.org/>

Vídeo com instruções (em inglês): <https://youtu.be/rH1L4dGIS4g>

VarElect



LOG IN SIGN UP

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The Next Generation Sequencing Phenotyper

Rapid prioritization of variant genes based on disease/phenotype of interest

SIGN UP FREE

Extensive disease – gene relationship data

Powered by LifeMap's GeneCards Suite and its extensive phenotype - gene relationships annotations.

Both direct, and indirect disease - gene links

Unlike other platforms, leverages unique, extensive data from the LifeMap Integrated Knowledgebase to infer direct, as well as indirect links, between genes and diseases/phenotypes.



Fazer login ou criar uma conta



Integrated
Biomedical
Knowledgebase



Premium Tools



Contact us

Log in to VarElect via the GeneCards Suite

Log in

[Don't have an account? Sign up for free!](#)

Email:

Password:

Remember Me

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Log In

Or log in with:



Sign up to VarElect and the GeneCards Suite

Sign up

[Registered user? Please log in](#)

First Name:

Last Name:

 Profile Type:

Academic

Company

Email:

 Password:

Confirm Password:

Next

Para professores e alunos, cadastrar usando o e-mail institucional

Sign up to VarElect and the GeneCards Suite

Sign up for VarElect

Country:

Organization:

Universidade Federal de Viçosa

Department:

Enter your department

Position:

Enter your position

Phone Number:

Enter your phone number

Where did you hear about us?

By clicking Join Now,
You agree to our [Terms & Conditions](#).

Join Now

Telefone não é de preenchimento obrigatório

Um código será enviado para seu e-mail

Sign up to VarElect and the GeneCards Suite

Dear [REDACTED],

Thank you for registering and Welcome to GeneCards Suite!

We have sent a verification code to [REDACTED]. Please validate this address by entering that code.

* **Verification Code:**

If you did not receive the verification code, you can resend the verification code or correct your email address by clicking on a button below.

or

If you are having trouble validating your account, please [contact our technical support](#).

Sign up to VarElect and the GeneCards Suite

Thank you for registering with VarElect

You will be redirected back to VarElect in 2 seconds

[Click here to go there now](#)

[Need help? contact our Customer Support](#)

Página inicial do VarElect

GeneCardsSuite GeneCards GeneCaRNA MalaCards PathCards **VarElect** GeneAnalytics GeneALaCart GenesLikeMe

 **VarElect**
NGS PHENOTYPER

About 

Examples: diarrhea | "capillary leak" | "cleft palate" My Analyses: **Open**

Start new Analysis

Rank genes related to phenotype and disease terms based on GeneCards and MalaCards
Analyze variants within GeneHancer regulatory elements ([learn more](#))
Need programmatic access via VarElect API? [Contact us](#)

1 Enter/Paste Gene/GeneHancer Symbols [?](#) [Upload File](#)

2 Enter Phenotype Keywords [?](#)

Start typing Phenotypes/Keywords.

Query output...

3 Limit to specific GeneCards section (Optional) [?](#)

Everywhere ▼

4 Enter Exclusion Phenotype Keywords (Optional) [?](#)

Start typing Phenotypes/Keywords to Exclude.

Query output...

5 **Remove results related to inferred diseases and common publications**

- Inserir a lista de genes no campo (1)
- Clicar em “*Symbolize*”

The screenshot shows the 'Enter/Paste Gene/GeneHancer Symbols' input field with a list of gene symbols: CRKL, AIFM3, LZTR1, THAP7, TUBA3E, LRRC74B, P2RX6, SLC7A4, MZT2B, SMPD4, MED15, KLHL22, SCARF2, ZNF74, LARP1B, ABHD18, MFSD8, PLK4, HSPA4L, SLC25A31, INTU, KLHL1, GAS2, HORMAD2, LIF, OSM. Below the input field is a 'Symbolize' button. The resulting table shows 111 genes ready for analysis.

Symbol	Name	
ABCC11	ATP Binding Cassette Subfamily C Member 11	×
ABHD18	Abhydrolase Domain Containing 18	×
ACSS2	Acyl-CoA Synthetase Short Chain Family Member 2	×
AIFM3	Apoptosis Inducing Factor Mitochondria Associated 3	×

This close-up shows the 'Symbolize' button highlighted with a red box. The input field above it contains the same list of gene symbols as the main screenshot.

