



Quartet DNA Reference Materials

Human DNA for Quality Control and Performance Assessment of Whole-Genome Sequencing (Chinese Quartet Family of Monozygotic Twin Daughters, Father, and Mother: D5, D6, F7, and M8)

The Quartet DNA Reference Materials suite (RM) was prepared as part of "*The Quartet Project: Quality Control and Data Integration of Multi-omics Profiling*" in which matched reference materials of DNA, RNA, proteins, and metabolites were simultaneously manufactured from the same batch of cultured cells. A unit of the Quartet DNA Reference Materials consists of four vials containing human genomic DNA isolated from the immortalized B-lymphoblastoid cell line of a specific family member of a Chinese Quartet family from Fudan Taizhou Cohort, including father (F7), mother (M8), and two monozygotic twin daughters (D5 and D6). Each vial contains approximately 10 µg of genomic DNA with the peak fragment size longer than 60 Kb, and the DNA is in TE buffer (10 mM TRIS, 1 mM EDTA, pH 8.0).

SPECIFICATIONS

Name of DNA Reference Material	Color	Concentration	Volume	DIN	A260/280	A260/230	Stock
FDU_Quartet_DNA_D5_20171028	●	~220 ng/µL	50 µL	≥8.5	1.80~1.92	>2.00	TE buffer
FDU_Quartet_DNA_D6_20171028	●						
FDU_Quartet_DNA_F7_20171028	●						
FDU_Quartet_DNA_M8_20171028	●						

Quartet DNA is stored at -80 °C and stable (DIN ≥8.5) for >3 years and was long-term monitored by Agilent 4200. The Quartet DNA Reference Materials suite is shipped with dry ice. Store at -80 °C for long-time storage, or at 4 °C for short-time storage.

INTENDED USES

The Quartet DNA Reference Materials suite is intended for quality control and performance assessment of human genome sequencing. It can measure and mitigate technical variation, enabling more accurate data integration in large cohort studies. The Quartet performance metrics for human genomic variant calling results are: 1) Reference datasets-based precision and recall; and 2) Quartet family based Mendelian concordance rate. This genomic DNA suite should be profiled in the same way as study samples in each batch within a lab. Because the RM is extracted DNA, it is not suitable for assessing the pre-analytical steps such as DNA extraction, but it aims to evaluate the whole process of library preparation, sequencing, and the bioinformatics pipelines such as mapping, post-alignment, and variant calling. This RM is not intended to assess subsequent bioinformatics steps such as functional or clinical interpretation. It is for research purpose only.

REFERENCE DATASETS

The Quartet DNA reference datasets are provided as a variant call file (vcf) that contains the high-confidence SNVs, small indels (less than 50 bp), and structural variants (insertions and deletion over than 50 bp), as well as a tab-delimited “bed” file that describes the high-confidence bed regions, using methods described in the Quartet DNA manuscript. The v1.0 of DNA reference datasets covers approximately 87.8% of the GRCh38 assembly (<https://gdc.cancer.gov/about-data/gdc-data-processing/gdc-reference-files>). As sequencing technologies and analysis methods improve, the reference datasets will be updated periodically. All the reference datasets and the quality assessment tools can be accessed through the Quartet Data Portal (<http://chinese-quartet.org/>).

NOTICE AND WARRANTIES TO USERS

The Quartet DNA Reference Materials are being provided on an "AS IS" basis. The provider hereby warrants that the Quartet DNA Reference Materials have been obtained or created a) in full compliance with all applicable local, governmental and international laws, regulations and guidelines, b) after obtaining and in full compliance with all necessary approvals from the relevant research ethics committees, and c) after obtaining and in full compliance with all necessary, properly signed informed consents and acknowledgement forms from any human subjects, or their legal guardians. The provider makes no representation or warranty, whether expressed or implied, with respect to the Quartet DNA Reference Materials, including any representation or warranty as to the durability, storage, disposal, merchantability or fitness for a particular purpose or to the non-infringement of the Quartet DNA Reference Materials on the proprietary rights of a third party. The recipient shall use the Quartet DNA Reference Materials obtained or created as described above at its sole risk and liability.

The Quartet DNA reference materials and raw datasets are publicly available and accessible. Researchers are encouraged to access and analyze the datasets. The recipients of the Reference Materials are highly encouraged to share their data with Fudan University through the Quartet Data Portal in order for us to improve the reference datasets and to better serve the community.

For other questions, please feel free to contact the Quartet Project team (quartet@fudan.edu.cn and <http://chinese-quartet.org/>), and/or Drs. Yuanting Zheng (zhengyuanting@fudan.edu.cn) and Leming Shi (lemingshi@fudan.edu.cn).



