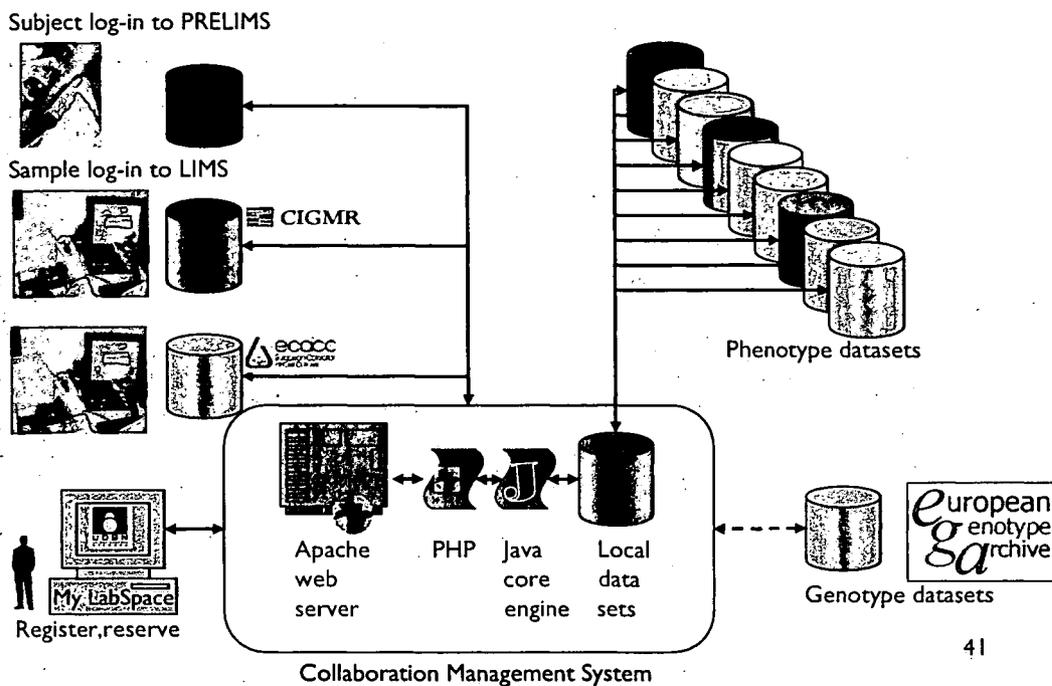
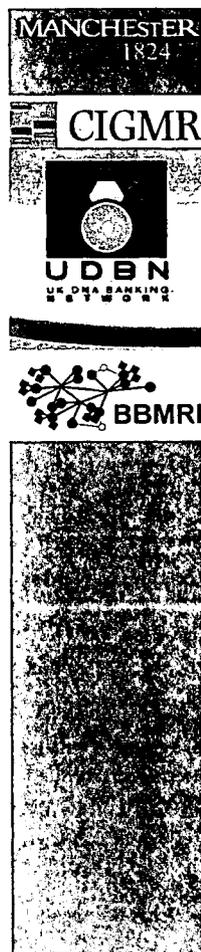




Data management



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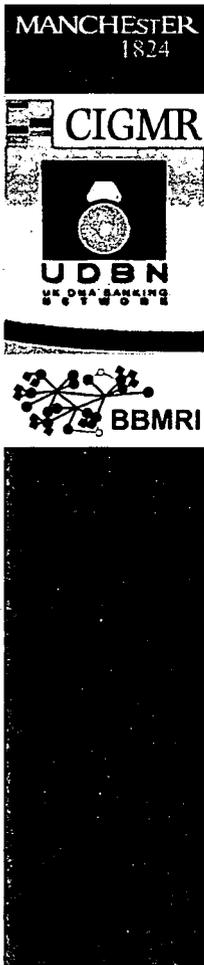


European Genotyping Archive



- EGA is part of Ensembl
- EGA
 - provides security to allow access rules defined by data depositor
 - is accessible www.ebi.ac.uk/ega/
 - is fully searchable with respect to SNPs, genomic regions, studies (case, population, family) etc
 - supports deposit and retrieval of genotype data in an internationally agreed standard format
 - will support appropriate raw data format in discussion with genotype providers and analysts
 - will exchange data with other appropriate data archive sites
 - will support necessary links to other related biological data resources such as genome browsers, clinical phenotype databases, etc

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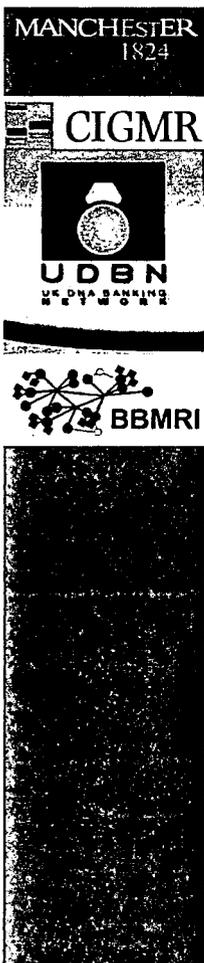


EGA services



- EGA will provide service resources:
 - Complete genotype estimation by imputing missing SNPs
 - Functional markup of genotyped individuals based on known SNP consequences provided by Ensembl
 - A population structure resource with the ability to study characteristics such as admixture
 - Separation of a publicly available list of variants and a restricted access collection of genotypes with potential phenotypic information associated to them
 - Downloads of all data in a single study
 - Queries on specific genome regions, diseases or phenotypes. Data mining based on BioMart.
 - Public data (summaries of information) will be provided through Ensembl and other sources