- Depois de clicar em “*Symbolize*” ele vai mostrar os genes que foram identificados em “*READY FOR ANALYSIS*”
- Se algum gene não foi identificado, estará na coluna “*UNIDENTIFIED*”

→ Somente à exemplo, abaixo estão genes que não seriam identificados

→ No caso, são microRNAs e ORF (*open reading frame*), uma fase de leitura aberta não identificada

1 Enter/Paste Gene/GeneHancer Symbols [?](#) [Upload File](#)

MZT2B, SMPD4, MED15, KLHL22, SCARF2, ZNF74, LARP1B, ABHD18, MFSD8, PLK4, HSPA4L, SLC25A31, INTU, KLHL1, GAS2, HORMAD2, LIF, OSM, bta-mir-12063, bta-mir-499, bta-mir-130b, bta-mir-301b, C17H5orf52

Symbolize

READY FOR ANALYSIS (111) **UNIDENTIFIED (5)**

Unidentified: 5 gene(s)

C17H5orf52	Not found in GeneCards		
bta-mir-12063	Not found in GeneCards		
bta-mir-130b	Not found in GeneCards		
bta-mir-301b	Not found in GeneCards		
bta-mir-499	Not found in GeneCards		

- Inserir o/s fenótipo/s no campo (2)
- Verificar mais detalhes nas (?), se necessário
- Os campos 3, 4, 5 e 6 são opcionais
- Clicar em “Analyze”

1 Enter/Paste Gene/GeneHancer Symbols ? Upload File

CRKL, AIFM3, LZTR1, THAP7, TUBA3E, LRRC74B, P2RX6, SLC7A4, MZT2B, SMPD4, MED15, KLHL22, SCARF2, ZNF74, LARP1B, ABHD18, MFSD8, PLK4, HSPA4L, SLC25A31, INTU, KLHL1, GAS2, HORMAD2, LIF, OSM

Symbolize

READY FOR ANALYSIS (111)
UNIDENTIFIED (0)

Ready for Analysis: 111 gene(s)

Symbol	Name	
ABCC11	ATP Binding Cassette Subfamily C Member 11	✕
ABHD18	Abhydrolase Domain Containing 18	✕
ACSS2	Acyl-CoA Synthetase Short Chain Family Member 2	✕
AIFM3	Apoptosis Inducing Factor Mitochondria Associated 3	✕

2 Enter Phenotype Keywords ?

thermoregulation AND "heat resistance" ✓

thermoregulation AND "heat resistance"

3 Limit to specific GeneCards section (Optional) ?

Everywhere

4 Enter Exclusion Phenotype Keywords (Optional) ?

Start typing Phenotypes/Keywords to Exclude.

Query output...

5 **Remove results related to inferred diseases and common publications**

6 **Mark cancer census genes in results for cancer related queries**

When searching, follow the rules below for optimal results.

- Enclose multi-word terms within quotation marks (e.g. "serotonin receptor")
- Use AND/OR to refine a multi-term search (e.g. "Glycerolipid metabolism" AND "execution phase of apoptosis")
- Delimiters (e.g. comma, tab, space, newline, slash) which are not within quoted strings are interpreted as OR

Reset

Analyze

Click to find direct and indirect relations between genes and phenotypes.

- A princípio, o próprio site passa um tutorial para você verificar os resultados da análise

The screenshot displays a web application interface with a table of results. At the top, there are three tabs: "DIRECTLY RELATED (0)", "INDIRECTLY RELATED (18)" (which is highlighted in purple), and "UNIFIED RESULTS (18)". Below the tabs is a search bar with the text "Enter filter text" and a magnifying glass icon. The table has columns for "#", "Symbol", "Description", "Type", "Score", and "Average Disease Causing Likelihood". A tutorial overlay is present in the center of the screen, titled "VarElect Tutorial : Results". The overlay contains the text: "This tutorial will guide you through the results of your query. Click on NEXT and PREV buttons to navigate this tutorial." Below the text are two buttons: "« Prev" and "Next »", and a button labeled "End tour".