“中华家系”1号人源基因组 DNA 参考物质

全基因组测序质量评价用人源基因组 DNA

(中国同卵双胞胎家庭，两个女儿、父亲和母亲：D5, D6, F7 和 M8)

“中华家系”1号 DNA 参考物质来源于“**Quartet 中华家系1号项目：多组学质量控制和数据整合**”，该项目研制了同批次细胞来源的 DNA、RNA、蛋白质、代谢物的多组学参考物质。“中华家系”1号 DNA 参考物质一套内含 4 管人源永生 B 淋巴母细胞系的全基因组 DNA，细胞系建立自复旦泰州队列的中国同卵双胞胎家庭，包括父亲(F7)、母亲(M8)和两个同卵双胞胎女儿 (D5 和 D6)。每管 DNA 参考物质包含 10 μg 基因组 DNA，DNA 主峰片段>60 Kb，溶解于 TE 溶液(10 mM TRIS, 1 mM EDTA, pH 8.0)。

规格参数

DNA 参考物质的名称	颜色	浓度	体积	DIN	A260/280	A260/230	储存条件
FDU_Quartet_DNA_D5_20171028	●	~220 ng/ μL	50 μL	≥ 8.5	1.80~1.92	>2.00	TE buffer
FDU_Quartet_DNA_D6_20171028	●						
FDU_Quartet_DNA_F7_20171028	●						
FDU_Quartet_DNA_M8_20171028	●						

“中华家系”1号 DNA 参考物质在-80 $^{\circ}\text{C}$ 条件下长期稳定性>3 年(DIN ≥ 8.5)，且采用 Agilent 4200 的 DIN 值(DNA Integrity Number)进行长期稳定性监测。DNA 参考物质采用干冰运输，收到后于-80 $^{\circ}\text{C}$ 长期保持，4 $^{\circ}\text{C}$ 短期保存。

预期用途

“中华家系”1号 DNA 参考物质的预期用途是对人基因组测序进行质量控制和性能评价，也可用于评估和消除多批次检测中的技术噪音，为多中心、长期大队列研究的数据整合提供质量保证。采用“中华家系”1号 DNA 参考物质进行基因组变异检测性能评价的指标包括：1)基于参考数据集的精密度和召回率；2)基于同卵双胞胎家系关系的孟德尔一致率。基因组 DNA 参考物质需要和研究样本平行检测，才能保证对每个实验室、每批次数据的质量进行有效的评价。本 DNA 参考物质适用于评价基因组变异检测的全过程，包括建库、测序、以及 mapping、post-alignment、variant calling 等生物信息学分析过程。不适用于评价 DNA 抽提等样本前处理过程，以及功能注释、临床解读等生物信息学分析过程。本 DNA 参考物质仅适用于科学研究。

参考数据集

“中华家系”1号 DNA 参考数据集包括变异检测结果文件(vcf)，涵盖高置信单碱基变异、小插入缺失变异(小于 50 bp)和结构变异(大于 50 bp 的插入和缺失)，以及描述高置信基因组区域的“bed”文件。v1.0 版本参考数据集覆盖 87.8% 基因组区域(GRCh38: <https://gdc.cancer.gov/about-data/gdc-data-processing/gdc-reference-files>)。随着高通量基因组测序技术和分析算法的进步，将定期发布参考数据集的升级版。“中华家系”1号 DNA 参考数据集、原始数据以及质量评价工具可以通过 Quartet Data Portal 获取使用(<http://chinese-quartet.org/>)。

用户使用条款

“中华家系”1号 DNA 参考物质按照“原样”提供。研究者保证本参考物质的研制：a) 完全符地区、政府和国际相关法律、法规和指导原则；b) 获得伦理委员会审批并遵守相关条款；c) 志愿者或其法定监护人已知情并签署知情同意书。研究者对“中华家系”1号 DNA 参考物质不作任何明示或暗示的陈述或保证，包括其耐用性、储存、处置、特定用途的适用性或适销性，或不侵犯“中华家系”1号 DNA 参考物质的第三方的所有权。物质接收者使用本参考物质涉及上述条款的，应自行承担相关风险和责任。

“中华家系”1号 DNA 参考物质和参考数据集提供开放获取和非商业使用。鼓励任何团队基于“中华家系”1号开放数据进行数据质量相关研究，研究不能涉及志愿者隐私和疾病风险预测。获取参考物质者需要通过 Quartet Data Portal 与复旦大学 Quartet 项目团队共享数据，以促进参考数据集的更新，更好地为领域服务。

如有其它问题，请联系“中华家系”1号项目团队(quartet@fudan.edu.cn 或 <http://chinese-quartet.org/>)，或者项目负责人郑媛婷博士(zhengyuanting@fudan.edu.cn) 和石乐明博士(lemingshi@fudan.edu.cn)。