

Ethical, legal and social implications: whole genome research

- Dr Harriet Teare
- HeLEX Centre, University of Oxford





Disclosure slide

• I have no conflicts of interest to declare





Outline

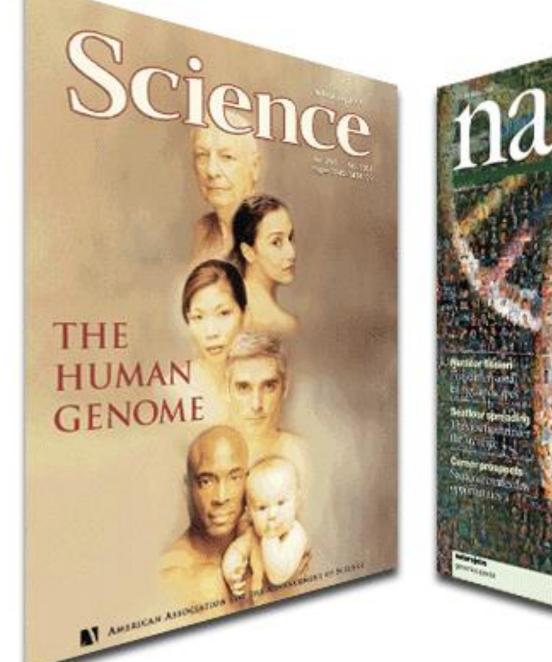
- How research is changing
- ELSI challenges
- Role of patients
- Example project RUDY

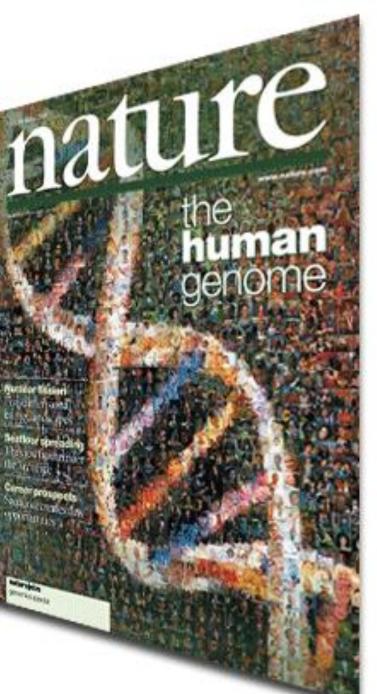


Outline

• How research is changing





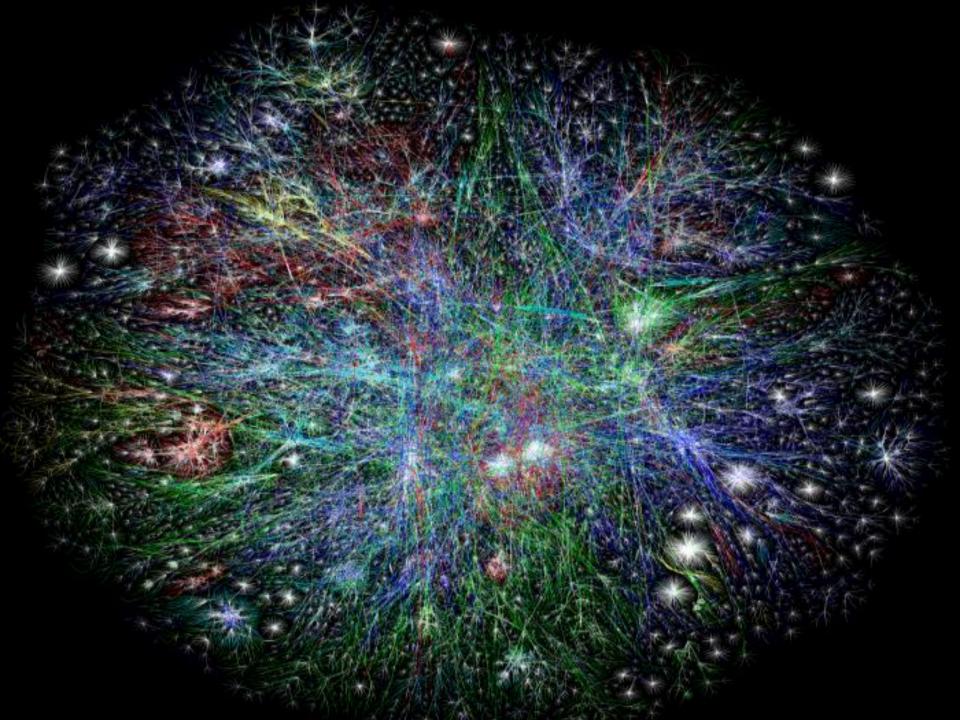


Human Genome Project



- First 'big science' project in genomics
- Not hypothesis-led but establishing an infrastructure
- Change from competitive to co-operative
- Co-ordination of groups around the world
- Interdisciplinary teams with specific skills
- Open access to data





Biobanks and patient registries





Technologies at Oxford



Outline

• ELSI challenges









UNIVERSITY OF



Protecting Privacy: Difficult to keep the identity of participants anonymous: - When data is shared - Placed on the web in open access repositories **BUT: not sharing goes against** current funders' policies scientific need for large sample sizes

HeLEX





Distinguishing Individuals

2008

Individuals could be distinguished in GWAS using only summary statistics Homer, N. *et al.* PLoS Genet. 4, e1000167 (2008).





