I am a clinical and molecular geneticist and my research interest and clinical expertise is in the field of childhood genetic deafness. My group investigates causes of inherited forms of deafness in order to understand the molecular mechanisms involved. We have identified several genes causing syndromic and non-syndromic deafness in children and have used these discoveries to improve molecular diagnostic services offered to families. Our Regional Laboratory is a national expert centre for genetic deafness. I am an invited assessor for the European Molecular Genetics Quality Network, an external quality assessment organization; the role of the group is to set cases and score molecular genetic reports from laboratories participating in the deafness scheme.

I am currently involved in delivery of the 100,000 Genomes Programme; this project, funded by the UK National Health Service, aims to transform healthcare by delivering Whole Genome Sequencing as part of routine clinical service. I am Lead for Rare Diseases for the North Thames Genomic Medicine Centre (GMC), a consortium of 7 hospitals and specialist centres in London. I hold a weekly genetic deafness clinic at Great Ormond Street Hospital for Children and the Royal National throat Nose and Ear Hospital and a monthly dual sensory impairment clinic at RNTNEH.