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UDBN promotes collaboration

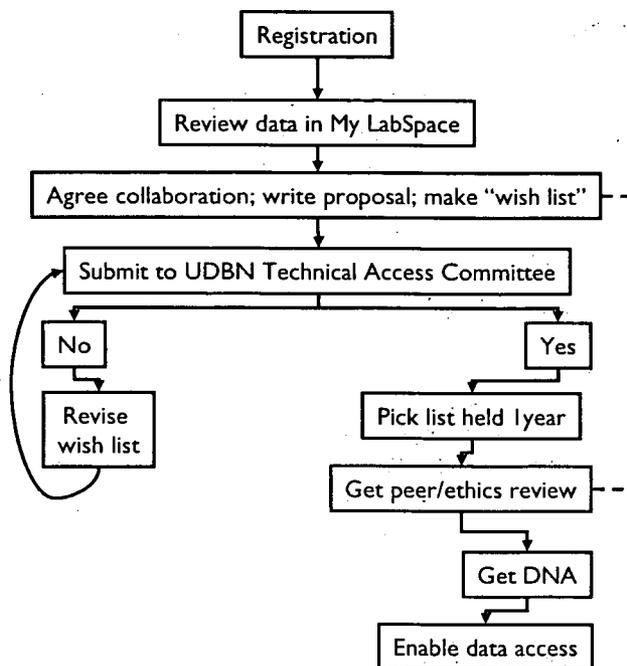
- Web-based process
 1. Registration of new users
 2. User browses summary data on collections
 3. User contacts a collection consortium
 4. Collaboration established
 5. Samples are identified
 6. Approval process
 7. Aliquots despatched
 8. Data returned



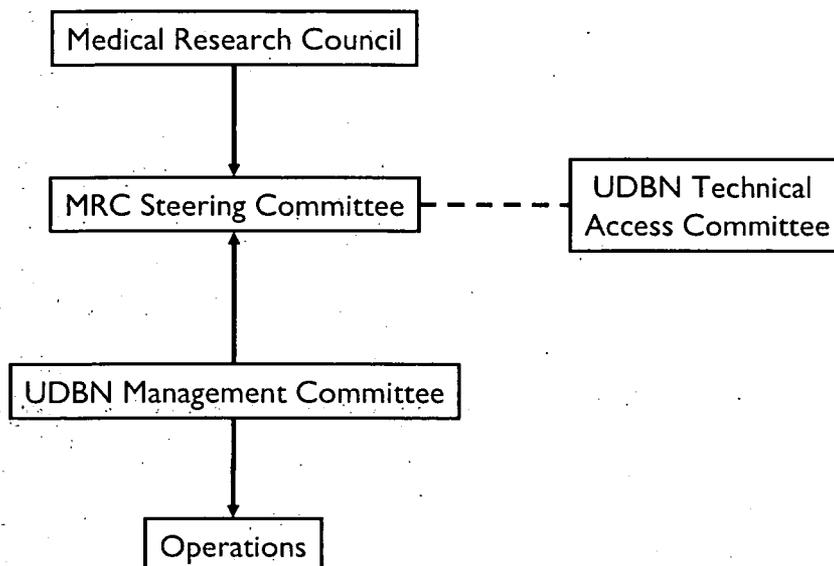
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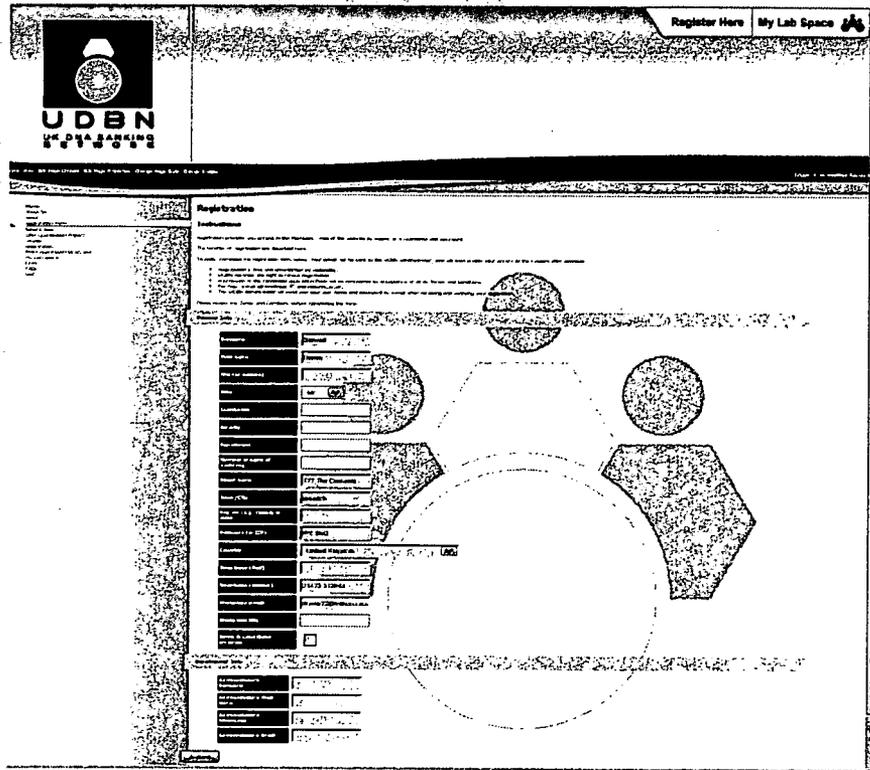
Overview of access



UDBN Technical Access Committee



Registration



The screenshot shows a web browser window with the UDBN logo in the top left corner. The page title is "Registration". The main content area contains a registration form with various fields and sections. The form includes a "Personal Information" section with fields for Name, Date of Birth, and Gender. There is a "Contact Information" section with fields for Email and Phone. A "Registration Details" section includes checkboxes for "I agree to the terms and conditions" and "I agree to the privacy policy". The form also features a "Submit" button and a "Cancel" button. The background of the form has a faint graphic of a person's head and shoulders.

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Registration

- The Registered User agrees
 - To use data only for the advancement of medical research.
 - To preserve the confidentiality of data and metadata.
 - Not to give access to data. UDBN can inspect user's data security.
 - To acknowledge UDBN in any publication.
- UDBN establishes *bona fides* of applicant
 - Confirms email address
 - Confirms employer is a legitimate research organisation

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Browse the collections...

