

## Molecular Testing of Blood Spot Cards for Newborn Screening EQA

- 12 bloodspots in total
- 4 distributions
- Varying genotypes included
- 4 weeks to submit results
- All EQA samples validated
- Detailed instructions provided
- Results assessed by expert panel
- Marking criteria based on professional guidelines

EQA	Type of EQA	Sample Type	ISO17043 Accreditation	Testing	Techniques
Molecular testing for cystic fibrosis (CF)	Genotyping	Blood spot cards	✓	CFTR gene	Any molecular technique
Molecular testing for MCADD	Genotyping	Blood spot cards	✓	ACADM c.985A>G p.(Lys329Glu) pathogenic variant	Any molecular technique

## Other Molecular Genetics EQAs for 2019

Arrhythmia & Cardiomyopathy Ataxia and Hereditary Spastic Paraplegia Breast and Ovarian Cancer Dementia Cystic Fibrosis Charcot Marie Tooth Disease Duchenne & Becker Muscular Dystrophies Fabry Disease Fragile X Syndrome Huntington Disease Hypotonic Infant (PWS, SMA and DM1) Imprinting & UPD	Lynch Syndrome MCADD Maternal cell contamination (MCC) & sexing Mitochondrial Disorders Neurofibromatosis & schwannomatosis Pathogenicity of Sequence Variants Pheochromocytoma Polyposis Retinal Disorders Skeletal Dysplasia Variant Validation X-inactivation
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Genotyping and interpretation EQAs with a single distribution per year.  
For further information on these EQAs contact us at [info@genqa.org](mailto:info@genqa.org)

Please see overleaf for more EQA information  
Email: [info@genqa.org](mailto:info@genqa.org)



## GenQA EQA Specialties

Molecular Genetic Disorders

Molecular Rapid Aneuploidy (MRA)

Molecular Pathology

Sample Handling: DNA extraction & quantification

Newborn Screening

Non-Invasive Prenatal Testing (NIPT)

Haematological Neoplasms

Technical: Next Generation Sequencing

Constitutional Postnatal Testing

Constitutional Prenatal Testing

Clinical Genetics

Individual Competency Assessment (G-TACT / Tissue-i)

For further information, please contact us at [info@genqa.org](mailto:info@genqa.org)  
Registration for all GenQA EQAs for 2020 will open in September 2019

## The EQA Cycle

