Genome-Phenome Report

Export report quickly and easily to lab's reporting platform using SimulConsult's API, or use cover letter with the default HTML output

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ed SimulConsult® software, database © 1998-2016 and pognosis tables © 2016. All

Report Features

- Clinical rationale for the diagnosis is clear
- Supports up to two diagnoses; the findings automatically group by diagnosis
- **Prognosis table** answers the clinician and patient question "what should I expect"
- **Contextual resource links** for the clinician and patient by diagnosis leverage the time the provider and patient spend together
- Structured terms (ICD 10, SNOMED, IMO[®] lexical ID) are available automatically via a partnership with Intelligent Medical Objects®, including patient-friendly terms for various medical concepts and HPO terms available for findings. (Orphanet available soon)

Genome-Phenome Analyzer

Analyze and interpret panel, exome and genome variant tables in the full clinical context to rapidly identify causative genes and variants

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Fast. In under 5 seconds, identify the pertinent gene or genes and their variants that could be causative in the patient, identified in a way that is hypothesis-independent as to mode of inheritance, number of genes involved and key clinical findings. Workflow allows clinical interpretation and reporting to be done in just a few minutes.

Accurate. Uses gene pertinence, a measure of confidence in the diagnosis. The top gene in confirmed diagnosis typically has >99% pertinence. (PMID 25156663, open access)

Cost effective. Reduce the need to do trios to diagnose known diseases. Make good use of scarce time of experts on clinical interpretation



ley Features

Clinical correlation of patient findings and genomic results identifies the most pertinent genes from among >6,100 described disorders, highlighting those most plausible as causative variants

Incidental findings with options of gene lists and individuals to report

Discovery gene list for genes with phenotypes not yet described in the literature

Genome Report workflow enables fast selection of diagnoses, genes and variants and export to reporting platform, including a Prognosis Table[®] to answer the question "what should I expect"

Complete coverage of known Mendelian disorders and many CNV disorders

Key Benefits

To set up a trial and order

genome@simulconsult.com 857-205-2914 (mobile) 617-879-1670 (office)

Analysis Workflow

MD orders test Findings can be collected in 1 of 3 ways



Genome-Phenome Analysis

Differential diagnosis	Add findings A	dd tests Phenotype	Genotype	Advanc	ed mode	Pertine	ence me	easure
Diseases	om the patient's 47 gene	variants:Set	variant parameters			for the	gene (g	reen
VLDLR-related cerebel	tinent cone veriente		Finding color key:			101 0110	8,000 (8	,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,,
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The order gene test butto	n at right has hyperinks to	o a variety of resources		Order gene	test	manid i		tation
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					Zy	osity		
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Repo. 4 NM 005529:ex6	CA3239C0.11747P	chr1:22168845	missense	50	0	50	0.0383	
Repo. 1 NM 005529:ex60):c.C7806Ap.V2602V	chr1:22174518	synonymous	50	100	50	0.0471	
Repo., 1 NM_005529:ex50):c.G6402A:p.V2134V	chr1:22180723	synonymous	50	50	50	0.0416	
Repo., 1 NM_005529:ex5	:c.G6552A:p.T2184T	chr1:22179451	synonymous	50	0	50	0.0378	
Repo 1 NM_005529:ex5	:c.G6673Ap.G22255	chr1:22179244	missense	50	100	100	0.0405	
= variants that could c Red= affected individuals; Black Hover over zygosity numbers for de Biallelic variant severity score of 4 a 2 Base score (biallelic) for missense. + 1 Functional scores damaging: Pol + 1 Conservation scores damaging:	ontribute to biallelic severity = unaffected; (<i>Gray</i> = unknown <i>pth and quality information. Chi</i> isigned to variant NM_005529:e: yPhen MutationTaster. GERP phylo?	omosome position numbers hyp	oerlink to GRCh37 reso osomal location chr1:2	ources 2181895.				