Home | About Us | test2 | Registration Form | What's New | DNA Quantitation Project | Studies | Registration | From registration to access | My Lab Space | Administration Protocols | Getting Started | Projects | Browse | Relationship Manager | System Administration | Entity Manager | Links | FAQs | test

Admin Area | Manage Entity | Manage Relationships | Add to My Lab Space | Logged in as martiny | log out

Browse

About Browsing

If you wish to identify a collection by its ICD number then you can click through using the *Areas of Study* table. If you already know the name of the study you are looking for, you can find it in *Quick Links*.

Areas of Study	Quick Links
C00-048 Neoplasms	Edlib UK The genetic basis of coronary heart disease through the study of affected sibling pairs (ASPs)
E00-E90 Endocrine, nutritional and metabolic diseases	P3G P3G Observatory Type 2 Diabetes T2D trios (Warren II) study. This study contains samples from patients with Type 2 Diabetes and both of their parents who may, or may not, have diabetes themselves.
F00-F99 Mental and behavioural disorders	Glomerulonephritis 2000 Glomerulonephritis DNA Bank Unipolar Depression Unipolar Depression
G00-G99 Diseases of the nervous system	Parkinsons Disease Parkinsons Disease Alzheimers Disease Late onset Alzheimers Disease Bright Study
H00-H59 Diseases of the eye and adnexa	The MRC BRIGHT study has recruited family-based and case control resources to provide a robust national repository for the evaluation of the genetic basis of hypertension.
I00-I99 Diseases of the circulatory system	
J00-J99 Diseases of the respiratory system	
L00-L99 Diseases of the skin and subcutaneous tissue	
N00-N99 Diseases of the genitourinary system	
Q00-Q99 Congenital malformations, deformations and chromosomal abnormalities	

Projects

DNA Quantitation Project
Click here to find a description of the pilot phase.



Browse the data...

Relationships | Add to My Lab Space

Data on Type 2 Diabetes

Use the query tool below to search the Type 2 Diabetes study. If the study looks to contain samples you are interested in, y collaboration.

Contact UDBN Partner

Options | **Details**

Change which phenotypes are displayed below | Currently displaying 35/35 phenotypes
Contact UDBN Partner | Contact the study investigators for more information

Patient type

Analyse this phenotype in more detail

Categories	Total
5	732

Discrete Values

Control	1	0.14%
Father	188	25.68%
Mother	176	24.04%
Proband	205	28.01%
Sibling	162	22.13%

Gender



...and contact the collectors

Relationships Add to My Lab Space

Data on Type 2 Diabetes

Use the query tool below to search the Type 2 Diabetes study. If the study looks to contain samples you are interested in, y collaborate with:

[Contact UDBN Partner.](#)

Options

Options **Details**
Change which phenotypes are displayed below **Currently displaying 35/35 phenotypes**
[Contact UDBN Partner](#) [Contact the study investigators for more information](#)

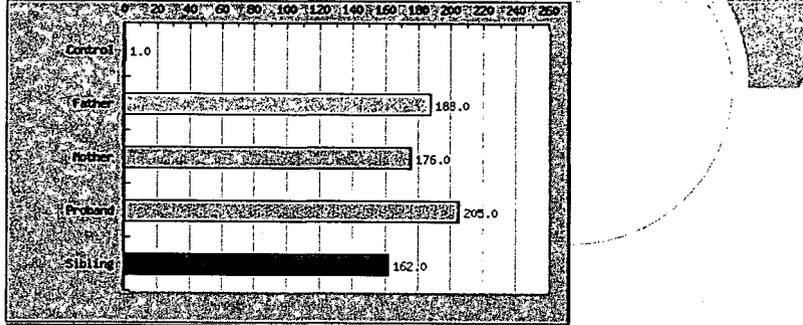
Patient type

Analyse this phenotype in more detail

Categories	Total
5	732

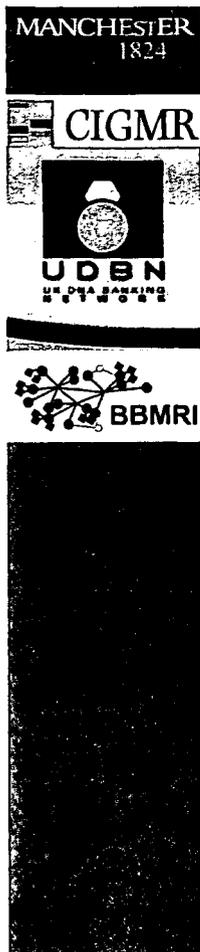
Discrete Values

Control	1	0.14%
Father	188	25.68%
Mother	176	24.04%
Proband	205	28.01%
Sibling	162	22.13%



The Technical Access Committee decides if ...

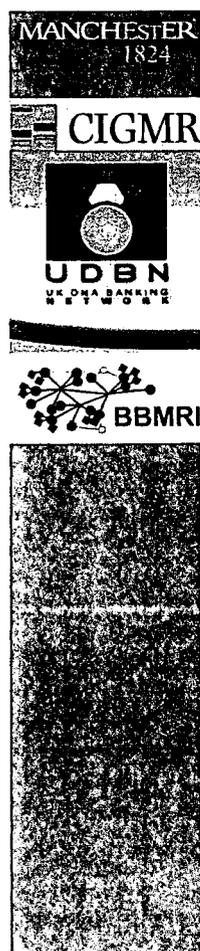
- The platform (s) is appropriate for the tests.
- There is no other platform available that uses significantly less DNA.
- The out-sourced genotyper is reputable.
- The amount of DNA/sample requested matches the amount of DNA/sample required.
- The requested concentration of DNA is reasonable for the platform.
- The test(s) can be performed with comparable reliability on cell line DNA or on WGA-DNA as on blood-derived DNA
- Less than 20% of the tests on the samples requested have not been performed or submitted for approval previously.
- The plan for data access is reasonable.
- The peer review processes of the funder are acceptable.