#	Symbol	Description	Type	Score	Average Disease Causing Likelihood
1	ITCH		Protein	0.14	85.6
2	P2RX6		Protein	0.05	32.1
3	TRPC4AP		Protein	0.04	78.9
4	ABCC11		Protein	0.02	1.8
5	GDF5	Growth Differentiation Factor 5	Protein	0.02	42.7
6	GAS2	Growth Arrest Specific 2	Protein	0.02	55.3
7	MORC2	MORC Family CW-Type Zinc Finger 2	Protein	0.01	80.8
8	HSPA4L	Heat Shock Protein Family A (Hsp70) Member 4 Like	Protein	0.01	53.5
9	OSM	Oncostatin M	Protein	0.01	45.7
10	NCOA6	Nuclear Receptor Coactivator 6	Protein	0.01	62.6
11	KLF15	Kruppel Like Factor 15	Protein	0	64.8

- “My analyses”:
- Use “Save” e “Save As” para salvar os resultados das análises
- Use “Open” para abrir análises que já foram salvas

The screenshot shows the top navigation bar with buttons for 'My Analyses: Save', 'Save As', 'Download Results', 'New', and 'Open'. Below this, the 'ANALYZED SYMBOLS: 111' section is visible, with tabs for 'PHENOTYPES', 'ALL (111 SYMBOLS)', 'DIRECTLY RELATED (0 SYMBOLS)', and 'INDIRECTLY RELATED'. The 'Query' field contains 'thermoregulation AND "heat resistance"', and the 'Limit to' dropdown is set to 'Everywhere'. The 'Exclude' field contains 'Phenotypes to Exclude'. A tooltip titled 'My Analyses' is overlaid on the interface, containing the text: 'Use the 'Save' and 'Save As' buttons to save your analyses and the 'Open' button to open previously saved ones.' The tooltip also features navigation buttons: '< Prev', 'Next >', and 'End tour'.

- Nesta parte tem os detalhes da análise: fenótipos incluídos e excluídos e mais...

The screenshot shows the 'Query Details' section of the interface. The top navigation bar is identical to the previous screenshot. Below it, the 'ANALYZED SYMBOLS: 111' section is visible, with tabs for 'PHENOTYPES', 'ALL (111 SYMBOLS)', 'DIRECTLY RELATED (0 SYMBOLS)', 'INDIRECTLY RELATED (18 SYMBOLS)', and 'UNRELATED (93 SYMBOLS)'. The 'Query' field contains 'thermoregulation AND "heat resistance"', and the 'Limit to' dropdown is set to 'Everywhere'. The 'Exclude' field contains 'Phenotypes to Exclude'. A 'Reanalyze' button is visible to the right of the query field. A tooltip titled 'Query Details' is overlaid on the interface, containing the text: 'Information on the query that was run.' The tooltip also features navigation buttons: '< Prev', 'Next >', and 'End tour'.

- Em “*DIRECTLY RELATED*” terá a lista de genes **diretamente** relacionados com o fenótipo estudado

*Como este é só um exemplo aleatório, nenhum gene diretamente relacionado com o/s fenótipo/s foi encontrado

ANALYZED SYMBOLS: 111 [Hide](#)

PHENOTYPES [ALL \(111 SYMBOLS\)](#) [DIRECTLY RELATED \(0 SYMBOLS\)](#) [INDIRECTLY RELATED \(18 SYMBOLS\)](#) [UNRELATED \(93 SYMBOLS\)](#)

Query: thermoregulation AND "heat resistance"

Limit to: Everywhere

Reanalyze

DIRECTLY RELATED (0) [INDIRECTLY RELATED \(18\)](#)

Direct Results
Information on genes directly related to the phenotype, including score and gene description

« Prev Next » End tour

No Data Found

- Em “*INDIRECTLY RELATED*” terá a lista de genes **indiretamente** relacionados com a característica estudada
- Esta parte é especialmente relevante quando nenhuma ligação direta foi encontrada
- A associação indireta entre estes genes e o fenótipo pode ser por meio de:
 - Vias compartilhadas (*shared pathways*), interação de proteína (*protein interaction*), relação de genes parálogos (*paralog relation*) e/ou publicações mútuas (*mutual publication*)

ANALYZED SYMBOLS: 111 [Hide](#)

PHENOTYPES [ALL \(111 SYMBOLS\)](#) [DIRECTLY RELATED \(0 SYMBOLS\)](#) [INDIRECTLY RELATED \(18 SYMBOLS\)](#) [UNRELATED \(93 SYMBOLS\)](#)

Query: [Reanalyze](#)

Limit to:

Indirect Results

Information on genes indirectly related to the phenotype, including score and gene description

[« Prev](#) [Next »](#) [End tour](#)

