NIHR | Oxford Biomedical Research Centre

Patient and Public Involvement

Patient involvement in research into genetic testing

Case Study

Background

Oxford's Genomic Medicine Theme works closely with Headlines, the UK-based charity supporting those with craniosynostosis and other rare craniofacial conditions. This case study shows how the Headlines patient group helped identify research priorities and subsequent research findings.

Around 1 in 2,000 babies are affected by craniosynostosis, a fusion between bones of the skull. The condition causes the skull to grow in a distorted shape, which can lead to serious complications involving vision, hearing, eating and brain function. The NHS Highly Specialised Service in Oxford provides the multidisciplinary assessment and surgery required to correct the alterations in skull shape, treating families from across England.

What we did

The patient representative on the NIHR Cleft and Craniofacial Clinical Studies group and the Headlines patient group identified the Top Ten Questions in Craniosynostosis.

One of the patients' questions was: "what are the causes of non-syndromic single suture synostosis?". This question relates to Oxford's long-standing programme of research, which discovered that a faulty gene or chromosome is responsible for craniosynostosis in around a quarter of children.

What difference did it make

Following identification of this research priority by the Headlines patient group, **further research** in Oxford discovered mutations in the gene SMAD6 affecting 26 children, 16 of whom had non-syndromic single suture synostosis. This research highlighted the importance of genetics in this type of craniosynostosis.

Articles in the Headlines magazine on SMAD6 and on diagnoses made in the **100,000 Genomes project** promoted knowledge of SMAD6 and its implications for patients and families.

From 2021, NHS testing for SMAD6 will be available to all children with non-syndromic single suture midline synostosis, helping to provide an answer for some families to one of Headlines' Top Ten questions.

Headlines changed their constitution in 2020 to include promotion of research as one of the primary goals of the organisation.