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Genes and transcripts are clearly key concepts in the interpretation of biological phenomena. This survey is intended to solicit information on how a 'primary' transcript should be chosen for a particular gene. Choosing a 'primary' transcript could be done from several perspectives: coding sequence content, expression levels, clinical variant reporting, historical usage, etc. Given the broad use of the transcripts, we would like your feedback for the impact on your work and to discover what different communities want in these transcript sets.

The two global sources of transcript annotation (RefSeq and Ensembl/GENCODE) will take your responses into account when formulating future strategies and resources.

This survey has four sections. It should take approximately 8 minutes to complete. The examples we use in the survey are all based on scenarios we frequently encounter during our curation.

Section 1 - Transcript choice (6 questions)

Section 2 - Variant interpretation and reporting (5 questions)

Section 3 - Reference sequence sources (2 questions)

Section 4 - About you

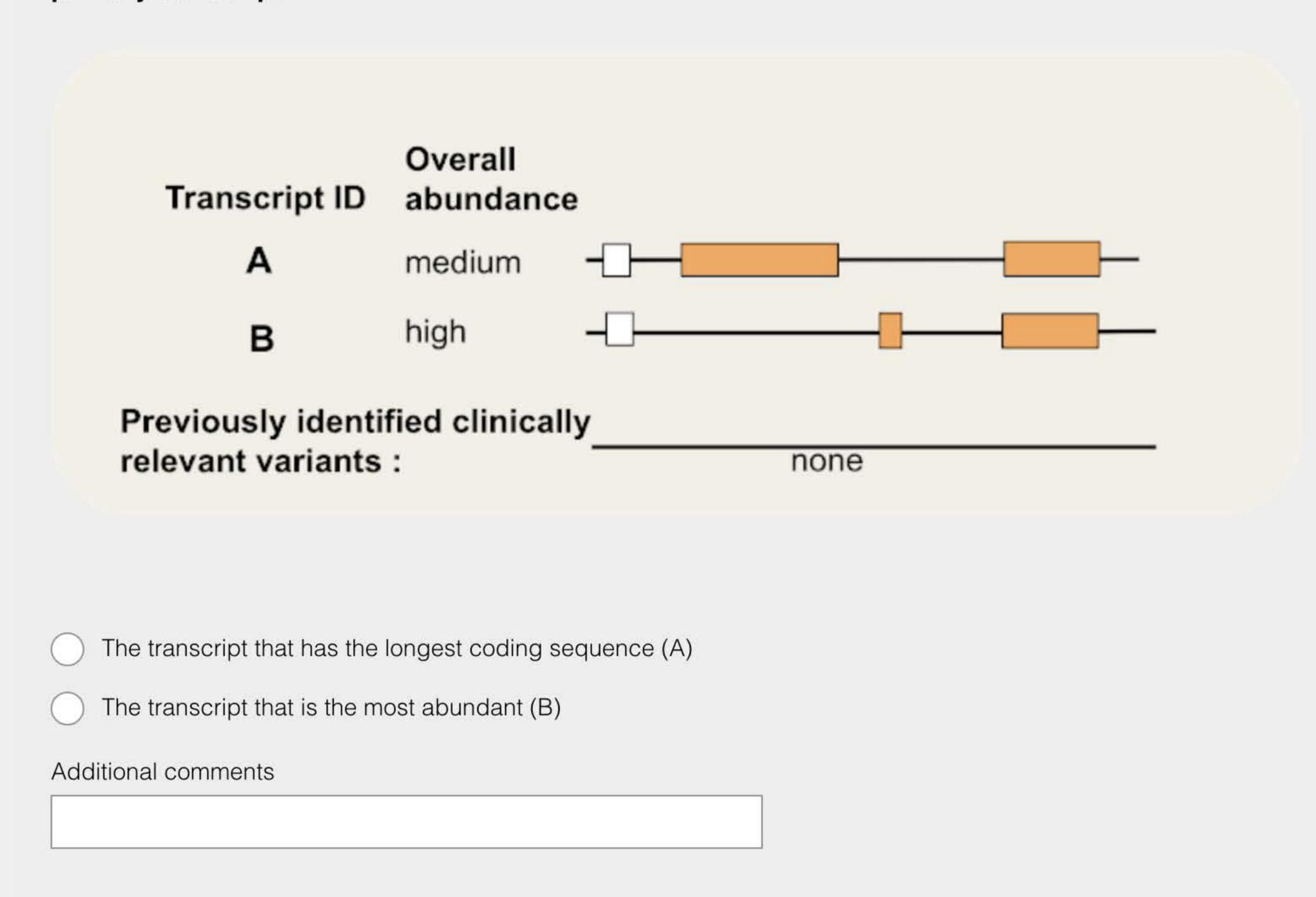
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Section 1 - Transcript choice

