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

	G	şul												
	4	-												
Genome report for a	ı 2 yea	ar old	boy											
Reason for testing:														
Deafness CT or MRI: brainstem atrop	ohy or hy	ypoplasi	a											
Other key clinical findings: Absence of: Regression														
Nystagmus, non-rotary														
Hyperreflexia Other prior test results:														
(none selected)														
Consanguinity of parents: 1	st cousi	in												
Ethnicity: (unspecified)														
Diagnosis # 1														-
Diagnosis # 1: VLDLR-relate Mode of inheritance: Autose	ed cereb	ellar hy	poplasia											
Gene symbol (HGNC): VLL	OLR													
Gene name: Very low densit Relevent variant:	y lipopro	otein rec	eptor											
Biallelic: Shared with both p	parents:	<u>NM_00</u>	3383.3:c.	1249_125	5delTA	CAAGT	Chrom	osomal	position	n: chr9::	2643480), Effect	t:	
rameshift														
Pertinent positive findings of	of the pa	atient fo	r this dia	gnosis:										
CT or MRI: brainstem atrop	ohy or hy	ypoplasi	a (present	now)										
Nystagmus, non-rotary (ons Hyperreflexia (onset by abo	ut 6 moi	nths old)											
Pertinent negative findings				omoele-										
(none entered)	or the p	adent I	л uns dia	agnosis:										
	eviews -	and OM	M											
Provider Resources: GeneR Patient and Family-Oriente	d Resou	urces: G	enetics H	ome Refe	man on or									
						d Disea	se-focu	sed patie	ent advo	cacy or	ganizati	ons		
					tence ai	id <u>Disea</u>	ise-focu	sed patie	ent advo	cacy or	ganizati	ons		
Prognosis														
Prognosis	At wh		do peop	le with V				ebellar	hypop	olasia l	nave th	iese fin]
	At wh	1			VLDLI		ed cer	ebellar 10	hypop 15	olasia l 25	ave th	ese fin	80	
Signs and Symptoms	Birth	1 month	3 months	le with \ 6 months	LDLI 1 year	R-relat 3 years	ed cero 6 years	bellar 10 years	hypor 15 years	25 years	40 years	60 years	80 years	
Signs and Symptoms Ataxia	Birth Few	1 month Few	3 months Some	le with V 6 months Some	1 year	R-relat 3 years Most	6 years	10 years Most	hypop 15 years Most	lasia l 25 years Most	40 years Most	60 years Most	80 years Most	
Signs and Symptoms Ataxia Intellectual disability	Birth Few NA	1 month Few Few	3 months Some Few	le with V 6 months Some Some	1 year Most Most	R-relat 3 years Most Most	ed cero 6 years Most Most	10 years Most Most	hypop 15 years Most Most	Dasia I 25 years Most Most	40 years Most Most	60 years Most Most	80 years Most Most	
Signs and Symptoms Ataxia	Birth Few NA NA	1 month Few	3 months Some	le with V 6 months Some	1 year	R-relat 3 years Most	6 years	10 years Most	hypop 15 years Most	lasia l 25 years Most	40 years Most	60 years Most	80 years Most	
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Signs and Symptoms Ataxia Intellectual disability Motor developmental delay	Birth Few NA NA NA Few	1 month Few Few NA Some	3 months Some Few Few NA Some	le with V 6 months Some Some NA Some	LDLI 1 year Most Some Some Some	R-relat 3 years Most Most Most Some	ed cere 6 years Most Most Most Some	bellar 10 years Most Most Most Some	hypop 15 years Most Most Most Some	Diasia I 25 years Most Most Most Some	40 years Most Most Most Some	ese fin 60 years Most Most Most Some	80 years Most Most Most Most Some	
Signs and Symptoms Ataxia Intellectual disability Motor developmental delay Gait disturbance Nystagmus, non-rotary	Birth Few NA NA NA	1 month Few Few NA	3 months Some Few Few NA	le with 6 months Some Some NA	LDLI 1 year Most Most Some Some	R-relat 3 years Most Most Most	6 years Most Most Most Most	10 years Most Most Most	hypop 15 years Most Most Most Most	Dasia I 25 years Most Most Most Most	40 years Most Most Most Most	60 years Most Most Most Most	80 years Most Most Most Most	
Signs and Symptoms Ataxia Intellectual disability Motor developmental delay Gait disturbance Nystagmus, non-rotary Eye movement deficit,	Birth Few NA NA NA Few	1 month Few Few NA Some	3 months Some Few Few NA Some	le with V 6 months Some Some NA Some	LDLI 1 year Most Some Some Some	R-relat 3 years Most Most Most Some	ed cere 6 years Most Most Most Some	bellar 10 years Most Most Most Some	hypop 15 years Most Most Most Some	Diasia I 25 years Most Most Most Some	40 years Most Most Most Some	ese fin 60 years Most Most Most Some	80 years Most Most Most Most Some	
