



Position statement: Use of LRG Reference Sequences for Variant Reporting

In recognition of the discontinuation of the LRG reference sequence project, EMQN CIC:

- Recommends laboratories continue to follow local guidelines on the use of transcript identifiers for reporting genomic variation.
- Encourages community engagement with the MANE initiative, a joint project between NCBI and EMBL-EBI, which aims to create a universal standard for transcript annotation in the genomics era.
- Supports the use of MANE Select and MANE Plus Clinical as denoted by the MANE initiative, for the standardization of variant annotation, interpretation and reporting.
- Recognizes that the MANE initiative is still in development and will not penalize
 participating laboratories for using the correct LRG reference in the 2023 scheme rounds.

For more information on MANE, please see our webinar <u>here</u>. Information on the initiative can be found at the <u>NCBI website</u> or <u>EBI website</u>.

Please contact us at office@emqn.org with any questions.

For issues on specific genes/transcripts, requests to review existing MANE transcripts or assign new ones, please contact: MANE-help@ncbi.nlm.nih.gov or mane-help@ebi.ac.uk