[DIRECTLY RELATED \(0\)](#) **[INDIRECTLY RELATED \(18\)](#)**

Enter filter text

<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>
#	Symbol	Description	Type	Score	Average Disease Causing Likelihood	
1	ITCH	Itchy E3 Ubiquitin Protein Ligase	Protein	0.14	85.6	<div style="width: 85.6%;"></div>
2	P2RX6	Purinergic Receptor P2X 6	Protein	0.05	32.1	<div style="width: 32.1%;"></div>
3	TRPC4AP	Transient Receptor Potential Cation Channel Subfamily C Member 4 Associated Protein	Protein	0.04	78.9	<div style="width: 78.9%;"></div>
4	ABCC11	ATP Binding Cassette Subfamily C Member 11	Protein	0.02	1.8	<div style="width: 1.8%;"></div>
5	GDF5	Growth Differentiation Factor 5	Protein	0.02	42.7	<div style="width: 42.7%;"></div>

- Ao clicar no sinal de + ao lado esquerdo do nome de cada gene, a linha será expandida mostrando detalhes da relação do gene com a característica

Implicating Genes

Expand a row of a gene appearing in the indirect results tab, to view implicating genes that connect it to the phenotype

« Prev Next » End tour

	Rank	Gene	Description	Type	Score	Average Disease Causing Likelihood
	1	ITCH	Itchy E3 Ubiquitin Protein Ligase	Protein	0.14	85.6
Implicating Genes for ITCH:						
		+ TRPV1	Transient Receptor Potential Cation Channel Subfamily V Member 1	Protein	0.14	55.2
	2	+ P2RX6	Purinergic Receptor P2X 6	Protein	0.05	32.1

- É possível expandir e encontrar mais detalhes de publicações que mencionaram o gene

Hit Context MiniCard

Expand an implicating gene row to view hit contexts in a MiniCard

« Prev Next » End tour

	Rank	Gene	Description	Type	Score	Average Disease Causing Likelihood
		- TRPV1	Transient Receptor Potential Cation Channel Subfamily V Member 1	Protein	0.14	55.2

TRPV1 → ITCH Gene Relation ?

Publications (5/7) See All

- [Excitation and modulation of TRPA1, TRPV1, and TRPM8...neurons by the pruritogen chloroquine.](#) (PMID: 23508958)
Abstract: The sensations of pain, *itch*, and cold often interact with each other. Pain inhibits *itch*, whereas cold inhibits both pain and *itch*. TRPV1 and TRPA1 channels transduce pain and *itch*, whereas TRPM8 transduces cold. The pruritogen chloroquine...reported to excite TRPA1, leading to the sensation of *itch*. It is unclear how CQ excites and modulates TRPA1(+)...neurons and thus affects the sensations of pain, *itch*, and cold. Here, we show that only 43% of CQ-excited...acting together with TRPA1 to mediate CQ-induced *itch*. CQ not only elicits *itch* by directly exciting *itch*-encoding neurons but also exerts previously unappreciated widespread actions on pain-, *itch*-, and cold-sensing neurons, leading to enhanced pain and *itch*.
- [Cutaneous innervation before and after one treatment period of acupuncture.](#) (PMID: 17034527)
Abstract: The effect of acupuncture on nociceptive pain is well documented, but effects on nociceptive *itch* have been contradictory. To evaluate possible effects...unmyelinated nerve fibres that transmit nociceptive pain and *itch*. A histamine prick test using planimetry was used to record experimental *itch* after acupuncture on the treated area and on the corresponding...and a visual analogue scale was used to evaluate *itch*. The mean +/- SEM number of CGRP-immunoreactive nerve...antibodies used in this study. Neither histamine-induced *itch* nor cutaneous responses were influenced by acupuncture...indicate an effect of acupuncture on neuropathic *itch* but not histamine-mediated *itch*. Our findings support the opinion that the pain-relieving...depend on its effect on the peripheral innervation.
- [Facilitation of TRPV4 by TRPV1 is required for *itch* transmission in some sensory neuron populations.](#) (PMID: 27432350)