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Diagnosis # 2: GJB2-rela Mode of inheritance: Au Gene symbol (HGNC): C Gene name: gap junction Relevent variant: Biallelic: Shared with bot	tosomal GJB2 protein, th paren	recessive , beta 2, 2 uts: <u>NM (</u>	6kDa; DF	NB1, DFN				chr13:2	0763492	, Effect.	: missen	se	
Pertinent positive finding - Deafness (onset at about	gs of the birth)	: patient	for this d	iagnosis:									
Pertinent negative findin		e nationt	for this c	lianneie									
- (none entered)	igs of th	e patient	ior uns c	nagnosis.									
Provider Resources: Ger	neReview	ws and O	MIM										
Patient and Family-Orie	ented Re	sources:	Genetics	Home Ref	erence a	nd Disea	ise-focu	sed patie	ent advo	cacy org	anizatio	ns	
Prognosis													
	At wh	at age d	lo people	with G	B2-re	ated de	afness	, AR, n	onsyn	lromic	have t	hese fi	ıd
	Birth	1 month	3 months	6 months	1 year	3 years	6 years	10	15	25	40	60	Г
Signs and Symptoms Deafness	Some	Some	Most	Most	Most	Most	Most	years Most	years Most	years Most	years Most	years Most	N
Vertigo, significant	Few	Some	Some	Some	Some		Some	Some	Some	Some	Some	Some	s
Findings detected by					1. Source	- one		1. Some	Johne	Joone			
	Tabora	nory tes											
GJB2 gene mutations (biallelic)	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	M
ABR abnormal	Some	Some	Most	Most	Most	Most	Most	Most	Most	Most	Most	Most	N
GJB2 gene deletion	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	F
(monoallelic) GJB2 gene mutation													Ļ
(monoallelic)	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	F
GJB2 gene deletions (biallelic)	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	Few	F
· · · ·													
KEY	None or	NA				ess than or	r equal to	Some is	more that	1 30%	Most is	more that	1 84
					30%								
													_
Incidental findings	ng Geisin	ger ACMC	3 superset (1	76), carrier g	enes, pha	rmacogen	etic gene	s. Genes o	examined	from P, N	1, F.		-
Incidental findings Incidental genes examined usi													
Incidental genes examined usi													
Incidental genes examined usi Incidental gene for P: Gi Gene name: Glucosidase Incidental gene type: Ca	, beta, ac												
Incidental genes examined usi Incidental gene for P: Gi Gene name: Glucosidase Incidental gene type: Ca Relevant variant:	, beta, ao rrier	cid; gluco	sylcerami	dase	34 n F	365K CH	romore	mal nos	ition: ch	r1-1550	06167	effect.	
Incidental genes examined usi Incidental gene for P: Gi Gene name: Glucosidase Incidental gene type: Ca	, beta, ao rrier	cid; gluco	sylcerami	dase	93A:p.E	<u>365K</u> Ch	romoso	mal pos	ition: ch	r1:1552	06167, 1	Effect:	
Incidental genes examined usi Incidental gene for P: Gj Gene name: Glucosidase Incidental gene type: Ca Relevant variant: Monoallelic: Apparently missense Provider Resources: OM	, beta, ac rrier de novo	eid; gluco : <u>NM_00</u>	sylcerami 1005741:e	dase x9:c.G109									
Incidental gene for P: G Gene name: Glucosidase Incidental gene type: Ca Relevant variant: Monoallelic: Apparently m missense	, beta, ac rrier de novo	eid; gluco : <u>NM_00</u>	sylcerami 1005741:e	dase x9:c.G109									
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Incidental genes examined usi Incidental gene for P: Gi Gene name: Glucosidase Incidental gene type: Ca Kelevant variantar: Monoalklie: Apparently missense Provider Resources: OM Patient and Family-Orie SimulConsult Ana	, beta, ac rrier de novo IIM ented Re alysis	cid; gluco : <u>NM 00</u> esources: and Qu	sylcerami 1005741:e Genetics uality F	dase ex9:c.G109 Home Ref	erence a								
Incidental gene examined usi Incidental gene for P: Gi Gene name: Glucosidase Incidental gene type: Ca Relevant variant: Monoallelic: Apparently, missense Provider Resources: OM Patient and Family-Orie	, beta, ac rrier de novo IIM ented Re alysis	cid; gluco : <u>NM 00</u> esources: and Qu	sylcerami 1005741:e Genetics uality F	dase ex9:c.G109 Home Ref	erence a								
Incidental genes examined usi Incidental gene for P: Gi Gene name: Glucosidase Incidental gene type: Ca Relevant variant: Monoallelis: Apparently . missense Provider Resources: OM Patient and Family-Orie SimulConsult Ana Quality metrics for anno . Rows read: 48	, beta, ac rrier <i>de novo</i> <u>mm</u> nted Re <u>alysis</u> otated va	cid; gluco : <u>NM 00</u> esources: and Qu ariant tal	sylcerami 1005741:e Genetics uality F ble	dase ex9:c.G109 Home Ref	erence a								
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sing patented 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