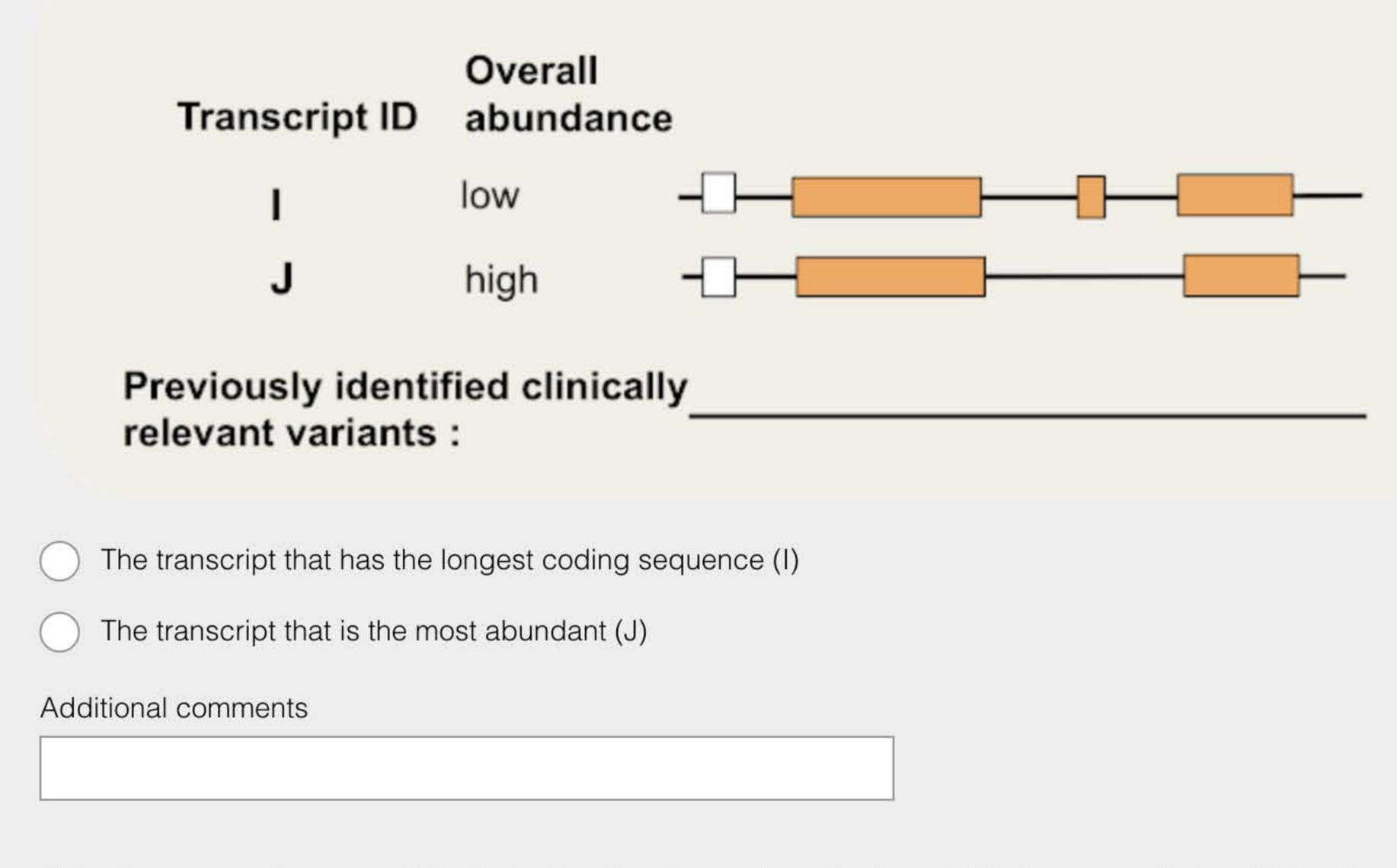
1.	. Considering the transcripts of a gene, t	for your work how important is it to have:
• •	. Considering the transcripts of a gene,	or your work now important is it to have.