Terms and conditions of release of DNA

- The Recipient is a registered user of the UDBN website and will:
 - Not sell DNA or use for profit or any other commercial gain
 - Only use DNA for previously specified tests
 - Give UDBN access to all the data within 10 days of the date of publication
 - Agree that UDBN will publish summary data via the website
 - Acknowledge UDBN

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Contents

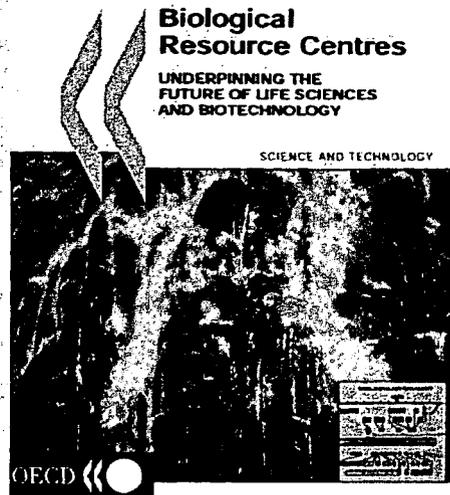
- Definitions
- Why is biobanking important?
- Types of biobank
- An example
- The growth of biobanking
- A development strategy
- Challenges in biobanking

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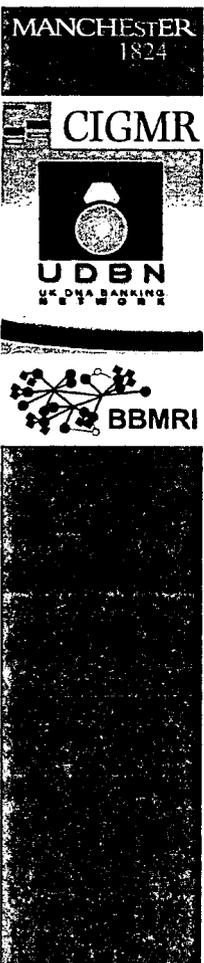


The growth of biobanking

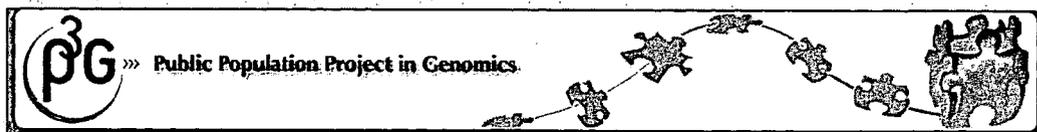
- Global policy context: OECD
- If BRCs are to underpin the future of life sciences and biotechnology, then we need to establish:
 - National BRCs
 - An accreditation system based on scientifically acceptable objective international criteria
 - International linkages
 - Internationally co-ordinated and harmonised operational parameters
 - A global BRC network
 - Paris, 2001



Organisation for Economic Cooperation and Development
Task Force Chair: Hideaki Sugawara

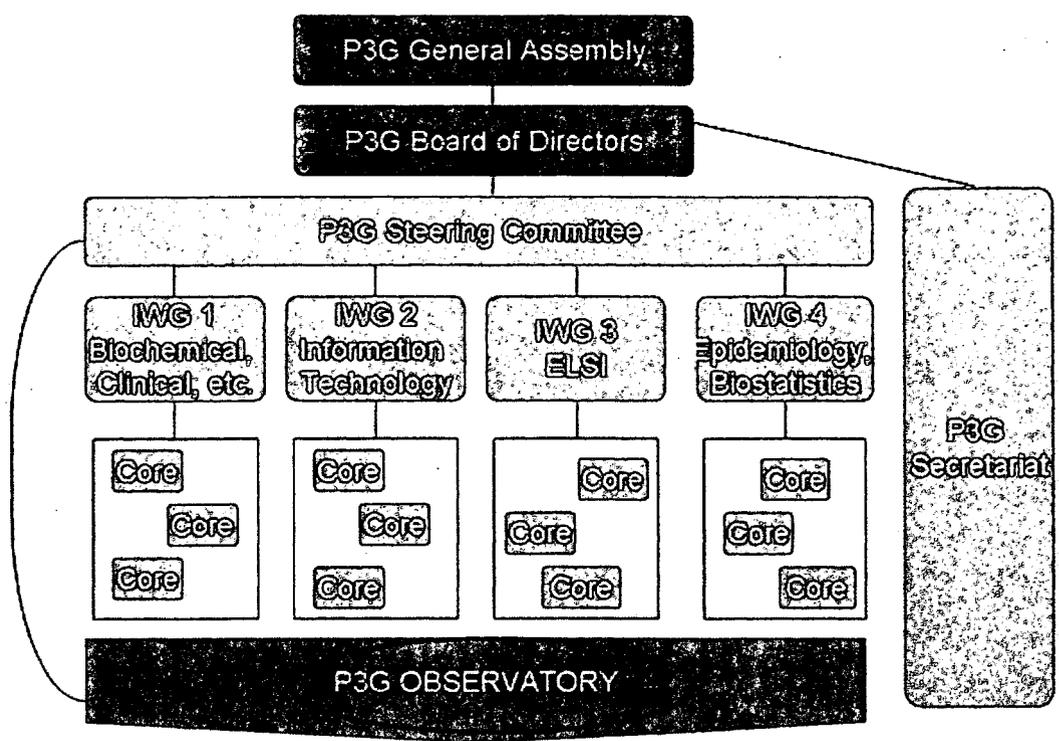


Global biobank harmonisation



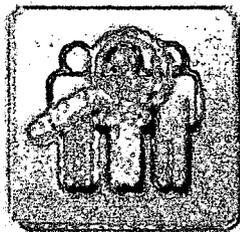
- The aims of P3G are to:
 - Foster collaboration between biobanks
 - Optimize the design, set up and research activities of population-based biobanks
 - Promote harmonisation
 - Facilitate transfer of knowledge and provide training