Dos 111 genes incluídos na análise, 18 foram selecionados

DIRECTLY RELATED (0)		INDIRECTLY RELATED (18)		UNIFIED RESULTS (18)	
Enter filter text					
#	Symbol	Description	Type	Score	Average Disease Causing Likelihood
1	ITCH	Itchy E3 Ubiquitin Protein Ligase	Protein	0.14	85.6
2	P2RX6	Purinergic Receptor P2X 6	Protein	0.05	32.1
3	TRPC4AP	Transient Receptor Potential Cation Channel Subfamily C Member 4 Associated Protein	Protein	0.04	78.9
4	ABCC11	ATP Binding Cassette Subfamily C Member 11	Protein	0.02	1.8
5	GDF5	Growth Differentiation Factor 5	Protein	0.02	42.7
6	GAS2	Growth Arrest Specific 2	Protein	0.02	55.3
7	MORC2	MORC Family CW-Type Zinc Finger 2	Protein	0.01	80.8
8	HSPA4L	Heat Shock Protein Family A (Hsp70) Member 4 Like	Protein	0.01	53.5
9	OSM	Oncostatin M	Protein	0.01	45.7
10	NCOA6	Nuclear Receptor Coactivator 6	Protein	0.01	62.6
11	KLF15	Kruppel Like Factor 15	Protein	0	64.8
12	MYH7B	Myosin Heavy Chain 7B	Protein	0	32.0
13	MAP1LC3A	Microtubule Associated Protein 1 Light Chain 3 Alpha	Protein	0	75.6
14	CLDN14	Claudin 14	Protein	0	53.4
15	HAPLN1	Hyaluronan And Proteoglycan Link Protein 1	Protein	0	55.3
16	LIF	LIF Interleukin 6 Family Cytokine	Protein	0	55.2
17	VCAN	Versican	Protein	0	16.7
18	CRKL	CRK Like Proto-Oncogene, Adaptor Protein	Protein	0	81.6

DIRECTLY RELATED (0)

INDIRECTLY RELATED (18)

UNIFIED RESULTS (18)

Enter filter text

#	Symbol	Description	Type	Score	Average Disease Causing Likelihood
1	ITCH	Itchy E3 Ubiquitin Protein Ligase	Protein	0.14	85.6
2	P2RX6	Purinergic Receptor P2X 6	Protein	0.05	32.1
3	TRPC4AP	Transient Receptor Potential Cation Channel Subfamily C Member 4 Associated Protein	Protein	0.04	78.9
4	ABCC11	ATP Binding Cassette Subfamily C Member 11	Protein	0.02	1.8

- No cabeçalho da tabela temos:
- **Symbol:** o símbolo do gene
- **Description:** a descrição/nome do gene
- **Type:** Tipo de gene, exemplo codificador de proteína (*protein-coding*), Pseudogene, gene RNA, Família de gene (gene cluster), Locus genético (*genetic locus*) ou não categorizado (*uncategorized*).
- **Score:** * próximo slide*
- **Average Disease Causing Likelihood:** A coluna Probabilidade de Causar Doenças reflete o princípio de que uma variante em um gene com alta intolerância a mutações tem maior probabilidade de causar doenças. RVIS é o escore de intolerância à variação residual (Petrovski et al) e GDI é o Índice de Danos Genéticos (Itan et al). As probabilidades de causar doenças são 100% menos o percentil RVIS ou 100% menos o percentil GDI (barras laranja), com a média de ambos mostrada numericamente. ND - não determinado, NA - não aplicável.

DIRECTLY RELATED (0)

INDIRECTLY RELATED (18)

UNIFIED RESULTS (18)

Enter filter text

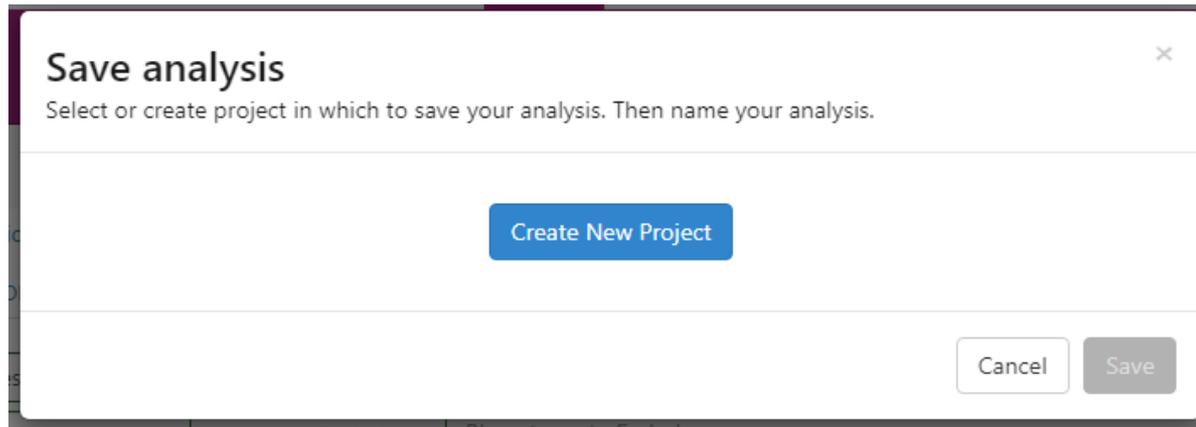


#	Symbol	Description	Type	Score	Average Disease Causing Likelihood
1	ITCH	Itchy E3 Ubiquitin Protein Ligase	Protein	0.14	85.6
2	P2RX6	Purinergic Receptor P2X 6	Protein	0.05	32.1
...	TRPC4AP	Transient Receptor Potential Cation Channel Subfamily C Member 4