OXFOR



Re-identification of male participants



- Using SNPs on Y chromosome
- Linked to data found in publicly available datasets on Internet
- Gymrek, M., Science 339, 321–324 (2013).





Incidental Findings

HeLEX



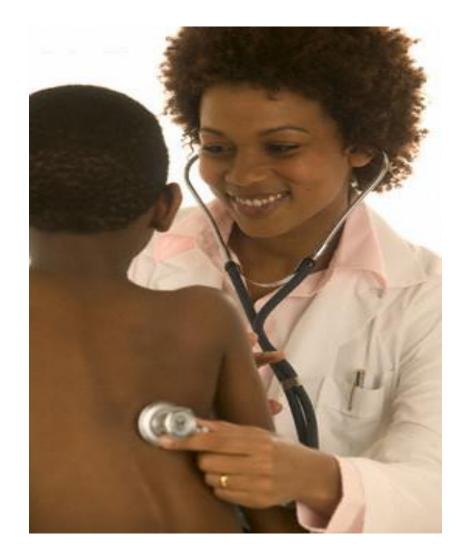
ntre for Health, Law and Emerging Technologies at Oxford

Findings that are not part of the original research aims but have <u>clinical significance</u>

- Comprehensive nature of WGS
- What should we feedback in terms of individual level findings?
- Who decides and who should feedback findings to whom?
- How should we protect the right NOT to know?
- What is the responsibility of the researcher:
 - To look?
 - To feedback?
 - Beyond the project?

Two different domains





Consent and withdrawal

Technologies at Oxford





IMPAKT Breast Cancer Conference 2014



The Albrecht Report - January 2013

(on the proposed EU data protection regulation)

- 1. Removes exemptions that applied for medical research
 - Cannot keep data indefinitely, or use for secondary research purposes without consent
 - Medical research must comply with general requirements
- 2. Tightens requirements for consent

'if you want my data, ask for my consent' - not implied or tick box

- Freely given, specific, informed and explicit
- Questions whether broad consent is lawful







Patient involvement – samples and data



- · Different risks related to donation of samples and data
- What does it mean to withdraw?



Outline

• Role of patients



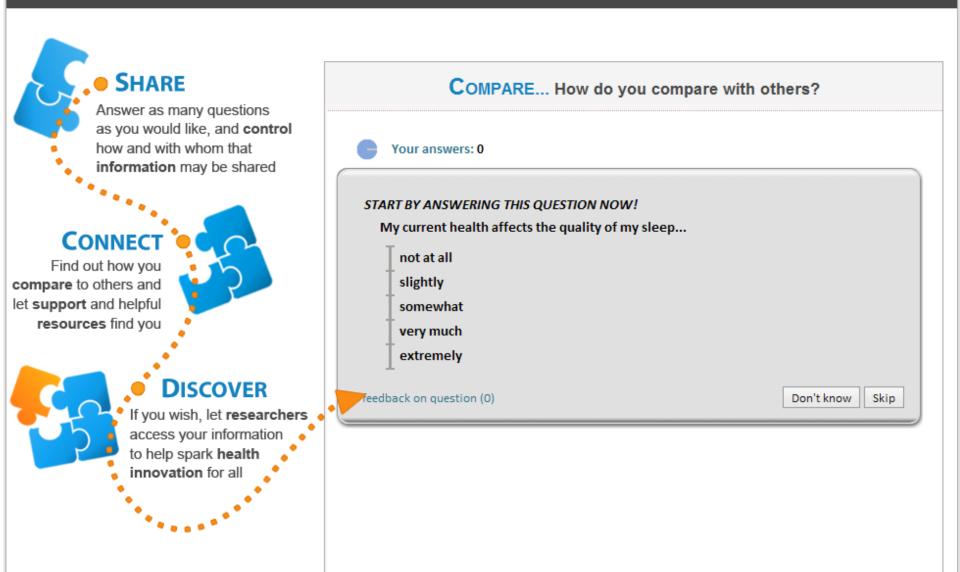


Patient Centric Initiatives (PCI)

Individuals at the centre of decision-making **Communication and interactions** over time **Empowerment of individuals Partnerships** Kaye J et al. 2012 Nature Review Genetics



Finding ways to improve health with the registry for all



patientslikeme*



ene Contact Un

Patients + Data = Insight

Patients are no longer just healthcare consumers. They are healthcare customers. PatientsLikeMe provides deep insight into your target patients' attitudes, beliefs, behaviors, and outcomes.