P³G Operational Chart

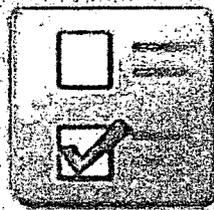


P3G Observatory

- Study Catalogue
 - A catalogue of 91 large population-based studies around the world
 - Plus comparison tools

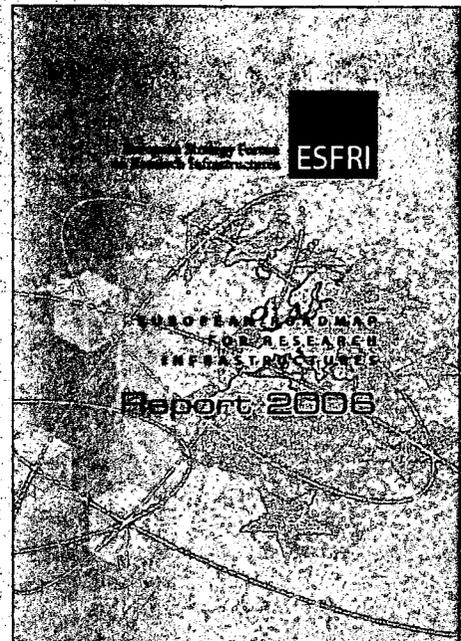


- Questionnaire Catalogue
 - A catalogue of 17 selected cross sectional questionnaires
 - Plus comparison tools



European infrastructure development

- European Roadmap for Research Infrastructures
 - Expensive facilities
 - Data links
 - Resource sharing
 - Projected budget: €27 billion
- Areas of research
 - Biomedical and life sciences
 - Social sciences and humanities
 - Environmental sciences
 - Computer science
 - Energy
 - Material sciences
 - Astronomy and particle physics



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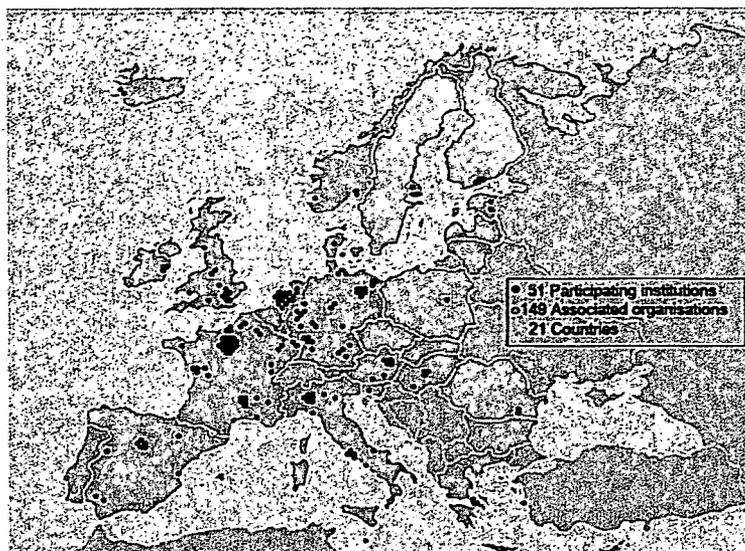
Biobanking and Biomolecular Resources Infrastructure (BBMRI)

- Preparatory phase funded Feb 2008
- Aim of preparatory phase
 - To prepare contracts between institutions and funders for “a pan-European and broadly accessible network of existing and de novo biobanks and biomolecular resources”

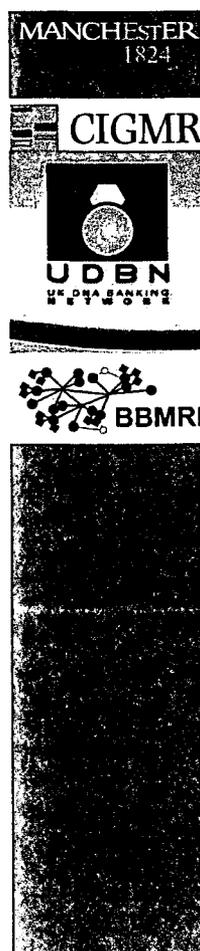
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BBMRI participants



- Statutory funders from 10 EU states
- 40 institutions with substantial resources



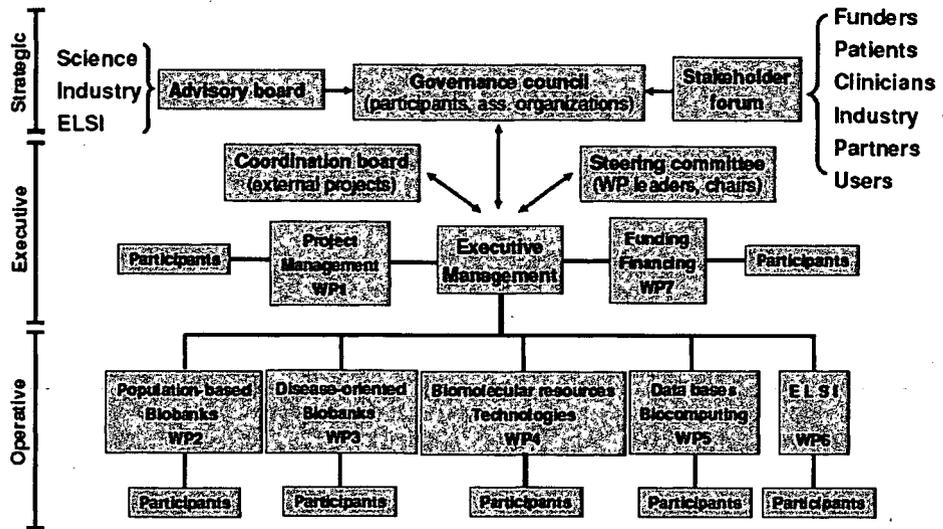
Current available resources

Biobank format	Number of biobanks	Number of samples
Population based	36	2.4m
Disease oriented	67	10m
Other	1	1500
Total	104	12.5m

