

North West Genomic Medicine Service Alliance Workshop:

Genomic Investigation of Deafness

Friends' Meeting House, Manchester

09:30-16:00, Friday 28th April 2023

Speakers include:

Martin McLean (National Deaf Children's Society), Professor Stavros Stivaros (University of Manchester), Dr Emma Clement (North Thames GMSA), Rachel Belk (North East and Yorkshire GMSA), Sarah Stewart, Dr Emma Burkitt Wright (North West GMSA)

This workshop is intended primarily for healthcare professionals who refer patients for genomic testing for deafness in the NHS in England

Free to attend (60 places available), lunch and refreshments provided: register by emailing both sarah.stewart@mft.nhs.uk and emma.burkitt-wright@mft.nhs.uk (please advise us of any specific communication or dietary requirements by 10th April 2023)

09:30 Registration and refreshments, Programme runs 10:00-16:00

Content will include:

Refreshers:

Genomic Testing for Deafness – how is it done?

To whom should we offer testing?

Consent and ethical considerations

Understanding molecular genetic reports

Current perspectives:

Imaging findings that can inform genetic diagnosis

Family and stakeholder perspectives

Outcomes and developments:

England-wide outcomes of R67 since April 2021

Family and professional resources to support understanding and consent

Implications for clinical practice:

Informative scenarios and syndromic presentations within R67

Management of syndromic health implications

Working towards greater understanding of genotype-phenotype correlation

In both morning and afternoon sessions, there will be breakout groups, for which you are welcome to bring details of cases that you have found interesting, challenging or otherwise informative

Aims: To equip attendees to identify patients who might benefit from testing, counsel appropriately, and understand and communicate the results of testing.

To further develop networks of interested professionals working with individuals and families and signpost clear routes for advice and further information.