Data sharing to support UK clinical genetics and genomics services: a UK perspective

Curating the Clinical Genome 2017

Alison Hall, Head of Humanities, PHG Foundation
The PHG Foundation

The PHG Foundation is a UK independent think-tank focusing on genomics and other emerging health technologies that can provide more accurate and effective personalised medicine.

MISSION - to make science work for health

• Enabling policymakers to deliver rational and responsible health policy
• Close links with Cambridge University
• Active since 1997

www.phgfoundation.org
Challenges associated with using genomic sequence data

- **Interpretation is difficult** and requires access to multiple data types
- **Multidisciplinary expertise** is often required
- **Data storage** is expensive
- **Precarious funding** Reduced funds and increasing demand

**Accurate diagnosis** often relies on finding an unrelated genotype / phenotype match

**Knowledge** is constantly evolving - existing resources may be unreliable

**Incidental findings** of uncertain clinical relevance

**Lack of data sharing** will lead to
  - Misdiagnoses
  - Inappropriate care
  - Inequalities – ‘diagnostic lottery’
  - Inefficiencies

**Lack of NHS designated infrastructure**
Data sharing within the NHS? What does this mean in practice for delivery of clinical services?

- **Policy**: Devolved health services i.e. to England and Wales, Northern Ireland and Scotland

- **Delivery**: Genetic testing services that are delivered by 23 regional laboratories (200,000 tests p.a.) supplemented by local labs having a range of expertise

- **IT**: Services and labs use multiple IT systems: interoperability is poor - data sharing is often technically challenging

- **Funding**: Lack of consistent funding arrangements (genetic/genomic services funded via local and specialised clinical commissioning)

- **Infrastructure**: Lack of designated NHS infrastructure to share clinical, phenotypic and genetic/genomic data

- **Research**: gaps sometimes plugged by hybrid projects having limited funding duration (e.g. Deciphering Developmental Disorders, 100,000 Genomes Project)
Multi-layered legal & regulatory landscape

**Rare disease**
Very rare genomic variants can be identifiable

**Complexity of laws**
Laws, regulations, contractual and professional obligations e.g. consent

**Evolving case law**
Significant cases on consent and duty of care are changing practice

**Confidentiality**
Other laws (common law) govern confidentiality. Some exemptions apply

**Interpretation**
Inconsistencies between local NHS Trust practice

**Data Protection**
Regards genetic and genomic data as ‘sensitive personal data’ having special protection
The dynamic regulatory landscape: EU General Data Protection Regulation 2016/679 (GDPR)

Regulation ‘with regard to the processing of personal data and on the free movement of such data’

- Replaces EU Data Protection Directive
- Directly enforceable in UK and Member States by May 2018
- Scope includes pseudonymised or coded data
- Genetic data may only be processed under certain conditions: but Member States are free to adopt stricter regulation

How the ICO will be supporting the implementation of the GDPR

The government has now confirmed that the UK will be implementing the General Data Protection Regulation (GDPR).

Perceived impediments to data sharing

Survey 2015 (15 respondents)

Survey 2016 (17 respondents)
Data sharing to support UK clinical genetics and genomics services

Challenges

- Legal / regulatory
- Which data
- Where
- How

Recommendations

- Strong leadership
- National agreement
- Operational agreement

Data sharing challenges

www.phgfoundation.org/project/ds/
But challenges remain to secure public trust and confidence

- **De-identification:** increasing the sophistication of technologies for de-identification has limited impact
- **Continuing reservations to data sharing for commercial use (IPSOS MORI)**
  - Low public awareness of current data use
  - Support for more effective ‘within-NHS’ sharing
  - Genetic data ‘both most private, and most potentially valuable’
  - 54% ‘ask permission’; 17% ‘opposed to sharing for research’
  - Hierarchy of WHY, WHO, WHAT and HOW
  - Changing safeguards did not change outcomes
- **Demonstrating trustworthiness:**
  - Privacy is not ‘absolute’
  - Wider sharing could improve diagnosis, care, and treatment
  - But privacy and security concerns need to be properly addressed through demonstrating ‘trustworthiness’
A brave new world for UK data sharing?
Regulation and governance

• **Legislation:** A new Data Protection Bill will implement GDPR and “create a data protection framework suitable for our new digital age” [“GDPR ++”]

• **Protecting against misuse of predictive genomic data for non-medical purposes**
  
  E.g. employment or insurance

• **Extension of UK Concordat and Moratorium**
  Prevents insurers from requesting disclosure of predictive genetic tests

• **Stronger sanctions for deliberate misuse** i.e. for deliberate and negligent re-identification of anonymised data
  (UK Digital Strategy)
A brave new world for UK data sharing? Infrastructures and processes

- **Secure, robust infrastructures for sharing genomic and phenotypic data**: Building on the Decipher NHS Consortium and the 100,000 Genomes Project, lab reprocurement will establish a co-ordinating centre and genomic knowledge centre to oversee local and regional labs.

- Adoption of structured data sets, agreements and frameworks

- **Improved standardisation**:  
  - Professional endorsement of consistent data collection, analysis, interpretation and reporting, e.g. BSGM adoption of ACMG Variant interpretation guidelines
  
- Robust and proportionate security standards 
  - e.g. National Data Guardian’s 10 Data Security Standards require that technology is secure and up-to-date
Promoting more radical approaches
Hearts and minds?

• **Proactively seeking patient preferences for wider sharing:** Model consent/opt-out (National Data Guardian – independent advisor to the NHS)

  Data can be shared for *direct care* on the basis of implied consent but new consent models are being developed which require explicit consent

  • Is clinical genomic data sharing part of ‘direct care’?
  • Is consent required to ensure a legitimate legal basis for international transfers (in absence of equivalency/adequacy?)

• **Improving genomic literacy** to promote understanding that only a small proportion of genomic data is a reliable predictor of future ill health

• Advocating for a **NOVEL SOCIAL CONTRACT**

Particular challenges faced by the UK

• Implementing the ‘right to be forgotten’ – on reaching 18, adults may force media platforms to delete information held about them.

➤ Uncertain relevance to clinical care and research

• The legal grounds for lawful processing change under the GDPR to remove some types of research using ‘legitimate’ processing which must be done in the ‘public interest’

➤ This needs clarification

• The regulatory framework needs to be consistent with mainstreaming these tests across clinical sectors

• The legislative timetable imposed by the BREXIT process, and political drivers mean that the shape of the future UK regulatory framework is currently unclear
Priorities for policy making
Prioritise why, who, what and how?

- Demonstrate necessity
- Emphasise clinical utility
  - Sharing across health and social care
  - More use of role-based access
- Evidence proportionality
  - Minimising disclosure - maximising utility
- Robust regulatory framework
  - Data security standards
  - Legislative support where necessary
  - New categories for accessing genomic data [Controlled – Restricted – Open Access]
- Greater harmonisation
  - Consistent processes and terminology
    - GA4GH Data Sharing Lexicon
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For more information contact:
Alison.Hall@phgfoundation.org
Sobia.Raza@phgfoundation.org