GHIF Practical guidelines for quality control of WGS results in population-scale initiatives

Resources

**Guidelines that mention sample QC**

1. Gargis, A. S. *et al.* Assuring the quality of next-generation sequencing in clinical laboratory practice. *Nat Biotechnol* **30**, 1033–1036 (2012).
2. Rehm, H. L. *et al.* ACMG clinical laboratory standards for next-generation sequencing. *Genet Med* **15**, 733–747 (2013).
3. Aziz, N. *et al.* College of American Pathologists’ Laboratory Standards for Next-Generation Sequencing Clinical Tests. *Arch Pathol Lab Med* **139**, 481–493 (2015).
4. Roy, S. et al. Standards and Guidelines for Validating Next-Generation Sequencing Bioinformatics Pipelines. J Mol Diagnostics 20, 4–27 (2018).
5. U.S. Food and Drug Administration (FDA). Considerations for Design, Development, and Analytical Validation of Next Generation Sequencing-Based In Vitro Diagnostics Intended to Aim in the Diagnosis of Suspected Germline Diseases. Updated 13 April 2018.
6. Medical Genome Initiative *et al.* Best practices for the analytical validation of clinical whole-genome sequencing intended for the diagnosis of germline disease. *Npj Genom Medicine* **5**, 47 (2020).
7. TC215/SC1 - ISO standard on “Quality Control Metrics for DNA Sequencing”

**QC frameworks**

1. Whalley, J. P. *et al.* Framework for quality assessment of whole genome cancer sequences. *Nat Commun* **11**, 5040 (2020).
2. Karczewski, K. J. *et al.* The mutational constraint spectrum quantified from variation in 141,456 humans. *Nature* **581**, 434–443 (2020).