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Enhance the value of your current and future products and services to these who actually use them.

-

Find out more

Health Plans w

Optimize healthcare resources by delivering the right intervention to the right patient at the right time.

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Not a representative of a pharmaceutical company or health plan? Learn how else we pertner on behalf of patients.

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We're crowd-sourcing health discovery by helping anyone create group health studies.





Clinical Studies at Scale

Need to conduct commercial or academic studies at Internet scale, engage our participant pool, and collect data with Genomera? Find out here how we can help.



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Genomera in the Press

People are excited about us! And we're excited to be a part of a growing movement, with recent coverage in Wired, Nature, and more. Read about the movement here.



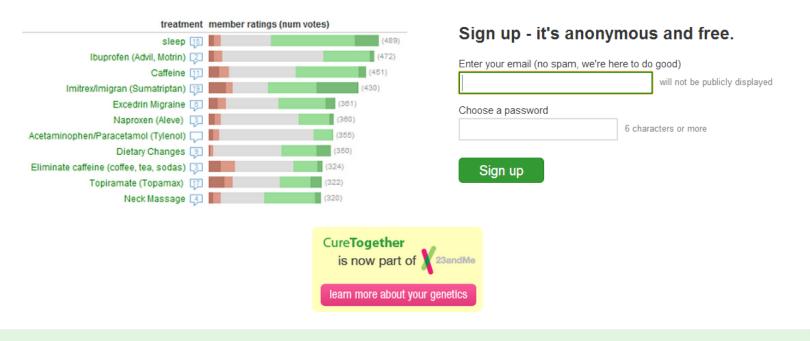
Rocking it with Rock Health

We're pleased to announce that we were selected from over 350 applicants to be part of the Rock Health accelerator in San Francisco. Check out Rock Health here.



The smarter way to find the best treatments.

Get access to millions of ratings comparing the real-world performance of treatments across 637 health conditions.



Why you will love CureTogether

- 1. You'll learn from people going through the same thing.
- 2. You won't see ads or time-wasting content.
- 3. We'll work hard to help you achieve better health.
- 4. It's anonymous, so you're safe and secure.

Recognition

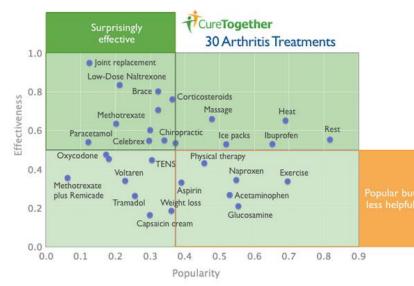




About CureTogether | Our Team | Ongoing Studies | Buzz

800 Patients with Arthritis Report Which Treatments Work Best

October 18th, 2011 Alexandra Carmichael Posted in Infographics, Research Findings | 1 Comment »



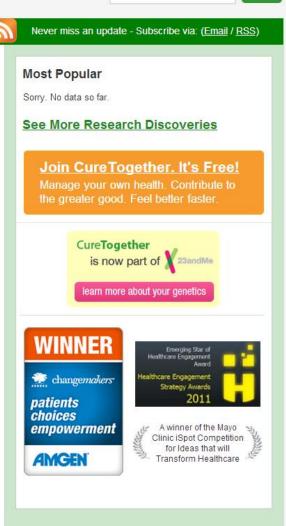
For the live-updated, interactive version of this infographic, click here.

We're excited to announce the very first results of our ongoing study of <u>Arthritis</u>. At CureTogether, 798 people with Crohn's came together to rate 30 treatments. When we gather 1,000 treatment ratings for a condition, we publish an infographic, and Arthritis has just crossed that threshold.

Patients rate heat, rest, and massage as helpful for their symptoms, as well as joint replacement and LDN (see the top of the green part of the chart). They rate Glucosamine, Acetaminophen, and Capsaicin cream as not helping as much. (see the bottom of the green part of the chart above.)

The top 10 overall treatments reported for Arthritis are, as a list:

- Joint replacement
 Low-Dose Naltrexone (LDN)
 Brace
 Corticosteroids
 Massage
 Heat
 Methotrexate
 Vicodin
- 9. Chiropractic adjustment
- 10. Rest



Make experience count

Transforming patient experiences into clear understanding



Home / What we do

Communities

Support and answers for patients, families and carers

Find your community ▶

GP Communities

All the tools a GP surgery needs to get patients' opinion and feedback

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Patient-reported outcomes made easy

Learn more Learn

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Outline

• Example project - RUDY









Uniting Patients and Researchers to Investigate Rare Diseases

National Institute for Health Research Rare Diseases Translational Research Collaboration in Musculoskeletal Diseases

- MK Javaid, R Luqmani, P Wordsworth
- H Teare, K Melham J Kaye
- National Collaboration...