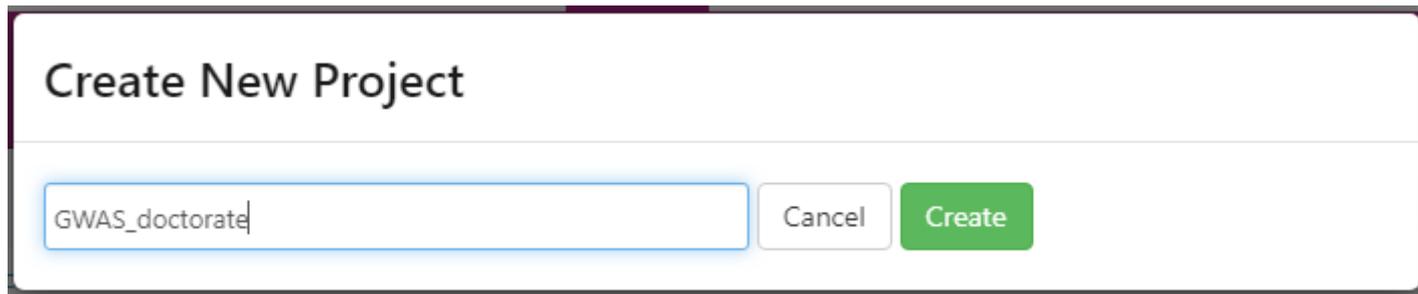
- **Score:** Essa pontuação é uma indicação da força da conexão entre o gene e o(s) fenótipo(s).
- As pontuações geralmente variam de 1 a 200.
- O objetivo principal da pontuação é permitir classificar e priorizar a lista de genes consultados por relevância para o(s) fenótipo(s) consultado(s).
- As pontuações de diferentes análises não podem e não devem ser comparadas, uma vez que as pontuações de relevância produzidas para cada análise são relativas dentro da análise e não absolutas.
- A barra verde representa a proporção de cada pontuação/pontuação máxima da análise atual.
- Ao analisar os elementos reguladores do GeneHancer, a pontuação do fenótipo é ajustada da seguinte forma: Cada associação gene-GeneHancer tem uma pontuação total, calculada pela multiplicação da pontuação de confiança GeneHancer pela pontuação da associação GeneHancer-Gene. As pontuações totais são normalizadas para um intervalo de 0,05 - 0,8 , e cada pontuação de gene-fenótipo do alvo do gene do elemento regulador é multiplicada pela pontuação total de GeneHancer normalizada.

Para salvar os resultados da análise

- Ir em “*My analyses*”, clicar em “*Save*”
- Clicar em “*Create New Project*” para criar um novo projeto, ou se já tiver um, basta apenas selecioná-lo



- Após descrever o nome do projeto, clicar em “*Create*”



- Definir um nome para a análise realizada e clicar em “Save”

The image shows a 'Save analysis' dialog box with a close button (X) in the top right corner. Below the title, there is a subtitle: 'Select or create project in which to save your analysis. Then name your analysis.' The dialog is divided into two main sections. The left section is titled 'Projects (1)' and contains a 'New' button and a list of projects. One project, 'GWAS_doctorate', is listed in a text box. The right section is titled 'GWAS_doctorate' and contains the text 'There are no saved Analyses in this project'. Below this text is a section titled 'Enter name for this analysis' with a text input field containing the placeholder text 'Name'. At the bottom of the dialog, there are three buttons: 'Manage', 'Cancel', and 'Save'.

Save analysis ×

Select or create project in which to save your analysis. Then name your analysis.

Projects (1) New **GWAS_doctorate**

GWAS_doctorate

There are no saved Analyses in this project

Enter name for this analysis

Name

Manage Cancel Save

- As descrições originais sobre o **Score** e **Average Disease Causing Likelihood** estão nas (?) acima de cada item.
- Para mais detalhes e informações, verificar o site: <https://varelect.genecards.org/>
- Links úteis:
- <https://varelect.genecards.org/about/#enhancers>
- <https://youtu.be/rH1L4dGIS4g>