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News Release

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Scotland study to probe causes of rare diseases signs up first recruits

People in Scotland with rare genetic diseases are set to benefit from a DNA study that seeks to improve their diagnoses and treatments.

The study – launched by the Scottish Genomes Partnership in collaboration with Genomics England – will analyse the entire genetic make-up of 330 people with rare diseases and members of their family.

Using advanced computing to link genetic data with patients' health information, scientists hope to pinpoint differences in their DNA code that may be responsible for their condition.

The research aims to offer more precise diagnoses for patients and explore how whole genome sequencing technology could be used in clinical practice to improve treatment and management of the diseases.

Participants will be identified by NHS Scotland Genetics Clinics and invited to take part in the research along with members of their close family.

A total of 1000 people will have their genome sequenced for the research, which will contribute to the 100,000 Genomes Project, a flagship project to advance clinical care through genome research.

Around 3.5 million people in the UK are living with a rare disease caused by a faulty gene, such as muscular dystrophies, rare forms of intellectual disability and rare inherited neurological problems. Although each disease affects fewer than one in 2,000 people in the population, there are between 6,000 and 8,000 known conditions which are often chronic and life-threatening. Doctors are describing new disorders every day but many people still do not receive an accurate diagnosis for their condition.

The study will take advantage of cutting-edge whole genome sequencing technology at the Universities of Edinburgh and Glasgow, advanced computing facilities at the University of Edinburgh and analytical expertise at Genomics England.

> Health Secretary Shona Robison said: "I am pleased that the Scottish Government is supporting this initiative together with the Medical Research Council. The launch of this study is a significant milestone towards embedding use of this cutting-edge technology by NHS Scotland to benefit patients with rare genetic diseases."

Professor Tim Aitman, Co-Chair of the Scottish Genomes Partnership and Director of the University of Edinburgh's Centre for Genomic and Experimental Medicine, said: "Genomics has come a very long way since the publication of the first human genome in 2001. This collaboration enables the investment made by the Universities of Edinburgh and Glasgow in the latest genome sequencing technology to make a direct and immediate impact on the healthcare of patients in Scotland. We look forward to working on this ground-breaking project with our partners in NHS Scotland and Genomics England."

Professor Zosia Miedzybrodzka, Professor of Medical Genetics at the University of Aberdeen and NHS Grampian, and Chief Investigator of the study said: "The clinicians and scientists delivering NHS Scotland Genetics Services are delighted that patients in Scotland with undiagnosed genetic conditions will have this opportunity to benefit from finding out the cause of their health problem."

Dr Marion Bain, Medical Director of NHS National Services Scotland said: "Cutting edge techniques and equipment mean that it is now possible to sequence an individual's genome in a matter of days. As the cost of sequencing comes down, new analytical techniques are developed and our knowledge in this area grows, there is huge potential to develop more effective and cost-effective diagnostic and treatment services."

Professor Mark Caulfield, Chief Scientist at Genomics England said: "I am delighted that patients living in Scotland will have the opportunity to benefit from genomic medicine by taking part in the 100,000 Genomes Project. Our aim is to bring new diagnoses for patients with rare disease and enable better treatments for patients across the UK."
Alastair Kent, Genetic Alliance UK said: "Whole genome sequencing has become an invaluable route for patients and families to be able to receive an accurate and timely diagnosis of their condition. The 100,000 Genomes Project has helped many families in England to understand their situation and plan for what awaits them. The development of the Scottish Genomes Partnership, and the linkage with Genomics England is very good news for Scottish families and for the whole rare disease community across the UK. We look forward to a long and productive collaboration that will boost research and bring exciting possibilities for patient benefit closer to being realised."
The Scottish Genomes Partnership is a collaboration between the Universities of Edinburgh, Aberdeen and Glasgow, four regional Clinical Genetics Units and four Genetic laboratories commissioned by NHS National Services Scotland. It is funded by the Scottish Government and the Medical Research Council.

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Notes to Editors
Further information about the SGP Rare Disease Research Study is available here:
www.scottishgenomespartnership.org/100,000genomesproject

The **Scottish Genomes Partnership** was founded in January 2015 with a £15m investment by the Universities of Edinburgh and Glasgow, to partner with Illumina for the purchase of state-of-the-art equipment for sequencing human genomes. This equipment enables researchers and clinicians in Scotland to study the genomes of both healthy and sick people on a large scale and faster than before.
In March 2016, the Scottish Government announced a £6 million investment to the Scottish Genomes Partnership, with an award of £4 million from the Chief Scientist Office of the Scottish Government Health Directorates and £2 million from the UK Medical Research Council's Whole Genome Sequencing for Health and Wealth Initiative:

http://news.gov.scot/news/investing-in-cutting-edge-medical-research. This funding allows scientists to decode and analyse the entire genetic make-up of more than 3000 people with cancer and genetic diseases in Scotland. Linking these data with clinical information will enable more precise molecular diagnoses for patients in the Scottish NHS, leading to more personalised treatment and selection of drug therapies. The SGP collaboration with Genomics England has been made possible through this investment.
SGP's research programme is part of the Scottish Precision Medicine Ecosystem which has received separate investment from the Scottish Government for research into pancreatic cancer and multiple sclerosis (http://news.scotland.gov.uk/News/Investing-in-the-healthcare-of-the-future-2255.aspx).
Scottish Enterprise has provided £3.5 million for industry-led projects through the Genomic Medicine Industrial Catalyst Fund: http://www.scottish-enterprise.com/knowledge-hub/articles/insight/genomic-medicine-industrial-catalyst-funding-call

Genomics England is a company owned by the Department of Health and was set up to deliver the 100,000 Genomes Project. This flagship project will sequence 100,000 whole genomes from NHS patients and their families. For more information visit www.genomicsengland.co.uk

How the SGP Rare Disease NHS Research Study will work in practice. The NHS Scotland Genetics Services will identify potential participants and their families. Potential participants will first be approached through their clinical geneticist in the normal course of their treatment. Clinicians in other specialties may discuss referral of potentially suitable patients with colleagues in their local clinical genetics team. The criteria agreed within the Scottish Clinical Genetics Forum are that patients considered will be those who have clear evidence of a genetic/family condition and fit into one of the rare disease subgroups in the list provided by Genomics England, and for whom current genetic and other diagnostic tests cannot identify the disease. If a patient is suitable, the NHS Scotland Genetics Services will manage the consent process, collect samples and data, extract DNA and provide de-identified data on participant symptoms to Genomics England. After whole genome sequencing and data analyses are complete, the NHS Scotland Genetics Services will check all the results received from Genomics England, and provide feedback and clinical results to participants. Clinical management of patients and their families will be carried out within the NHS in the normal way.

Rare Genetic Diseases in Scotland. Rare Disease UK estimates that up to 300,000 people in Scotland's population may be affected by a rare disease over their lifetime. Scottish groups were involved in work to develop a UK Strategy for Rare Diseases, which was endorsed by the Scottish Minister for Health and Wellbeing at its launch in November 2013. A Scottish Implementation Plan, "It's Not Rare to Have a Rare Disease" was published in July 2014 (http://www.gov.scot/Resource/0045/00455471.pdf).
Rare Disease UK hosted a briefing in the Scottish Parliament on 29/11/16 about rare, genetic and undiagnosed conditions and work being carried out in Scotland to put into practice the Scottish Implementation Plan. More information about this event can be found here: https://www.raredisease.org.uk/news-events/news/members-of-scottish-parliament-meet-rare-disease-patients/
The SWAN UK website (www.undiagnosed.org.uk) also provides information about undiagnosed genetic diseases.