Total current investment €340m
 Total approved investment €138m
 Anticipated BBMRI infrastructure investment €170m



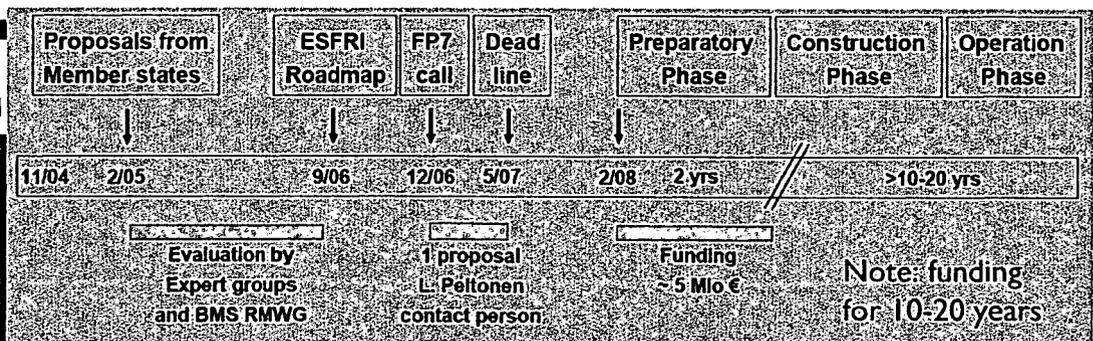
Preparing BBMRI



- N.B. Advisory Board is international: its aim is to ensure harmonised standards worldwide

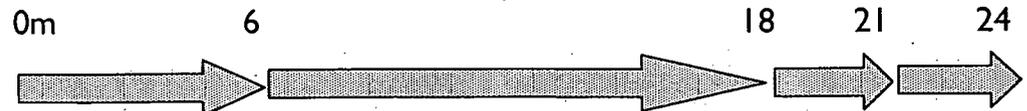


BBMRI timetable





Timetable details



• Evaluate existing resources and solutions
 • Identify open issues

Concept for:
 • Integration of existing resources
 • Integration of future resources
 • Process standardisation, certification
 • Access rules
 • Incentives and benefits
 • International exchange of samples and data

Operations concept

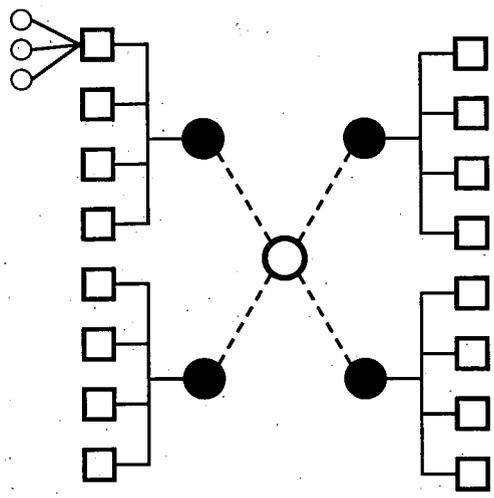
Financial plan

Implementation plan

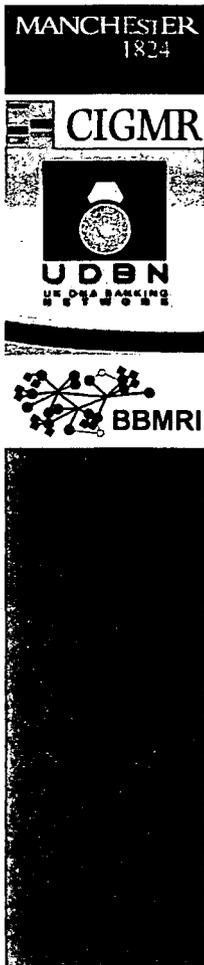
Contracts



What will the BBMRI network look like?



○	□	●	○	—	---
Associated partners, hospitals, universities, service providers	Domains (biobanks, biomolecular resources, tech centres)	Hubs (resource-based, geographically-based)	Virtual hub	Samples and data	Data



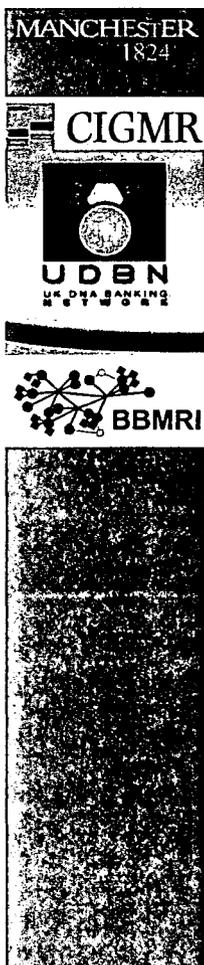
BBMRI in the UK



RESEARCH
COUNCILS UK

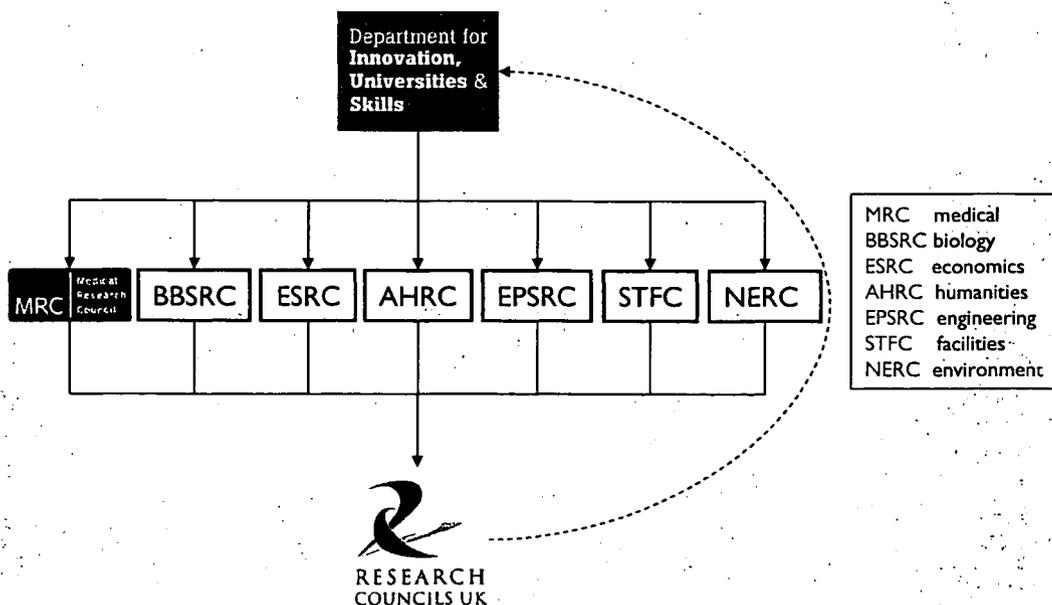
- Research Councils UK has adopted the European Roadmap for research infrastructures as part of the creation of the European Research Area
- BBMRI is a 'planned facility' named in the Large Facilities Roadmap of RCUK