	Critical	Nice to have	Not needed
Only ONE primary transcript			
A minimal set of transcripts to cover ALL EXONS with evidence of CLINICAL SIGNIFICANCE			
A minimal set of transcripts to cover ALL ABUNDANT PROTEIN-CODING EXONS			
A minimal set of transcripts to cover ALL ABUNDANT EXONS			
A larger set of ALL known transcripts			
All of the above			
ther (please specify)			

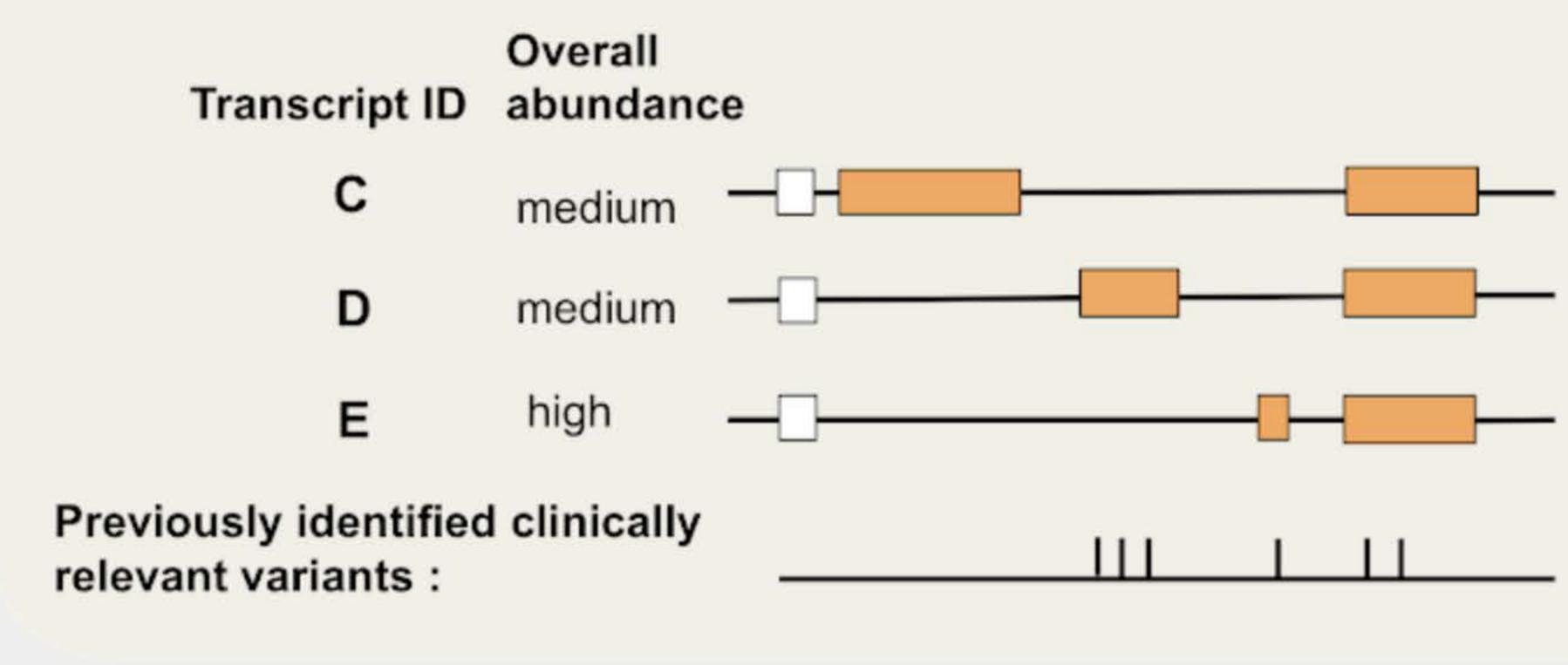
2. In the case of a gene WITHOUT any known clinically relevant variants, which transcript do you think should be the primary transcript?