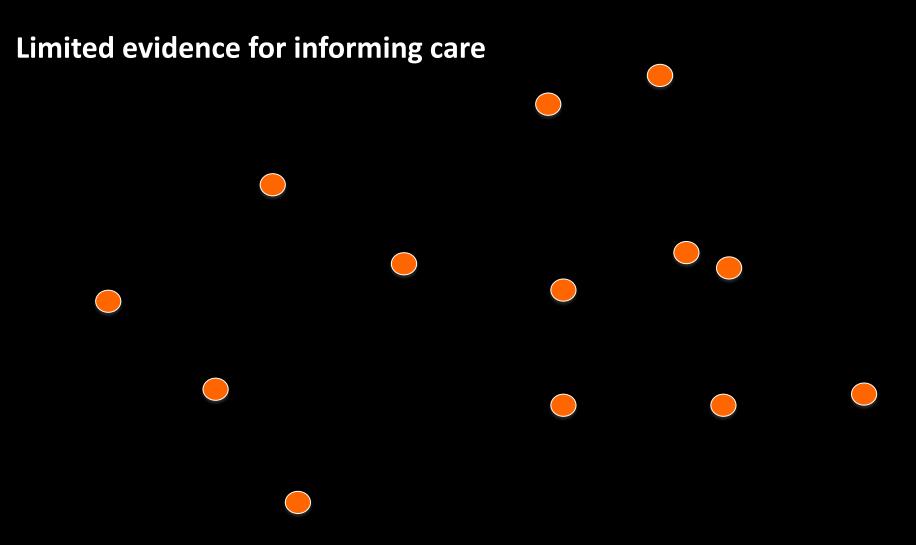
Nuffield Department of Orthopaedics, Rheumatology and Musculoskeletal Sciences

Nuffield Department of Population Health

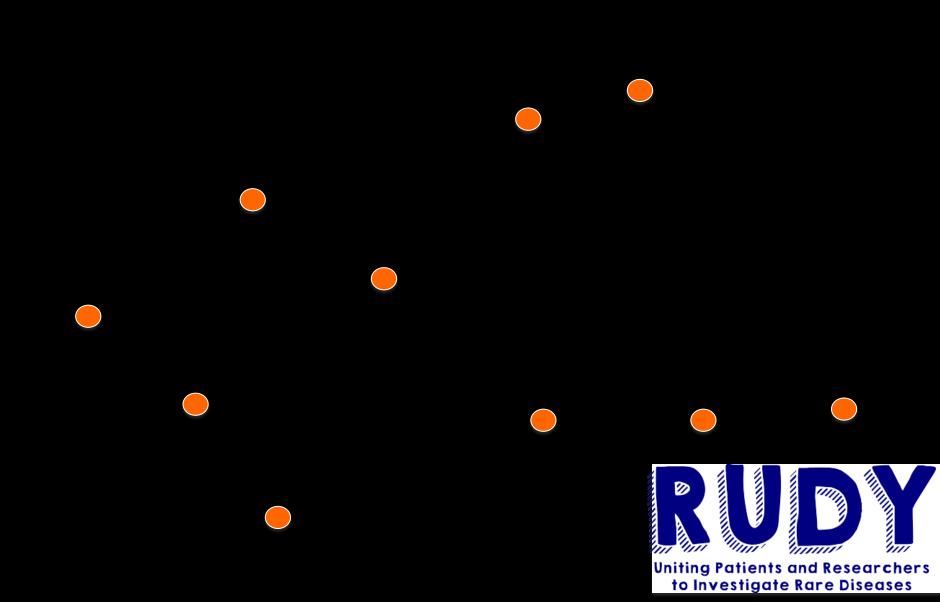


Rare diseases are under researched

Information for patients and doctors is fragmented



HOW?



Identify and recruit

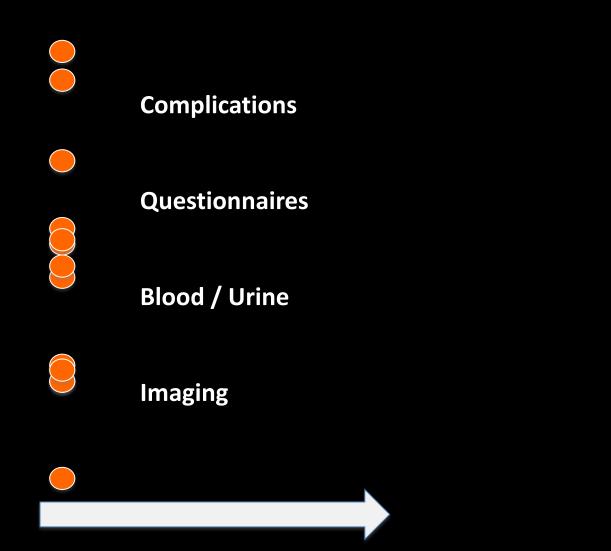


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Uniting Patients and Researchers to Investigate Rare Diseases