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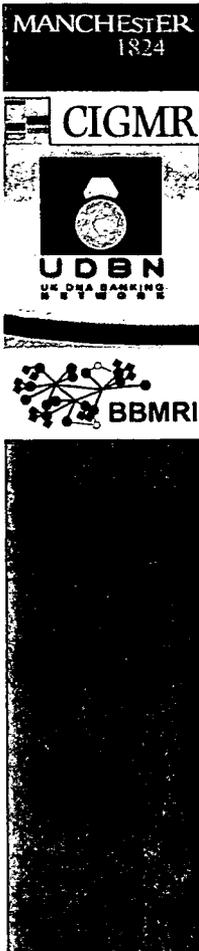
What is RCUK?

simplified



RCUK is responsible for production of a Large Facilities Roadmap

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DIUS supports Large Facilities

- Large Facilities Capital Fund
 - £100m (= ¥200m) p.a. support for Research Councils' investments that "could not be sensibly accommodated" from within Research Council budgets.
 - Used to help pay capital costs
- Work has started to develop the scientific and business case for UK BBMRI
- This will then allow
 - Short-listing by MRC
 - Prioritisation by RCUK
 - Consideration by government
 - Agreement by government and RCUK
 - Approval by minister

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Contents

- Definitions
- Why is biobanking important?
- Types of biobank
- An example
- The growth of biobanking
- A development strategy
- Challenges in biobanking

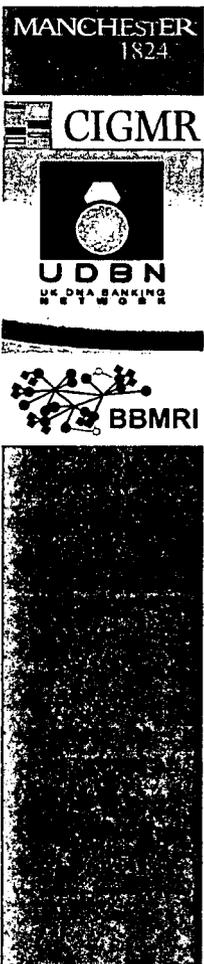
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A development strategy

- Fund first things first
- Prepare for the infrastructure
- Call for infrastructure proposals
- Build the infrastructure
- Fund the infrastructure
- Pathfinder projects
- 'Fair access' principles

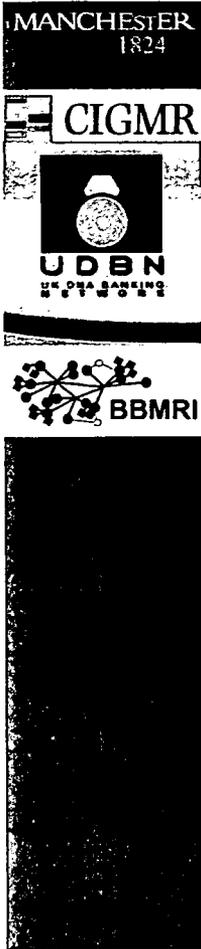
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Fund first things first

- Post-genomic molecular epidemiology requires the efficient accrual, proper management and seamless integration of very large numbers of samples and datasets
 - This is the 'cutting edge' of the science today
 - The primary weakness (loss of statistical power) in large genome wide association studies has arisen from
 - Poor quality samples
 - Poor quality sample management
 - Inadequate phenotyping can also contribute
 - This loss of power will increase risk of failure when we want to look for more subtle genetic effects
 - We shall be forced to start accrual all over again
 - But many investigators will lobby primarily for 'cutting edge' technology
 - There is evidence that cutting edge genotyping technologies are delivered better by industrial enterprises than academic institutions
- Fund management separately from accrual
 - Management requires expertise in logistics and laboratory methods
 - Accrual requires epidemiological and clinical expertise
- This means building the management infrastructure first
 - In the UK, accrual was funded first causing delay, conflict and confusion

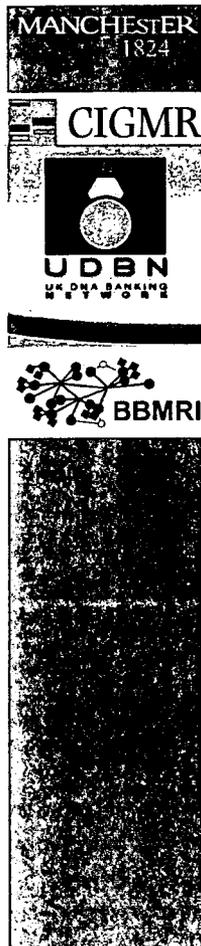
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Prepare for the infrastructure

- Preparatory Phase
 - Funder organises national workshops to
 - explain the vision for national biobanking infrastructure to
 - overcome the fragmentation of national resources
 - meet the challenges of post-genomic molecular epidemiology
 - examine issues of ownership, sharing, security
 - Funder establishes an international committee to facilitate
 - OECD vision of global harmonisation, standardisation and integration
 - increased cohesion of the national research communities
 - Funder develops 'fair access' principles
 - Funder promotes harmonisation and standardisation with other relevant funders
 - Complex projects may have >1 source of funding

