HIPS/19/17 - Should Scotland provide whole genomic sequencing for diagnosis of rare disorders: A health economic analysis

Around 6-8% of the Scottish population have a rare disease, 80% of which have a genetic origin. Most rare diseases are severe, life-threatening and have a profound impact upon the individual and their family. The journey to diagnosis, often referred to as a ‘diagnostic odyssey’, can take many years, involves numerous hospital visits, costly and invasive tests, multiple misdiagnoses, and is often unsuccessful leaving shattered hopes and expectations. Recent developments in genomic medicine bring promise of improved diagnostic rates, potentially enabling earlier and more accurate diagnosis. Such information is useful for future family planning and even treatments. Whilst the new genomic tests are more expensive than standard care, fewer hospital visits and tests and opportunities for targeted treatments, could offset NHS costs of current testing, and be highly valued by patients and their families. NHS Scotland must decide soon which patients should receive which genomic tests. As science pushes forward, it is important not to lose sight of users at the centre of care. We propose to compare the costs and benefits of genomic testing for rare disorders, from the perspectives of service-users and the NHS, with NHS current practice.