From research to action: advances in rare disease

1865
Gregor Mendel starts describing principles of inheritance.

1903
Archibold Garrod's book, *Inborn Errors of Metabolism*, is published, igniting interest in the study of genetic diseases.

1946
First genetic counselling clinic in Europe started at Great Ormond Street Hospital.

1959
An additional copy of chromosome 21 is determined to be the cause of Down Syndrome.

1969
Department of Health introduces newborn screening using the Guthrie method.

1983
The genetic marker for Huntington disease is found, making it the first genetic disease to be mapped.

1985
The first NHS DNA diagnostic service is launched.

1989
The gene involved in cystic fibrosis, *CFTR*, is identified.

1990
The ground-breaking Human Genome Project is launched.

1992
Emergence of amniocentesis method to test for certain genetic conditions.

1997
Orphanet, a profoundly important resource for rare disease still widely used by clinicians today, is established.

1997
NHS begins using DNA sequencing for familial hypercholesterolaemia.

2001
Rhys Evans becomes first child in the UK treated with gene therapy for severe combined immunodeficiency.

2003
The first entire human genome sequence is published by The Human Genome Project.

2008
Rare Disease Day established by EURORDIS.

2013
UK government publishes its first strategy for rare disease and launches the 100,000 Genomes Project.

2014
The Early Access to Medicines Scheme is launched to enable earlier access to potentially life-changing treatments.

2015
The National Congenital Anomaly and Rare Disease Registration Service is established.

2018
The NHS Genomic Medicine Service is established in England.

2019
Rapid exome sequencing is made available nationally for acutely unwell babies.

2020
Crispr-Cas9 gene therapy is used inside the body for the first time, for inherited blindness disorder Leber's congenital amaurosis 10.

2020
Genome UK published.

2021
Five-month old baby with spinal muscular atrophy is first NHS patient to be treated with life-changing gene therapy.

2021
Non-invasive prenatal testing added to NHS pregnancy screening programme.

2021
The UK Rare Disease Framework published.

2022
The UK Rare Disease Action Plan launched.

The 100,000 Genomes Project