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Master HL7 genetic variant reporting panel (2016-11-21)

Date Done	Time Done	Where Done	Comment
03/21/2017	Type a value	Select or type a value	Type a value

Name	Value	Units
Form configuration		
Choose kind of mutations targeted	× Discrete variants × Complex variants × Pharmacogenomics Select one or more	
Choose region of interest specification	× Specific targeted mutations × Range targeted in the reference sequence Select one or more	
Default transcript reference sequence coding system	Select one or type a value	
Default genomic reference sequence coding system	Select one or type a value	
Variables that apply to the overall study		
Reason for study	Type a value	
Genetic disease(s) assessed	× Abnormal arm span Search for or type values	
Medications assessed	× Arginine × aripiprazole Search for or type values	
Gene(s) assessed	× ACAP3 × ACTR3BP1 × ACTRT3 Search for values	
Gene mutations tested	× NM_138413.3(HOGA1):c.289C>T (p.Arg97Cys) Search for or type values	
1.1 Ranges of DNA sequences examined	Type a value	
Add another 'Ranges of DNA sequences examined'		
Description of ranges of DNA sequences examined	Type a value	
Summary results		
Discrete variation analysis overall interpretation	Positive	
Deletion-duplication overall interpretation	No deletion or duplication detected in studied region	
Full narrative report	Type a value	
Variant ISCN	Type a value	
Versions of Coding Systems		
Human reference sequence assembly	GRCh37	
HGVS version [ID]	Type a value	
dbSNP version [Num]	Type a number	
COSMIC version [Num]	Type a number	
ClinVar version [ID]	Type a number	
1 Discrete genetic variant panel		
Variant category	Simple Variant	
Genetic variant coding system	ClinVar Variants	
Discrete genetic variant	NM_138413.3(HOGA1):c.289C>T (p.Arg97Cys)	
Transcript specification		
Gene studied	HOGA1	
Transcript RefSeq ID	NM_138413.3	
DNA change c.HGVS	c.289C>T	
Amino acid change p.HGVS	p.Arg97Cys	

DNA change type	SNV
Amino acid change type	Select one or type a value
Genomic specification	
Genomic reference sequence	NC_000010.10
Genomic DNA change (gHGVS)	Type a value
Genomic ref allele	C
Genomic allele start-end	99358609^99358609
Genomic alt allele	T
Other optional codes related to a discrete genetic variant	
Haplotype Name	Type a value
dbSNP ID	rs267606762
CIGAR	Type a value
Other possible attributes	
Cytogenetic location of variant	10q24.2
Genomic source class	Select one
Variant analysis method type	Select one or type a value
Interpretations	
Clinical significance	Select one
Genomic variant assessment	Select one
Probable associated phenotype	Primary hyperoxaluria, type III
Allelic state/phase information	
Allelic state	Select one
Allelic Frequency NFR	Type a number
Allelic read depth	Type a number
Allelic phase [Type]	Select one or type a value
Basis for allelic phase	Select one

Add another 'Discrete genetic variant panel'

1 Complex genetic variant - panel

Complex variant ID	9818
Complex variant HGVS name	NM_000044.3:c.172_174CAG(10_36)
Complex variant type	Select one
Associated phenotype	Bulbo-spinal atrophy X-linked
Clinical significance	Select one
Allelic state	Select one
Basis for allelic phase	Select one

1.1 Discrete genetic variant panel -

Variant category	Simple Variant
Genetic variant coding system	ClinVar Variants
Discrete genetic variant	NM_014630.2(ZNF592):c.3136G>A (p.Gly1046Arg)
Transcript specification	
Gene studied	ZNF592
Transcript RefSeq ID	NM_014630.2
DNA change c.HGVS	c.3136G>A
Amino acid change p.HGVS	p.Gly1046Arg
DNA change type	SNV

Amino acid change type	Select one or type a value
Genomic specification	
Genomic reference sequence	NC_000015.9
Genomic DNA change (gHGVS)	Type a value
Genomic ref allele	G
Genomic allele start-end	85342440^85342440
Genomic alt allele	A
Other optional codes related to a discrete genetic variant	
Haplotype Name	Type a value
dbSNP ID	rs150829393
CIGAR	Type a value
Other possible attributes	
Cytogenetic location of variant	15q25
Genomic source class	Select one
Variant analysis method type	Select one or type a value
Interpretations	
Clinical significance	Select one
Genomic variant assessment	Select one
Probable associated phenotype	Spinocerebellar ataxia autosomal recessive 5
Allelic state/phase information	
Allelic state	Select one
Allelic Frequency NFR	Type a number
Allelic read depth	Type a number
Allelic phase [Type]	Select one or type a value
Basis for allelic phase	Select one

1.2 Discrete genetic variant panel -

Variant category	Simple Variant
Genetic variant coding system	ClinVar Variants
Discrete genetic variant	NM_001201543.1(FAM161A):c.1309A>T (p.Arg437Ter)
Transcript specification	
Gene studied	FAM161A
Transcript RefSeq ID	NM_001201543.1
DNA change c.HGVS	c.1309A>T
Amino acid change p.HGVS	p.Arg437Ter
DNA change type	SNV
Amino acid change type	Select one or type a value
Genomic specification	
Genomic reference sequence	NC_000002.11
Genomic DNA change (gHGVS)	Type a value
Genomic ref allele	T
Genomic allele start-end	62066830^62066830
Genomic alt allele	A
Other optional codes related to a discrete genetic variant	
Haplotype Name	Type a value
dbSNP ID	rs200691042
CIGAR	

	Type a value
Other possible attributes	
Cytogenetic location of variant	2p15
Genomic source class	Select one
Variant analysis method type	Select one or type a value
Interpretations	
Clinical significance	Select one
Genomic variant assessment	Select one
Probable associated phenotype	Retinitis pigmentosa 28
Allelic state/phase information	
Allelic state	Select one
Allelic Frequency NFR	Type a number
Allelic read depth	Type a number
Allelic phase [Type]	Select one or type a value
Basis for allelic phase	Select one

Add another 'Discrete genetic variant panel'

Add another 'Complex genetic variant - panel'

1 Pharmacogenomics gene results panel -

Gene(s) studied	× ACAP3 Search for or type values
Genotype display name	Type a value
Cross reference to full genetic details	Type a value
Genetic variation's effect on drug metabolism	Ultrarapid metabolizer
Genetic variation's effect on drug efficacy	Select one or type a value
Genetic variation's risk for hypersensitivity	Select one or type a value

1.1 Medication usage implications panel -

Medication assessed	Arginine
Medication usage suggestion [type]	Increase Dose
Medication usage suggestion [narrative]	Type a value

1.2 Medication usage implications panel -

Medication assessed	aripiprazole
Medication usage suggestion [type]	Decrease Dose
Medication usage suggestion [narrative]	Type a value

Add another 'Medication usage implications panel'

2 Pharmacogenomics gene results panel -

Gene(s) studied	× ACTR1A Search for or type values
Genotype display name	Type a value
Cross reference to full genetic details	Type a value
Genetic variation's effect on drug metabolism	Ultrarapid metabolizer
Genetic variation's effect on drug efficacy	Select one or type a value
Genetic variation's risk for hypersensitivity	Select one or type a value

2.1 Medication usage implications panel

Medication assessed	armodafinil
Medication usage suggestion [type]	Use Caution

Medication usage suggestion [narrative]	**Type a value**

Add another 'Medication usage implications panel'

Add another 'Pharmacogenomics gene results panel'