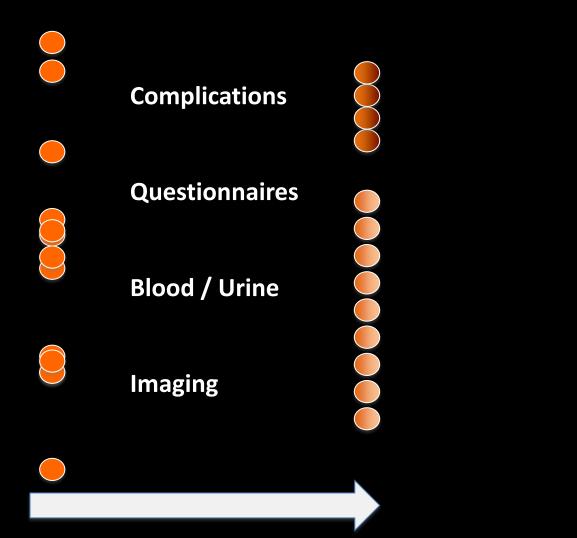
hand

Systematic assessment



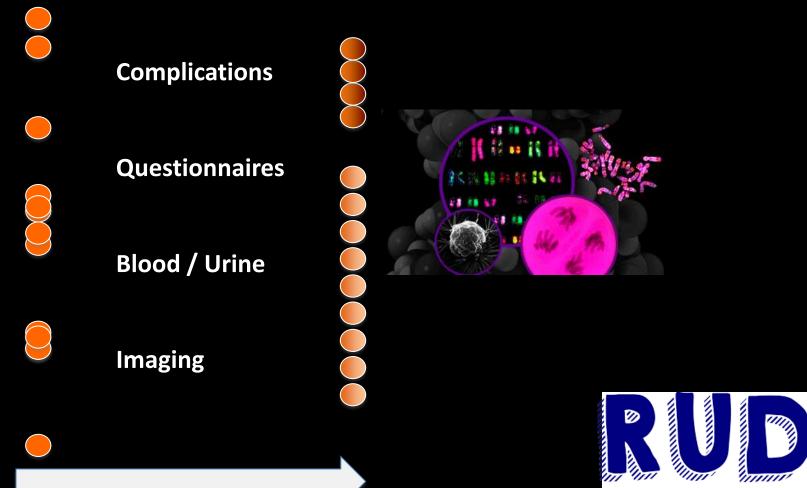


Identify novel subgroups

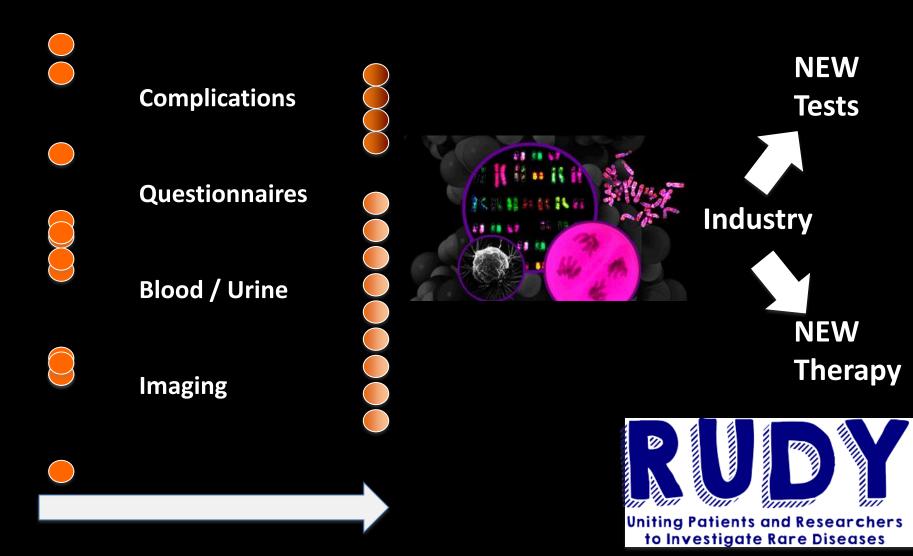


