

☐ Display Question Code☐ Show Help/Description☐ Keyboard Navigation On Input Fields

Total # of Questions: 149

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 ⚙	<input checked="" type="checkbox"/> Discrete variants <input checked="" type="checkbox"/> Complex variants <input checked="" type="checkbox"/> Pharmacogenomics Select one or more	
Choose region of interest specification ? ⚙	<input checked="" type="checkbox"/> Specific targeted mutations <input checked="" type="checkbox"/> 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 ? ⚙	<input checked="" type="checkbox"/> Abnormal arm span Search for or type values	
Medications assessed ⚙	<input checked="" type="checkbox"/> Arginine <input checked="" type="checkbox"/> aripiprazole Search for or type values	
Gene(s) assessed ⚙	<input checked="" type="checkbox"/> ACAP3 <input checked="" type="checkbox"/> ACTR3BP1 <input checked="" type="checkbox"/> ACTRT3 Search for values	
Gene mutations tested ⚙	<input checked="" type="checkbox"/> 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'