

## Molecular Testing for Newborn Screening

- 8 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	Neonatal screening cards	✓	CFTR gene	Any molecular technique
Molecular testing for MCADD	Genotyping	Neonatal screening cards	✓	ACADM gene including c.985A>G p.(Lys329Glu) pathogenic variant	Any molecular technique

### New for 2020

EQA	Type of EQA	Sample Type	ISO17043 Accreditation	Testing	Techniques
Molecular testing for Severe Combined Immunodeficiency (SCID) PILOT	Analytical	Neonatal screening cards	X	T-cell Receptor Excision Circles (TRECs)	Any molecular technique

SCID Pilot EQA – single distribution of 3 bloodspot cards  
No performance criteria will be applied

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)



# 2020 EQA



Collaboration between CEQAS and UK NEQAS Molecular Genetics  
Members of UK NEQAS consortium

## GenQA EQA Specialties

Molecular Genetic Disorders

Variant Classification and Interpretation

Molecular Pathology

Sample Handling: DNA extraction & quantification

Newborn Screening

Preimplantation Genetic Testing

Haematological Neoplasms

Technical: Next Generation Sequencing

Constitutional Postnatal Testing

Prenatal (including Non-Invasive) Testing

Clinical Genetics

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

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## The EQA Cycle