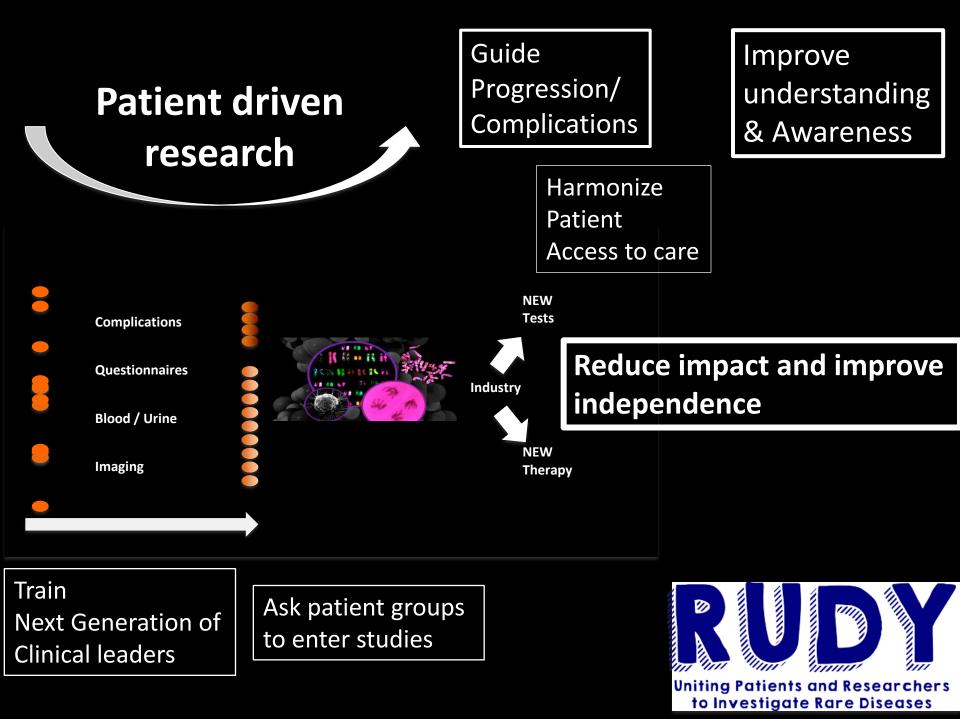
RUDY

Explore biology

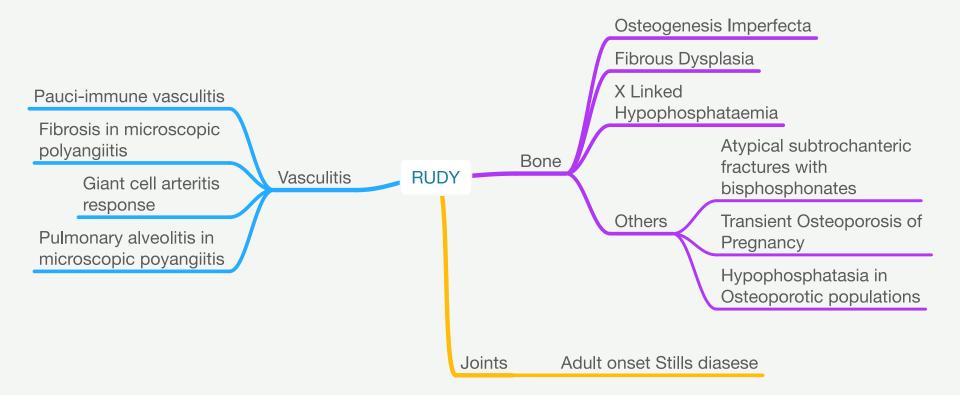


Transform clinical care





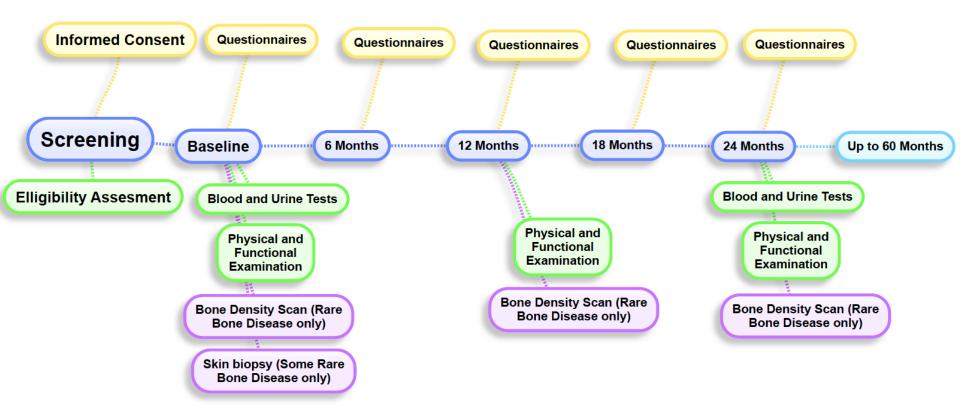
Rudy disease scope:

