genedrive

MT-RNR1

Genedrive® MT-RNR1 ID Kit: The world's first point-of-care genetic test to reduce aminoglycoside induced hearing loss (AIHL)

The Problem

1 in 500[1] people are born with a genetic variant that results in profound hearing loss when exposed to gentamicin (an aminoglycoside antibiotic), the first line treatment for suspected sepsis in newborn babies.[2]

Treatment must be within the golden hour[2]

1 in 500 born with **MT-RNR1 1.555A>G gene variant**

Administer aminoglycoside antibiotics

Patient suffers severe hearing loss

The Impact

Pre-speech hearing loss has a lifelong impact on the baby and their family, including social and emotional development, language, education and career.

Dual cochlear implant surgery costs **£60,282** in the first year and **£808.77** per year recurrently. Nationally, 200 babies per year require this surgery because of genetic AIHL. This is a cost burden on the NHS of **£13 million anually.**[3]

The environmental impact of additional hospital visits is substantial. Each patient will contribute **1,444kg** of CO_2 in the first year and **228kg** per year recurrently.[4]

The Solution: Genedrive® MT-RNR1 ID Kit using the Genedrive® System

Key Features

- ✓ Rapid, point-of-care genetic test that delivers results in **26 minutes**
- ✓ Simple test procedure using non-invasive buccal swab samples
- ✓ Easy to interpret results displayed on touch screen
- ✓ Connectivity to various middleware platforms to automatically record results

CPIC Recommend MT-RNR1 variant carriers avoid aminoglycosides[5]

NICE Recommended[6]

Expected Benefits

- ✓ Reduced risk of AIHL improves patient safety
- ✓ Safe antibiotic administration within the golden hour
- ✓ Reduced demand for cochlear implant surgeries
- ✓ Reduced environmental impact of recurrent hospital visits

Research Case Study

Pharmacogenetics to Avoid Loss Of Hearing (PALOH)[7]

PALOH

Can rapid, point-of-care genetic testing be implemented in an acute neonatal setting to avoid antibiotic induced hearing loss, without disrupting normal standards of care?

751 babies recruited

3 tested positive for MT-RNR1 m.1555A>G

NO siginificant change in time to antibiotic administration

"Until now there has not been a test quick enough to ensure that newborn babies with a bacterial infection and the m.1555A>G variant gene are treated with an appropriate antibiotic. Having this test available to NHS staff can avoid the risk of hearing loss in babies with the variant who need treatment with antibiotics.

Mark Chapman, Interim Director of Medical Technology at NICE

"Over 600 babies have been tested. Three babies tested positive for the gene change that would cause them to go deaf if given gentamicin and were successfully given an alternative antibiotic within the NICE recommended golden hour."

Dr Ajit Mahaveer, Consultant Neonatologist at Saint Mary's Hospital

"Genomic medicine is transforming healthcare, and this is a powerful example of how genetic testing can now be done extremely quickly and become a vital part of triage – not only in intensive care but across our services.

Professor Dame Sue Hill, Chief Scientific Officer for England and Senior Responsible Officer for Genomics in the NHS

Limitations

There are other MT-RNR1 variants associated with a risk of hearing loss

Proposed Future Work

Implementation data to be collected in smaller, non-specialist centres

Further evidence generation required in centres with different patient demographics e.g. diverse ethnicities

References

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4 Romanello 2021. Calculating the carbon footprint of the NHS in England Carbon Brief. https://www.carbonbrief.org/guest-post-calculating-the-carbon-footprint-of-the-nhs-in-england/

5. Clinical Pharmacogenetics Implementaton Consortium (CPIC). 2022. Guideline for the use of Aminoglycosides based on MT-RNR1 genotype.

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