Service Profile

Digital PCR Services

Mutation Detection & Copy Number Variation

Helping you simplify going digital with an all-in-one solution

Digital PCR (dPCR) is revolutionizing the way rare target detection and copy number variation analyses are performed. dPCR now provides unparalleled sensitivity, precision and reproducibility for detecting low-abundance targets, allelic variants and small fold-change differences. However, onboarding dPCR in your lab can seem daunting given the investments in training, standardization and resources required.

QIAGEN® Genomic Services overcomes these hurdles by providing a convenient, all-in-one dPCR mutation and copy number variation detection service. Using our proven sample preparation technologies and powerful QIAcuity® Digital PCR System, we offer the following benefits:

- **End-to-end service**: we take care of every step, from sample preparation to data analysis
- **Guidance and flexibility**: we aid in designing your project and help you make the right decisions
- **Expertise in nanoplate-based dPCR technology**: we provide years of experience in nanoplate-based dPCR using our sensitive QIAcuity Digital PCR System
- **Ready-to-publish data**: we deliver comprehensive reports and data packages, and provide guidance on the next steps

Partner with us for expert guidance and dedicated service – from Sample to Insight® – to detect the single positive today.

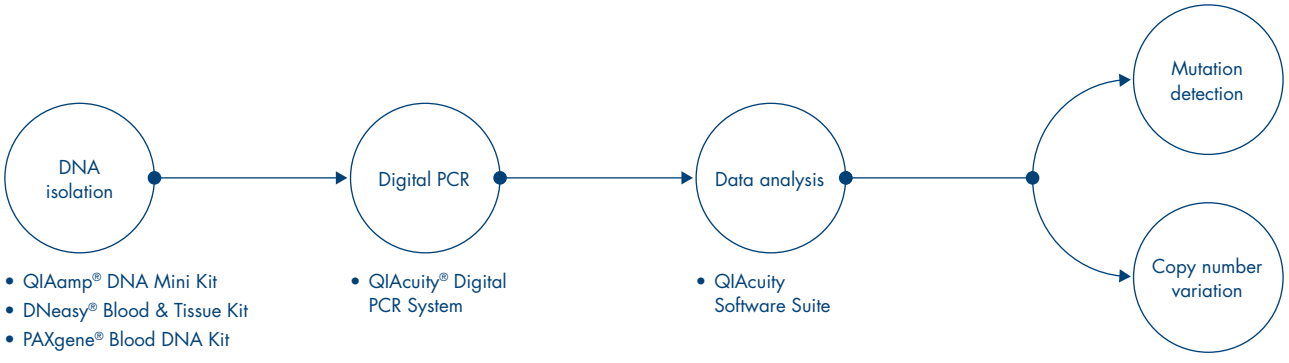

Figure 1. Digital PCR Mutation Detection and Copy Number Variation workflow.

Transforming the PCR experience with digital PCR

dPCR is a highly precise approach to sensitive and reproducible nucleic acid detection and quantification. Measurements are performed by dividing the sample into partitions, with zero or one target molecule present in any individual reaction. Each partition is analyzed after end-point PCR cycling for the presence or absence of a fluorescent signal, and the absolute number of molecules present in the sample is calculated. Compared to other techniques, dPCR provides:

Figure 2. The top 5 benefits of dPCR.

Comprehensive data analysis

QIAGEN Genomic Services performs your data analysis using the QIAcuity Software Suite. Your final data package will contain absolute quantifications of your samples, with the concentration in copies per microliter of your target sequence. Further, a complete overview of the mutation detection results presented as a mutation fraction table or the copy number variation results are delivered with detailed explanations.