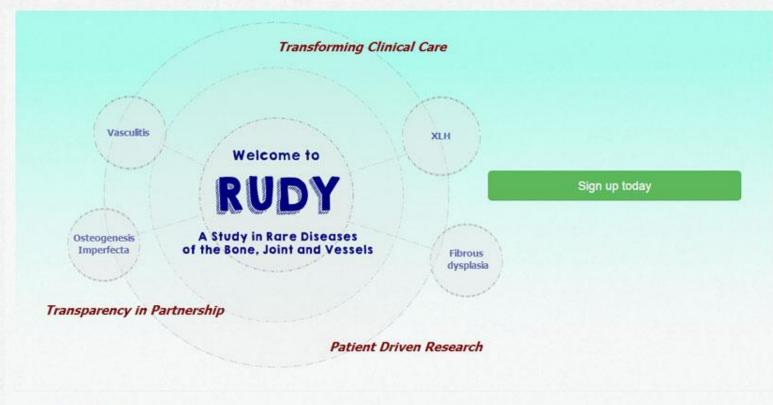


+ More added



Study Schedule





What is Rudy?

Rudy is a study in Rare diseases of the bones, joints and blood vessels. Headed up by a research team at the University of Oxford, Rudy aims to transform clinical care for participants through patient driven research.

Which rare diseases are we interested in?

At present we are prioritizing rare diseases affecting the bone, joints and/or blood vessels (also known as vasculitis). The number of diseases we are interested in is growing as more researchers join the network. If your rare disease is not currently included in this list, please register on the website anyway. We will email you when we have a plan in place for participants with your rare disease.

Who are we?

We are a national network of doctors, researchers, patients and families working to together to improve our understanding of rare diseases and developing new tests and treatments.

Who funds us?

The NIHR Rare diseases of Bone, Joint and blood vessels is funded by a partnership between the NIHR Rare Diseases Translational Research Collaboration and the Oxford NIHR Musculoskeletal Biomedical Research

How can I find out more?

We have a lot of information about Rudy that answers most of the questions we have. If you don't find an answer then please use the comment box and send us an email or call us on xxx xxxx xxx and we will get back to









Two-way conversation

Two-way conversation

- Your information
- Your experiences
- Your choices

• Your samples / data

- Your disease record
 - Updates on RUDY



- Feedback on the websiteComments on the project
- Patient forum / governance

Consent in the RUDY project

Consent in the RUDY project

- Opportunity to change your mind
- Choose how much you want to be involved:
 - What information you want to provide
 - Which parts of the project you contribute to
 - How often you are contacted
 - Who can see and use the information you provide

* My Consent

Events

I agree to provide information about *previous* events and consequences using my secure personal profile on the RUDY website and for this information to be made available to the RUDY research team

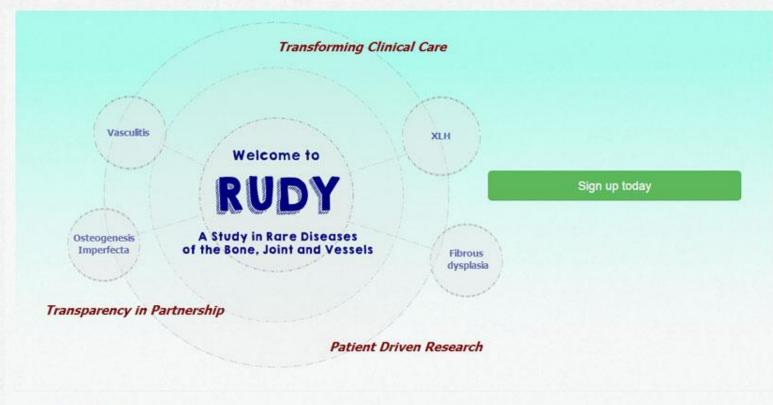
I agree to provide information about current and future events and consequences using my secure personal profile on the RUDY website and for this information to be made available to the RUDY research team

Records

I agree that my medical records held at previous NHS hospitals, GP surgeries and local authorities can be made available to researchers as these are relevant for this study.

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0	Samples	
	I agree that any surplus tissue that is removed as part of my medical care can be used by the researchers as part of this study.	××
	I agree that blood and urine samples can be taken for use in this research and understand that there will not be any direct participant commercial benefit from this.	<××
	I agree that the research team can contact me to arrange appointments to visit my nearest research facility for blood, urine and height tests every two years until the study is completed.	<x<< td=""></x<<>
	I agree that the research team can contact me to arrange appointments to visit my nearest research facility for bone density tests every two years and that these results will be made available to me as well as my current GP and hospital doctor, if I have one.	√ ×
	I agree to have punch skin biopsies, and for the skin samples collected to be used	



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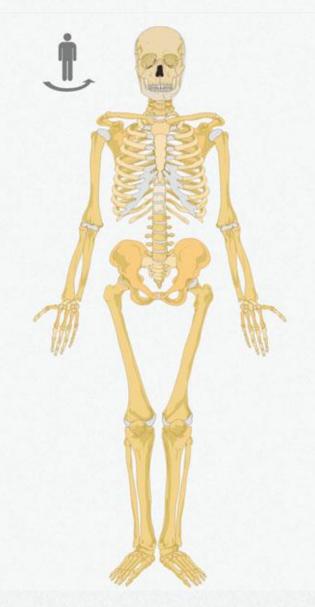
Events

Ever broken a bone?