MD's staff uses simplified SimulConsult interface

Diagnoses hidden so labs can include it on their public websites

× • Other • Regression × • • Early death if undiag Home ts-Click a gene to see varia

findings Add tests Phenotype

Patient: 2 year old boy

≤ 1m ▼ Other

PCH8: p

Incidental (or discovery) genes

Interpreter sets preferences for genes to report (ACMG or more), who to examine (proband, trio...), whether only to report childhood onset and for whom to report results

Keppert GBA monoallelic 3 P Carrier Report if LP Adult MIM PubMed HGNC Report IMEM216 monoallelic 4 P Carrier Report if LP Adult OMIM PubMed HGNC Image: Carrier Image: Carrier Report if LP Adult OMIM PubMed HGNC Image: Carrier Image: Carrier Report if LP Adult OMIM PubMed HGNC Image: Carrier Image: Carrier Report if LP Adult OMIM PubMed HGNC Image: Carrier Image: Carrier <th></th> <th></th> <th>Incidental gene</th> <th>es</th> <th></th> <th></th> <th></th> <th></th> <th></th>			Incidental gene	es					
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+1 De nove bost.								 variants that could contribute to monsulfic severity (incidental) Purple - selected individual Purple - selected individual Purple - selected individual Purple - selected individual Purple - selected - selec	

Confirm the output in the report "cart" and export Genome Report

Genome report: items selected
Reason for testing: CT or MRI: brainstem atrophy or hypoplasia (present now) Other findings: Regression (absent) Nystagmus, non-rotary (onset by about 1 month old) Hyperreflexia (onset by about 6 months old)
Diagnosis 1: VLDLR-related cerebellar hypoplasia VLDLR gene mutations (biallelic) Zygosity 100 NM_003383.3:c.1249_1255delTACAAGT Incidental in P:GBA gene mutation (monoallelic) Zygosity 50 NM_001005741:ex9:c.G1093A:p.E365K
Clear all selections Output report XML report

Export to SimulConsult's standard Genome-Phenome Report (shown on next page) or xml API to lab's reporting platform. Prognosis information about the disease in each diagnosis helps the provider and patient know what to expect and informs care.

Genome Reporting Workflow

Change initial information

Phenome & Genome

Diffe	erential diagnosis	Add findings	Add tests Pher	notype Genoty	TDe \		Advanced	mode
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	PCH8: pontocerebenar	3 🗸 🗸 🖌	(not selected) il gene	mutations (biallel	ic)			
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	LIS2: RELN-related liss	2 • 🗸 •	 EIF2AK3 ge 	ne mutations (biall	elic)		Incidental ger	ies
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	PCH9: pontocerebellar						Come panel	-
	PEHO-like syndrome						Assess findin	g
-	PCH1A: pontocerebella						Profile findin	g
-	PCH3: pontocerebellar						Database	
-	Microcephaly, postnata						Search Fi	le
-	Galactosialidosis syndro						Home	elp
Tip: <u>GFM</u>	ER images: Cerebellar hyp	oplasia, VLDLR-assoc	iated			More tips	Summary No	ote
Tip: <u>Gene</u>	eReviews: VLDLR-Associat	ed Cerebellar Hypopl	asia			OMIM	Order gene te	st
Gene vari	iants: Show the 1	/LDLR variant ascerta	ined reliably					
VLDLR b	iallelic gene severity scor	e 4				HGMD for VLDLR		
					•	Zvgosit	tv	
	Variant severity score and	sequence	chrPosition	effect	Р	м	F	_
🗹 Repo	4 NM_003383.3:c1249_12	SSdelTACAAGT	chr9:2643480	frameshift	100	50	50	
Red= affec Hover over	= variants that could contribut ted individuals; Black = unal zygosity numbers for depth and	e to biallelic severity ffected; Gray= unknown quality information. Chron	tosome position numbers h	vperlink to GRCh37 reso	urces			
Biallelic vari 4 Base scor +2 from Pat	iant severity score of 4 assigned e (biallelic) for frameshift. thogenicity database score.	to variant NM_003383.3x.1	249_1255delTACAAGT at ch	romosomal location chr	9:2643480.			



See report on next page

