Genetics and hearing loss – research and practice

For many years, although research into genetic forms of hearing loss had identified numerous genes responsible, particularly in childhood- and early adult-onset deafness, very few of these advances had any impact on services for families. Scientists had identified over one hundred genes which could cause non-syndromic deafness, and hundreds more causing deafness in association with other medical features (syndromic deafness), but most families still remained without a genetic cause for their deafness.

More recently there has been a step-change in genetic diagnosis as a result of technological advances in DNA analysis. In the 100,000 Genomes Programme in England, Whole Genome Sequencing has been applied to diagnose many rare conditions including deafness. By the end of the programme, WGS will be a standard diagnostic tool in the National Health Service. We will explore what this means for families currently and in the future and will discuss whether new treatments really will be developed as a result of the advances in genetics.