Enter details of any fractures here

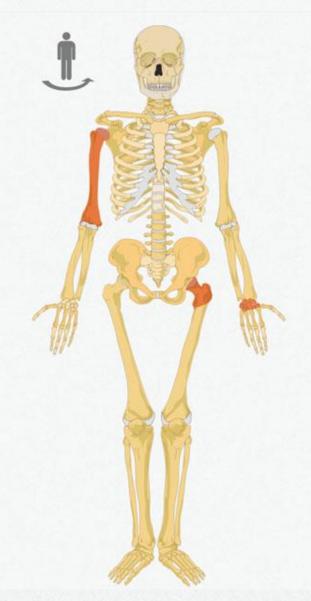
Add a fracture event

L Fracture Event:



Fracture Details	
Date of Fracture	
	ш
evel of Trauma	
Trauma Leve	el 1. e.g. Spontaneous
Trauma Leve	el 1. e.g. Spontaneous Search

L Fracture Event:

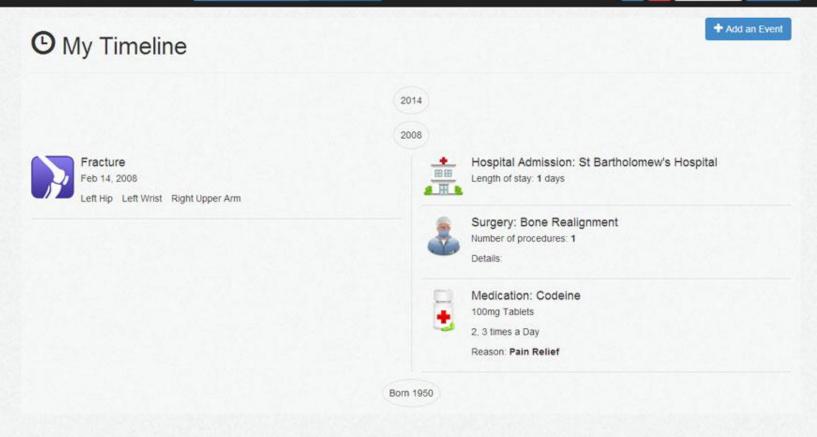


Date of Fracture	e					
14/02/2008	-					
evel of Trauma						
			1			
		Tr	auma	i Level 3' e g. inter	mediate	
ractured Bones		Tr	auma	i Level 3: e.g. inter	Upper Arm	
ractured Bones		Tri	auma	i Level 3: e.g. inter		
	1	Tri 2	auma 3	Level 3: e.g. inter		×
Left Hip		2				
Left Hip Fracture Count:		2				
Left Hip Fracture Count:	irpal:	2 5) 2	3	More than 3 More than 3		×

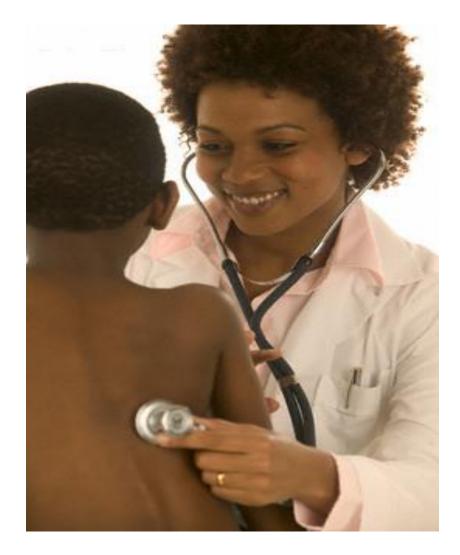
Treatments

What treatment did you receive for the Fracture?

Were you		Admitted to hospital	Treated as outpatient	Treated by GP	Treated at home	
Surgery						
You haven'	added any surgeries f	or this event				
						Add Surgery
Medication						
Name	Strength	Formulation	Dose	Times	Frequency	Reason
You haven't a	added any medications	for this event				
						Add a Medication
Physiotherapy						
You haven	added any physiother	apies for this event				
						Add Physiotherapy







To benefit research and clinical care

Summary

- How research is changing
- ELSI challenges
- Role of patients
- Example project RUDY

Acknowledgements

- Making Connections and ELSI
 2.0 collaborators
- EnCoRe Project members and Team at HeLEX
- RUDY project

- Oxford Musculoskeletal biobank
- Oxford Sarcoma Group
- Oxford Biobank
- OxPaRRM



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