Healthcare Improvement Scotland | SHTG Advice on health technologies

Plain Language Summary

Genetic testing to guide antibiotic use and prevent hearing loss in newborn babies | October 2024

Key messages

- Newborn babies with a mutation in a gene called MT-RNR1 have an increased risk of hearing loss when given the antibiotic gentamicin. Approximately 1 in 500 babies have this mutation.
- Genetic testing of 424 babies found three babies who had the MT-RNR1 gene mutation. These babies were given a different antibiotic.
- Babies with an infection need antibiotics within 1 hour. Genetic testing did not delay starting antibiotic treatment for babies with suspected infections.
- Hearing loss has a serious effect on the quality of life of affected children and their families. It affects a child's ability to communicate, their social and emotional development, and their opportunities in life. Children with hearing loss need to wear hearing aids or cochlear implants.
- Over a 3-year period, genetic testing of newborn babies needing antibiotics is expected to prevent hearing loss in three babies at an estimated cost to the NHS of £29,000.

What is aminoglycoside induced hearing loss?

Aminoglycosides are a group of antibiotics. The most common aminoglycoside used to treat infections in newborn babies is gentamicin. Aminoglycoside antibiotics have side effects that can damage your ears causing permanent deafness. This is called aminoglycoside induced hearing loss.

Newborn babies who have a specific mutation of the MT-RNR1 gene are at increased risk of aminoglycoside induced hearing loss. Alternative antibiotics may have fewer side effects but have a higher risk of antibiotic resistance.

What is MT-RNR1 genetic testing?

Genetic testing can determine whether a baby has the MT-RNR1 gene mutation. A swab is taken from the baby's mouth. The swab sample is mixed with chemicals and inserted into a small machine. The machine tests the genetic material on the swab and tells doctors whether the baby has the mutation. The test can be performed in the hospital ward and takes about 30 minutes. If the baby has the mutation, they can be treated with a different type of antibiotic.

Why is this important?

Aminoglycoside antibiotics have side effects that can permanently damage your ears. Babies who have a specific mutation of the MT-RNR1 gene are at increased risk of aminoglycoside induced hearing loss. Newborn babies who show signs of having a serious infection need antibiotics within 1 hour of deciding to treat them. Only genetic testing on the hospital ward can provide results fast enough to inform the choice of antibiotics in this situation.

What we did

We looked at whether genetic testing in newborn babies was effective and safe for identifying babies that could become deaf if given antibiotics such as gentamicin. We calculated the effects on the NHS budget of introducing this type of genetic testing for babies in Scotland.

What we found

One study used genetic testing to guide the choice of antibiotics for babies in two neonatal intensive care units (NICUs) in England. Of 424 babies tested at admission to the NICU, three had the gene mutation that increased their risk of deafness. These three babies were given a different, equally effective antibiotic.

Five babies had an incorrect positive result: the test said they had the mutation, but they did not. This means they were given a different antibiotic unnecessarily.

Genetic testing did not cause delays in treating the babies' infections. Approximately two tests in 100 failed. This test failure rate might be higher in less specialist hospital wards.

Hearing loss has a serious effect on the quality of life of affected children and their families. This can include children's ability to communicate, their social and emotional development, and their education and employment opportunities.

Parents of newborn babies with suspected infections wanted information about:

- how common the gene mutation linked with aminoglycoside induced hearing loss is
- genetic testing

- the risks associated with hearing loss and infection
- the effectiveness of alternative antibiotics.

If genetic testing of babies who need antibiotics was made available in all NICUs in Scotland, it would cost the NHS an estimated £29,000 over 3 years. Testing over this 3-year period would likely prevent three babies from hearing loss. The exact costs of genetic testing were difficult to estimate.

Expanding the number of wards that offer genetic testing could prevent more babies from going deaf and generate savings for the NHS. This expansion of genetic testing would not be based on the evidence that is currently available.

What is our conclusion?

Babies who have a mutation in the MT-RNR1 gene are at increased risk of becoming deaf if they are given gentamicin to treat an infection. Genetic testing can identify babies with this gene mutation, allowing doctors to prescribe a different antibiotic. The genetic test does not delay treatment for babies with infections.

Alternative antibiotics have a higher risk of antibiotic resistance. This is likely a small risk because the gene mutation is rare, so few babies will need an alternative antibiotic. The benefits of preventing hearing loss in babies likely outweigh the risks of increased antibiotic resistance.

Introducing MT-RNR1 genetic testing for newborn babies in NICUs will likely cost the NHS £29,000 over a period of 3 years and result in three fewer babies becoming deaf.

What next?

Genetic testing will be considered for national rollout to hospital wards that care for newborn babies.

This plain language summary has been produced based on an SHTG Assessment