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Call for infrastructure proposals

- Preparatory Phase
 - Funder issues call for proposals to host the infrastructure
 - Select applicants with experience in
 1. service provision
 2. large-scale infrastructure management
 3. web-based data networks
 4. laboratory methods research
 5. genetics, epidemiology, cell culture
 - Do not accept commercial applicants
 - problems of public trust and engagement
 - but public-private partnerships may address these problems

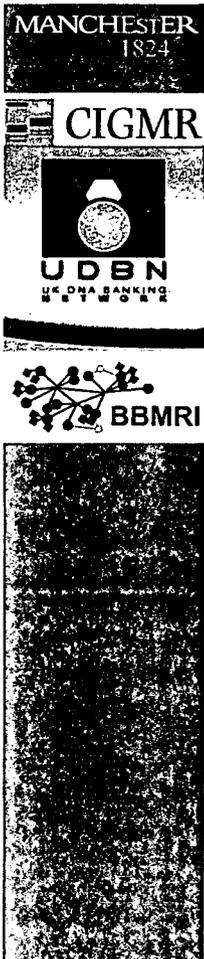
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Build the infrastructure

- Construction Phase by selected applicant
 - Policy development
 - Hold regular networking events nationwide
 - Work with stakeholders to seek to undertake management of legacy collections
 - Make policy recommendations on consent, privacy, public engagement, access
 - Implement pathfinder collection projects that
 - Infrastructure development
 - Develop web-based accrual project management software
 - Develop web-based access mechanisms
 - Identify / create a suitable common data centre
 - Develop central consent and IPR management
 - Implement high throughput sample processing (DNA extraction; serum aliquotting; quantitation, storage, retrieval)
 - Second site storage to guard against catastrophic loss
 - Establish sample replenishment methods
 - Gain ISO accreditation for sample and data management
 - Identify research needs for sample and data management methods
 - Pilot operations
 - Work closely with pathfinder projects
 - Identify and harmonise with genotypers etc
 - Propose cost recovery mechanisms

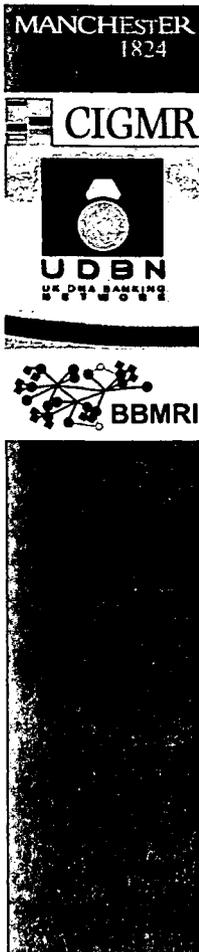
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Fund the infrastructure

- Rolling funding mechanism is needed
 - First phase: can the applicant meet the targets?
 - Second phase: provide secure long-term funding to re-assure collectors
 - Biobanking as part of the national research infrastructure development programme

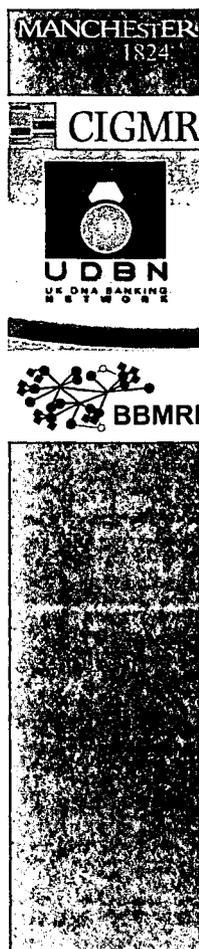
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Pathfinder projects

- As soon as the infrastructure has been funded, issue a call for proposals
 - for new collections
 - to investigate high-impact health problems
 - with evidence of international cooperation / collaboration
 - with novel conditions of grant
 - Patient (subject) gifts sample to funder
 - Patient gives informed general consent
 - Accrual should focus on improving phenotype data
 - Use standard questions where possible
 - Use data standards (HL7 / SNOMED)
 - Collection is a “shared scientific resource”
 - Make resource available after publication
 - On-time completion of pathfinder deliverables will lead to a tailored call for genotyping etc.

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Funding to analyse a resource

- Investigator agrees
 - To use a genotyper etc approved by the biobanking infrastructure
 - That genotyping data will go directly from genotyper to common database
 - That all versions of data will be stored there
 - That third party *bona fide* potential collaborators will have access to summary data at time of publication

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UDBN's 'fair access' principles

- Based on 2003 UNESCO International Declaration on Human Genetic Data
 - Article 18 : Circulation and International Cooperation
 - States should regulate the cross-border flow of data and samples “so as to foster international cooperation and ensure fair access”

Article 18 – Circulation and international cooperation

(a) States should regulate, in accordance with their domestic law and international agreements, the cross-border flow of human genetic data, human proteomic data and biological samples, so as to foster international medical and scientific cooperation and ensure fair access to these data. Such a system should seek to ensure that the receiving party provides adequate protection in accordance with the principles set out in this Declaration.

(b) States should make every effort, with due and appropriate regard for the principles set

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UDBN's 'fair access' principles

- Fair to the subject
 - Privacy and confidentiality
 - Ethical use of samples and data
 - Consent management: national open methods to permit effective withdrawal of consent
 - Public engagement: understanding and goal-setting
- Fair to the collector
 - Right to first access
- Fair to the recipient
 - Collaboration management: ensure transparency
 - Access to usable published / unpublished data
 - Long term availability of sample: stock control
 - Minimum of administration
- Fair to collector's and investigator's institution
 - IPR management: long term tracking of samples and data



The critical advantage of access via collaboration is that it corresponds to existing practices which are self-monitoring and which we know work well

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