3. In the case of a gene WITHOUT any known clinically relevant variants, which transcript do you think should be the primary transcript?



4. In the case of a gene WITH clinically relevant variants, which transcript should be the single primary transcript? Please answer based on which is best for your work.



D	medium -	
E	high -	
Previously identified relevant variants :	ed clinically	
Transcript that has the longe	est coding sequenc	e (C)
Transcript that covers the m	ost clinically relevar	nt variants (D)
Transcript that is the most al	oundant (E)	
Transcript that has been mo	st used historically	
Additional comments		
5. Considering the sequence	e of a transcrip	t, which is the most important to you?
That the sequence matches	the reference asse	embly sequence (e.g. GRCh37/ hg19), even if it contains minor alleles
That the sequence does not	contain any pathog	genic alleles
That the sequence matches	the global major al	llele
That the sequence does not	change	

6. For	your work,	when is i	t appropriate t	o make an	update to	the primary	transcript?	Please check a	III that apply.

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A change in coding sequence	
A change in UTR length	
A change to transcript splicing	
Never update	
Other (please specify)	

It doesn't matter to me

Section 2 - Variant interpretation and reporting

7. I	f there is one prima	ary transcript pe	r locus, v	what would y	ou do?		
0	Use it, and only it, for	INTERPRETING the	conseque	ence of variants?)		
0	I wouldn't use just on	e transcript for INTE	RPRETATIO	ON unless it was	the only one know	'n	
0	Other (please specify	/)					
O 1	f tha maat aawara s	variant affact to b		ad ia nat an t	ha aalaatad muin	navy transarint (E) v	what wanta wan dag
0. 1	i the most severe v	variant enect to t	e report	ea is not on t	ne selected prii	nary transcript (r), v	what would you do?
	Transcript ID					Consequence	
	F	Selected -				synonymous	
	G	Affected				missense	
	Н	Other				synonymous	
	Variant to	be reported:		1			
	Report the variant on	the selected primar	v transcrip	t (F) only			
	Report the variant on						
0	Report the variant on	both the selected p	rimary tran	script (F) and th	e affected transcri	ot (G)	
\bigcirc	Report the variant on	all transcripts (F, G,	H)				
\bigcirc	I don't know						
0	Other (please specify	/)					
9. \			use for i	reporting vari	ants? Please ch	neck all that apply.	
	RefSeq transcripts or						
	Ensembl/GENCODE		1S				
	GRCh38/hg38 genon						
	GRCh38/hg38 genon						
	LRG transcripts or LF	ra proteiris					
				Prev	Next		

Section 3 - Reference sequence sources 10. Please check all that are true: I use RefSeq transcripts for my work I do not use RefSeq I use Ensembl/GENCODE transcripts for my work I do not use Ensembl/GENCODE Both RefSeq and Ensembl/GENCODE transcripts are useful for my work I do not know whether RefSeq or Ensembl/GENCODE produce the best transcripts for my work 11. What is most important to you? Having RefSeq and Ensembl/GENCODE agree on one primary transcript per gene Having different sets as each set has different strengths I have no preference Other (please specify) Prev Next

Section 4 - About you
12. Which professional categories best describe you? Please check all that apply.
Healthcare professional
Diagnostician
Bioinformatics professional
Life Science researcher
Developer/engineer
Educator
Student
Other (please specify)
13. Where do you work? Check all that apply.
University/College/Academia/Non-profit/Research
Clinical diagnostics
Clinical research
Commercial/Industry
Government
Other (please specify)
14. In what country do you work?
15. <u>Question 1 revisited - a primary transcript</u>
Do you want us to provide one primary transcript?
Yes
O No
Um not sure
Please note, you may contact us at the <u>help-desk</u> to provide additional feedback about the resources
in this survey or any other NCBI resource.
Prev Done