

## FOCUS ON GenQA EQAs for Molecular Newborn Screening

EQA for three autosomal recessive genetic disorders:

- **Cystic Fibrosis (CF)**
- **Medium-chain acyl-CoA dehydrogenase deficiency (MCADD)**
- **Severe Combined Immunodeficiency Disease (SCID)**



SCID is a rare condition of the immune system that leads to severe abnormalities of the immune system development and function. The incidence of SCID is ~1 in 100 000 live births.

TRECs are small circles of DNA created in the T-cell lymphocytes, and indicate the presence of mature T-cells; TRECs are reduced in SCID patients. Early detection of SCID improves the patient's likelihood of a successful bone marrow transplant.

In 2020 GenQA launched a SCID pilot EQA, providing **8 samples** over **two distributions**.

- Blood is spotted on to **neonatal screening cards**, to mimic a heel-prick sample.
- Participation is open to all interested laboratories.
- Participants are required to test for T-cell receptor excisions circles (**TREC**) levels, **determine the clinical interpretation** and submit their findings on a proforma.
- An EQA Summary report is published after every EQA run, detailing the EQA data, participant issues and summarising the methods used by all participants.
- This pilot EQA is supported by Public Health England.

### Molecular Newborn screening webinar Monday 30<sup>th</sup> November 2020, 11am GMT

- Overview of GenQA molecular newborn screening EQAs
- The challenges of testing for SCID
- SCID pilot EQA for 2020 and 2021
- LIVE question and answer session

Register for the free webinar and live Q&A session, at

<https://attendee.gotowebinar.com/register/930572076806458125> by **25<sup>th</sup> November 2020**.

If you wish to submit a question for the Q&A session, then please email [info@genqa.org](mailto:info@genqa.org)

Registered participants will have priority access to the webinar for one month (if you are unable to join us on the 30<sup>th</sup> November).

For further information on these EQAs please contact us at [info@genqa.org](mailto:info@genqa.org)



Public Health  
England

**FOCUS ON next month:**

**Individual Competency Assessment  
for BRCA variant classification**