



**Genetic Alliance UK**  
Supporting. Campaigning. Uniting.



# Patient Involvement and Ethics in Rare Disease Research

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# Genetic Alliance UK

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- 180+ patient organisations
- Across the spectrum of genetic disease
- Intractable, incurable, progressive, lethal
- Significant unmet need – a public health challenge
- Research the key to progress
- Translation essential



# The importance of IRDiRC

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- Rare diseases not uncommon
- Create the critical mass
  - of patients
  - of researchers
  - for commercial engagement
  - for planning and delivering services
  - for sustainability
- Build public and patient confidence
- Generate resources for discovery and delivery



# The contribution of patients and families

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- Insight into natural history
- Prioritisation of symptoms and impacts
- Real world data
  
- Advocacy
- Engagement, recruitment and retention
- Urgency and a sense of purpose



# Key Issues – making things happen

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- An appropriate and proportionate ethical framework
  - Honesty – why do you want to do this and what do you hope to discover
  - A realistic time frame
  - Consent – broad, narrow, time-limited, open ended?
  - Re-contact policy
  - Access – who, where, for what purpose
  - Commercialisation
  - Transitions
  - Withdrawal



# Regulations that work

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- **Benefits and risks**
  - The end user perspective
- **The organisation of projects and trials**
  - Make research fit patients' lives, don't expect patients to fit their lives to the research
- **Data Protection**
  - Privacy not necessarily a trump card
- **Licensing**
  - Follow biology
- **HTA**
  - What things mean, not just what can be measured



# History

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Science



Industry



Medicine



Patients



# The Way Forward

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Science

Industry

**PARTNERSHIP**

Medicine

Patients



# Genomics England

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- 100,000 NHS patients
- Rare Diseases, Cancer and Infectious diseases
- For research and health care
- Patients with rare diseases
  - Are very supportive
  - Want to see maximum progress
  - Trust governance arrangements
  - Feel ownership of their genome
  - Wish to have the choice about unrelated findings
  - Are concerned about the capacity for change in the NHS
  - Worry about non-health and care related mission creep



# A sustainable framework for progress

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- Robust
- Rational
- Comprehensive
- Comprehensible
- Honest
- Transparent
- Reviewable
- Appealable
- Policed
- Enforced



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Thank you for listening

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