SimulCo													
Genome report for a	a 2 ye	ar old	boy										
Reason for testing: Deafness CT or MRI: brainstem atroj Other key clinical findings: Absence of: Regression Nystagmus, non-rotary Hyperreflexia Uther prior test results: (none selected) Consanguinity of parents: Ethnicity: (unspecified)			a										
Diagnosis # 1													
Sene niame: Very low densit Relevent variant: Stallelic: Shared with both p rameshift Pertinent positive findings CT or MRI: brainstem atroj Nystagmus, non-rotary (ons Hyperreflexia (onset by abc	of the parents: of the parents	atient fo	r this dia a (present onth old)	gnosis:	5delTA	CAAGT	Chron	osomal	position	n: chr9:	2643480), Effec	t:
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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



lev 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 Add tes	ts Phenotype	Genotype	Advanced	mode	Pertine	nce me	asure
Diseases	rom the patient's 47 gene varia	ints: Set	variant parameters]		for the	gene (g	rreen
VLDLR-related cerebel			Finding color key:				J v	,
Alcardi-Goutieres synd	ertinent gene variants		Pertinence			shading	g), toge	ther with
PCH2: pontocerebellar	4 • VLDLR gene muta	tions (biallelic)		1		c	,, 0	
Alström syndrome	3 ▼ ✓ ▼ ALMS1 gene muta	tions (biallelic)		Differential	-	details	about t	the relevant
CDG1A: PMM2-related	3c ▼ ✓ ▼ HSPG2 gene muta	tions (biallelic) 🥄		Gene discove		• •	1	1 1.
PCH8: CHMP1A-relate	3 ▼ ✓ ▼ DSPP gene mutatio	on (monoallelic)		Assess findin	ng	variants	s (oran	ge shading
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Schwartz-Jampel chond	2 V V EIF2AK3 gene mu	tations (biallelic)		Database		Delow)	and the	e factors
LIS1: Lissencephaly, iso				-	ile	1 1		1
Trisomy 21 (Down synd					alm	underly	ng eao	ch gene
			More tips		ote			- 11 f
Tin: The "Order gone test" butt	on at right has hyperlinks to a var	ioty of recourses	OMIM	Order gene te	art	severity	score,	allow for
The Order gene test but	on at right has hyperinks to a var	lety of resources		Order gene te	st		+	Lation
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- nepon	47:c.G5899A;p.V1967J	<u>chr1:22181895</u>	missense	50	50	0	0.0405	
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	50:c.C7806A:b.V2602V	chr1:22174518	synonymous	50	100	50	0.0383	
	50:c.G6402Ap.V2134V	chr1:22180723	synonymous	50	50	50	0.0416	
	51x.G6552Ap.T2184T	chr1:22179451	synonymous	50	0	50	0.0378	
	52:c.G6673Ap.G22255	chr1:22179244	missense	50	100	100	0.0405	
Red= affected individuals; Black								
Hover over zygosity numbers for a	lepth and quality information. Chromoso	me position numbers hyp	erlink to GRCh37 reso	urces				
	assigned to variant NM_005529:ex47:c.G5	899A:p.V1967I at chrome	osomal location chr1:22	2181895.				
2 Base score (biallelic) for missens +1 Functional scores damaging: Pe								
+1 Conservation scores damaging:								

