Evidence-Based CFTR Variant Coverage

Optimized Advanta™CFTR NGS Library Prep Assay design supports target enrichment of all 27 CFTR exons and select intronic regions, which cover >250 variants derived from the CFTR2 mutation list*. Sequence the spectrum of variant types, from simple SNPs to large deletions, in a single NGS-based workflow.

Table 1. CFTR variants targeted by the Advanta CFTR NGS Library Prep Assay. Ordered by variant legacy name (alphanumerically). (a) indicates variants (23) within the ACMG/ACOG recommendations. (d) indicates the minimum variants (31) required by German guidelines. Bold indicates the inclusion of at least one variant-bearing sample in the analytical validation.

*The Clinical and Functional Translation of CFTR (CFTR2); available at cftr2.org (CF-causing variants from CFTR2_8August2016.xlsx)
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**Variant cDNA name**

| Variant cDNA name | Table 2. CFTR variants targeted by the Advanta CFTR NGS Library Prep Assay. Ordered by variant cDNA name (5' to 3'). (a) indicates variants (23) within the ACMG/ACOG recommendations. (d) indicates the minimum variants (31) required by German guidelines. Bold indicates the inclusion of at least one variant-bearing sample in the analytical validation.


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Panel availability subject to confirmation.

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