Executive Summary

- **The Scottish Genomes Partnership (SGP) is a flagship Scotland-wide genomic research programme between the Universities of Edinburgh, Glasgow, Dundee and Aberdeen, and NHS Scotland.**
- It is a unique synthesis of academic and clinical research expertise in genomics from across Scotland, to improve clinical practice, diagnosis and treatment and to establish optimum ways in which genome technology can be brought into routine clinical care
- The **overall aims of the SGP programme** are:
    - the sequencing of more than 2500 Scottish genomes to understand more about the causes of rare genetic disease and cancer;
    - the diagnosis of rare genetic diseases in a strategic collaboration with the Genomics England 100,000 Genomes Project;
    - the sharing of genetic and genomic information with parallel national and international genome projects; and
    - the development of expertise and awareness of the impact of whole genome sequencing in Scottish healthcare and society.
- SGP was launched by a £15m investment in January 2015 by the Universities of Edinburgh and Glasgow, which led to installation of 10 state-of-the-art Illumina HiSeqX genome sequencing instruments divided between Edinburgh Genomics Clinical Division and the Glasgow Precision Oncology Laboratory.
- SGP's research is funded from February 2016 – February 2019 by a £6 Million joint award from the Chief Scientist Office of the Scottish Government (£4m) and the UK Medical Research Council (£2m).
- Scottish Enterprise has committed up to £3.5m as part of the SGP initiative, by establishing a Genomic Medicine Industrial Catalyst Fund.
- Within Scotland, SGP capitalises on an outstanding ecosystem, which includes the £25m Stratified Medicine Scotland Innovation Centre funded by the Scottish Funding Council, the £7m MRC Farr Institute Scottish Hub and two MRC Molecular Pathology nodes.
- **The SGP has made significant progress since funding was announced in February 2016.**
- A Project Governance structure was established for effective day-to-day management and communication. Specialist project teams manage individual work strands and these are led by cross-programme Steering Group and Operational Management Committee.
- A Scientific Advisory Board, composed of leading experts from the UK and USA, was established in order to advise on current activities and future development.
    - At a meeting in April 2017, the SAB unanimously endorsed the exceptionally high standard of the SGP research and made a small number of recommendations to build further on the tremendous opportunities they saw for the SGP research to transform healthcare in Scotland.
- **A strategic collaboration with the Genomics England 100,000 Genomes Project was established with the direct aim of providing evidence to inform the future delivery of NHSS genetics services.** 1,000 participants (affected individuals and close family members) are being recruited for the Rare Diseases arm of the Genomics England study.

- Intensive working between February 2016 and February 2017 aligned the Scottish protocol with the Genomics England approach, and enabled regulatory approvals from the Public Benefit and Privacy Panel for Health and Social Care, the Research Ethics Committee and NHS Research Scotland.
- Recruitment to the SGP-NHSS study opened in March 2017 and is progressing very well.
- The NHSS Genetics Laboratory leads and Edinburgh Genomics Clinical Division have ensured joint working to develop robust sample handling protocols.
- SGP bioinformaticians and Edinburgh Genomics Clinical Division have invested significant time in establishing the secure transfer of SGP sequence data into the Genomics England bioinformatics pipeline.
- The SGP Steering Group and Operational Management Committee both include health commissioning expertise to ensure that SGP findings can be taken up effectively by NHS Scotland.
- Health Economics data collection will provide essential evidence for NHS Scotland service development.
- The project is establishing joint working between SGP bioinformatics expertise and NHS Scotland clinical scientists to explore how to develop skills and infrastructure for genomic data interpretation in NHS Scotland. This vital collaboration is also reaching out to relevant service leads in NHS England, to ensure learning from other early work in this area.

- **In the cancer work strand, the SGP is providing cutting-edge sequencing and analysis for clinically important and recalcitrant cancers from Scottish cohorts.**
  - The Glasgow Precision Oncology Laboratory is delivering sequencing for all the SGP cancer projects, and this is expected to be complete by the end of 2017.
  - Seven studies have been selected, including retrospective pancreatic, oesophageal and ovarian cancer samples.
  - An exciting new collaboration with AstraZeneca and the Stratified Medicine Scotland Innovation Centre will perform whole genome sequencing on 200 ovarian cancer cases.
  - Using the SGP award, the bioinformatics team in Glasgow is developing new cancer genome analysis pipelines. This leading-edge work is expected to be used for future clinical trials, and the learning from this will facilitate the development of rapid turnaround genomic screening for cancer.
  - SGP bioinformaticians in Edinburgh will soon begin analysis of whole genome sequencing data from the strategically important AstraZeneca study.

- **SGP Rare Disease academic studies are building on Scotland's outstanding academic track record in gene identification and functional analysis of single gene human disorders.**
  - Four studies are underway, seeking new information about unknown molecular influences on motor neurone disease, eye malformation, microcephalic dwarfism and disorders of sexual development.
  - The sequencing of 825 genomes through Edinburgh Genomics Clinical Division is already two thirds complete, with extremely high quality data produced.
  - Initial analyses are underway by SGP bioinformaticians at the University of Edinburgh prior to the selection of the final cases for these projects.

- **An SGP population cohort study is providing a different approach to understanding how the genome influences health and disease.**
    - This is benefiting from previous research with a unique family-based and isolated population cohort, the Shetland-based Viking Health Study.
    - Whole genome sequencing has been completed by Edinburgh Genomics Clinical Division for 500 participants in the Viking Health Study and analysis of these data is underway with linkage to NHS health records.
- **Data sharing has been established as a central principle of SGP, to make best use of resources and expertise.**
    - All SGP's bioinformatics work is underpinned by a commitment to common, optimal informatics solutions for genomics, with standardised data formats and analysis pipelines.
    - All SGP data will be lodged into the European Genome-phenome Archive (EGA) under their controlled access framework. This will ensure that, following publication, the data sets are available to *bona fide* researchers in line with the consent and ethical approvals for each sample set.
    - Exciting work is underway to create a Scottish genomic variation repository using SGP and other available Scottish whole genome sequencing data. This will be a key legacy from the SGP project, as it will provide decisive information for making sense of genomic data, and will underpin genomic interpretation in both clinical and academic environments.
- **Translating research into practice is a key aim of the SGP, and much progress has already been made with this.**
    - The placement of SGP within world class centres for rare diseases, cancer and population genomics, working closely with clinical colleagues, has led to strong integration.
    - SGP activities are shaping the policy direction for genetics in NHSS.
- **Future plans are being configured for sustainable funding that builds on the SGP progress and successes described in this report.**
    - The NHS across the UK is considering how to make the most of genomics for healthcare.
    - The 2016 annual report of the Chief Medical Officer for England, "Generation Genome" was published in July 2017 and makes the case for developing the use of genomics in many areas of healthcare.
    - The NHS Scottish Diagnostic Steering Group and the Scottish Government Rare Disease Implementation Oversight Group are both monitoring SGP progress to consider how the outputs may be used to develop NHSS services.
    - New research funding proposals are being shaped with a view to using genomics to make further advances in understanding the molecular basis of cancer and rare diseases.