Genome Reporting Workflow

Dis	seases	Patient: 2	year	r old be	oy	Cha	ange initial information		
•	VLDLR-related cerebel PCH2: pontocerebellar	Pertinent	posit	tive fin	din	18	Finding color key: Pertinence		
•	Aicardi-Goutières syndi			Reason		CT or MRI: brainstem atrophy or hyp	onlasia		
-	PCH8: pontocerebellar		im •	Other	-	Hyperreflexia	opusu		
-	Alström syndrome		lm •		-	Nystagmus, non-rotary			
	PCH10: Pontocerebella		m 🔻	Other	-	Microcephaly		Different	tial Dx
	LIS2: RELN-related liss		-		-	History of a similar disorder in family	or contacts	Incidenta	l gene
	CDG1A: PMM2-related		_		_			Genome	report
-	Schwartz-Jampel chond							Gene dise	
	PCH1B: pontocerebella	Pertinent	nega	tive fir	ıdir	gs			
-	Muscular dystrophy-dy							Gene p	anel
-	PCH9: pontocerebellar	×	-	Other	•	Regression		Assess fi	inding
-	PEHO-like syndrome	×	•	1	-	Early death if undiagnosed		Profile f	inding
-	PCH1A: pontocerebella							Datab	
-	PCH3: pontocerebellar								
-	Microcephaly, postnata							Search	File
-	Galactosialidosis syndro							Home	Help
			_		_			1	
							More tips	Summary	Note
: Simu	ulConsult resource: Detail:	s on judging	reare	ession			OMIM	Order ge	ne test

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

Incidental genes	
Report GBA monoallelic 3 P Carrier Report if LP Adult OMIM PubMed HGNC	GBA monoallelic gene severity score 3 Haw monoallelic gene severity score 3 Zygosity
	Variant severity score and sequence chrPosition effect P M F freq1
Report TMEM216 monoallelic 4 P Carrier Report if LP Adult OMIM PubMed HGNC	✓ Repo. 3 NM_001005741=x9::G1093Ap.E365K chr1:155206167 missense 50 0 0 0.0136
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Report TMEM216 monoallelic 4 F Carrier Report if LP Adult OMIM PubMed HGNC	Repo. 3 NM_001171811xx7xc632Ap_E278K chr1:155206167 missense 50 0 0 0.0136 Repo. 3 NM_001171812xx7xc632Ap_E378K chr1:155206167 missense 50 0 0 0.0136
	- variants that could contribute to monalistic severity (incidental) Furplin = testocie individual Woodwork over garger humbers for depth and quality information. Chromosome position numbers hyperfluit to GRC317 resources Monalistic variant servity score of 1 assigned to variant MW 001005711:nb2cfC0374.pcBsf21657 at chromosomal location chr.1355206167. 1 Bas score (monalistic for misses) reformed from the usual score by one due to frequency > 0.011. + 1 Denote bott.

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

Genome report: items se	lected
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 repor

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.

Phenome & Genome

Diffe	rential diagnosis A	Add findings	Add tests	Phen	otype Geno	type		🗹 Advan	iced mode
Dis	eases	From 47 gene va	ariants:			Set	variant parameters		
Dx1 -	VLDLR-related cerebel PCH2: pontocerebellar	Pertinent gene v	ariants				Finding color key: Pertinence		
	Alcardi-Goutières syndi PCIB: pontocerebellar Alström syndrome PCIII0: Pontocerebellan LIS2: RFL-N-related IIS COGIA: PMM2-related Schwartz-Jampel chond PCIII3: pontocerebellan PCII0: pontocerebellan PCII3: pontocerebellan PCII3: pontocerebellan	4 x √ x 3 x √ x 3 x √ x 3 x √ x 3 x √ x	(not selected) Diagnosis 1 Diagnosis 2 CT EIF	31 gene 2 gene 1 SA gene m 32AK3 gen	mutations (biall mutations (biall nutations (bialle utations (biallel e mutations (bia tation (monoall	elic) elic) ic) illelic)		Differentia Incidental g Genome reg Gene discor Gene pan Assess find Profile find Databas	genes port very nel ding ding
	Microcephaly, postnata Galactosialidosis syndro								File Helr
	R images: Cerebellar hyp Reviews: VLDLR-Associat ants: Show the 1 V		olasia	·		+	More tip	Summary Order gene	Note test
VLDLR bi	iallelic gene severity score	<u>e 4</u>					HEMD for VLDL8	ty	
Repo.	Variant severity score and 4 NM 003383.3::1249 125			osition	effect frameshift	P 100	M 50	F 50	
Hover over 2 Biallelic varia Base score	= variants that could contribut ted individuals; Black = unaf sygosity numbers for depth and ant severity score of 4 assigned t (biallelic) for frameshift. hogenicity database score.	fected; Gray= unknown quality information. Chr		,					

Incidental (or discovery) genes



See report on next page