Table 1. Mutation fraction table

	Sample/ NTC/control	Type	Reaction mix	Target	IC	Concentration (copies/µl)	CI (95%)	Mutation fraction	CI (95%)
A1	Sample 1	Mutation	BRAF V600M	MT	–	1979.3	2.60%	100%	4%
A1	Sample 1	Wildtype	BRAF V600M	WT	–	0	–	–	–
A2	Sample 2	Mutation	BRAF V600M	MT	–	927.1	3.9%	47.23%	11%
A2	Sample 2	Wildtype	BRAF V600M	MT	–	1035.7	3.7%	–	–
B5	NTC	Mutation	BRAF V600M	WT	–	0	–	n.a.	n.a.
B5	NTC	Wildtype	BRAF V600M	MT	–	0	–	–	–

Service specifications

Consultation	Free consultation with an expert to design an experimental setup that best meets your needs.		
Sample requirements	**Sample input**	**Isolation kit**	**Input requirements**
	Customer-isolated human DNA	N/A	500 ng (>10 ng/µl)
	Cell-free circulating DNA (ccfDNA) from human plasma or serum	QIAamp ccfDNA/RNA Kit QIAamp MinElute® ccfDNA Kit	Minimum: 1 ml Maximum: 4 ml
	Cells (human)	QIAamp DNA Mini Kit	Minimum: 1 x 10^6 cells pelleted and frozen
	Fresh-frozen tissue (human)	DNeasy Blood & Tissue Kit	Minimum: 4–5 mg Maximum: 50 mg
	FFPE samples (human)	QIAamp DNA FFPE Tissue Kit GeneRead® DNA FFPE Kit	Minimum: 2 x 10 µm sections of 250 mm^2 Maximum: 4 x 10 µm sections of 250 mm^2
	Blood (PAXgene®, human)	PAXgene Blood DNA Kit	Recommended: 1 tube
	Other	**Please inquire**	
Sample quality control	OD measurement for determination of sample concentration. Optional gel electrophoresis for determination of DNA integrity (e.g., DIN value from capillary gel electrophoresis). This is a STOP/GO point where it is possible to omit samples or replace samples before proceeding.		
Digital PCR	Mutation detection is performed using the QIAcuity Probe PCR Kit and the dPCR LNA Mutation Assay. • QIAcuity Nanoplate 26k is used for highly sensitive and accurate detection of mutations with a high reaction volume		
	CNV analysis is performed using the QIAcuity EG PCR Kit and the dPCR Copy Number Assay. • QIAcuity Nanoplate 26 k is used for sensitive detection of rare CNV events with a high reaction volume • QIAcuity Nanoplate 8.5 k is used for high-throughput routine CNV screening with a low reaction volume		
	For CNV analysis, 2 reference samples and 2 reference assays need to be provided for normalization.		
Complete data analysis	Results can only be provided per plate		
	Raw data	Number of partitions valid/positive /negative	
	Raw data quality control	Image quality controls and image corrective measures included in QIAcuity Software Suite	
	Normalization approach	For CNV normalization to reference assays/samples	
	Absolute quantification	Absolute quantification data (concentration in copies/µl and confidence interval at a 95% confidence level)	
	Mutation detection	Mutation detection statistics (Mutation fraction in percent and confidence interval at a 95% confidence level) For simplex tests, individual replicate results are not available.	
	Copy number variation analysis	Copy number variation statistics (number of copies per genome and confidence interval at a 95% confidence level) For simplex tests, individual replicate results are not available.	
Final report and consultation	The final data analysis package contains an overview of data analysis and algorithms used, the files and tables listed above and publication-ready figures (PDFs provided as standard, please inquire for SVG or other formats). A teleconference is scheduled with QIAGEN scientists to discuss analysis and validation of results. Consultation and support will be provided for 90 days following delivery of data (for data delivery only projects), or delivery of data analysis (for data analysis inclusive projects). For extended support beyond 90 days, please inquire.		

Note: Service specifications might be tailored to the needs of the project on a case-by-case basis.

How can we accelerate your research?

Our expert team is looking forward to learning about your research project and designing your customized service with QIAGEN.

Tell us about your project

The QIAGEN Genomics Service is intended exclusively for research use only (RUO). This service is not intended for the diagnosis, prevention or treatment of a disease.

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