Joint working executive summary

Project title	A joint working project with Academic Health Science Network North East and North Cumbria (AHSN NENC, now known as Health Innovation) & Sanofi to develop a proactive case finding programme in Primary care to identify patients with Rare Diseases
Partner organisation/s	AHSN NENC (now known as Health Innovation) Sanofi
Project rationale	• Children and adults with these rare, lysosomal storage disorders are often misdiagnosed or remain undiagnosed. This means they can go many years without a correct diagnosis or appropriate disease management, negatively impacting their health • This issue of underdiagnosis has been recognised within The UK Strategy for Rare Diseases (published by the Department of Health in February 2020) and most recently the UK Rare Disease Framework (published by the Department of Health in January 2021) • Conventional disease awareness initiatives have been used to raise the awareness of treatable lysosomal storage disorders, however, the rarity of the conditions means that the average primary care physician is unlikely to ever encounter a patient with Pompe, Fabry or Gaucher disease. Diagnosis relies on an individual to suspect, make the connection and either diagnose themselves or refer the patient for testing The project will address several issues related to rare diseases, all in line with the UK Rare Disease Framework. To do this we will deploy a diagnostic tool to sit within GP clinical systems to identify patients with a high clinical suspicion of Pompe, Fabry and Gaucher diseases, with the aims of: 1. Facilitating earlier diagnosis of these rare diseases with a standardised process 2. Earlier access to metabolic specialists and treatments 3. Prevention of irreversible complications of disease 4. Streamlining the referral pathway to metabolic centres from general practice

Project period	Q2 2022 – Q4 2024
Project objectives	• The aim of this project is to facilitate earlier diagnosis of Fabry, Pompe and Gaucher diseases through the use of disease prediction models using real-world data from primary care records. • Sanofi will partner with AHSN NENC to programme the disease prediction models into a Clinical Digital Resource Collaborative (CDRC) Precision tool which will sit within GP clinical systems in the NENC AHSN. The screening tool will be able to be used on GP practice medical records software (SystmOne and EMIS). • A list of patients with a high probability of Pompe, Fabry and Gaucher diseases will be produced for the doctor in charge to review and decide whether the patient should be referred for disease testing. The vision is for this to sit alongside conventional methods of diagnosis. **Benefits to patients**: • Earlier diagnosis of these life-limiting diseases will mean earlier clinical intervention and correct therapeutic support **Benefits to NHS** • This will reduce the need for reliance on an individual healthcare professional having the knowledge of a rare condition to make the diagnosis, one of the main reasons for many patients being mis-diagnosed or diagnosed late • Aside from improved outcomes, earlier diagnosis may also produce cost-efficiency by reducing the pattern of interventions and referrals often seen in these patients pre-diagnosis **Benefits to Sanofi:** • Sanofi produce therapies which may be used to treat patients in line with local/national guidelines, should any positive diagnosis be confirmed This project has pooled both direct and indirect financial resources.
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