



# Rare Disease Patients & Genome editing: Perspectives and Engagement

13 November 2017, Paris

**EURORDIS.ORG**

# EURORDIS WORKING GROUP GENOME EDITING

- Patient experts, geneticist, bioethicists and social scientist

**Daniel Renault**, FEDERG and PAC member of RD-Connect

**Joseph Irwin**, SMA Support group and PAC member of RD-Connect

**Chris Sotirelis**, UK Thalassaemia Society and PAC member of RD-Connect

**Françoise Rouault**, AFM and PAC member of RD-Connect

**Elizabeth Vroom**, Dutch Duchenne Parent Project and PAC member of RD-Connect

**Oliver Timmis**, AKU Society and PAC member of RD-Connect

**Rainald von Gizicky**, Retina International and (new) PAC member of RD-Connect

**Pauline McCormack**, PEALS Newcastle University and chair of the PEC of RD-Connect

**Rick Thompson**, Scientific Director at Findacure

**Michele Lipucci di Paola**, AVLTTalassaemia Italy

**Anne-Sophie Lapointe**, Vaincre les maladies lysosomales and EURORDIS board member

**Françoise Salama**, AFM and EURORDIS board member

**John Dart**, DEBRA International and EURORDIS board member

**Christine Grabowski**, Morbus Osler Selbsthilfe e.V. Germany

**Rebecca Dimond**, Cardiff School of Social Sciences

# EURORDIS initiatives to engage RD patients:

- Informal Discussion ECRD with a group of patient experts, Edinburgh May 2016
- Introduction on Genome editing (fact sheet) September 2016
- Genome Editing Webinar October 2016 (Scientific and Ethical aspects)
- Genome Editing Face to Face Workshop November 2016
- Dissemination of key publications and relevant communication to a wide group of interested patient representatives and organisations

# Genome editing Webinar :

- **Professor Fulvio Mavilio, Scientific Director of Généthon, France**
  - therapeutic potentials for genetic diseases: The hype and the reality using a rare disease as an illustrative example.
- **Dr Heidi Howard, biologist and bioethicist at Uppsala University, Sweden**
  - ethical, legal and social considerations relating to the use of genome editing in clinical application.
- Around 50 participants

# Genome editing Workshop: Council of European Federations

## SESSION 1: WHAT IS GENOME EDITING? THE SCIENCE AND BEYOND

Chair: Dr Rick Thompson, Findacure

Gene therapy and genome editing technologies for the study and potential treatment of:

- Hematological genetic disorders, **Dr Annarita Miccio, Imagine Institute, Paris**
- Duchenne Muscular Dystrophy, **Dr France Piétri-Rouxel, Institut de Myologie, Paris**
- Retinitis Pigmentosa by **Professor Dominik Fisher, University Eyeclinics, Tuebingen**

Genome editing research using stem cells, embryos and germline: opportunities and challenges, **Professor Anna Veiga, Centre for Regenerative Medicine, Barcelona**

## SESSION 2: Ethical, Legal and Social Issues

Chair: Dr Pauline McCormack, PEALS Newcastle

- The ethics of genome editing: a very short history  
**Dr Anneliena Bredenoord, Medical Center Utrecht**
  
- The legal framework for genome editing  
**Dr Ilke Turkmendag, Newcastle University**

## SESSION 3: PERSPECTIVES OF THE BIOTECH SECTOR AND REGULATORY ASPECTS

- Spark Therapeutics, **Dr Siobhan Gaynor**
- Intellia Therapeutics, **Dr Sean Burns**
- Vertex, **Dr Andrew Fadden**
- The regulatory landscape, **Dr Nicolas Ferry, Former member of the Committee for Advanced Therapy, European Medicine Agency**

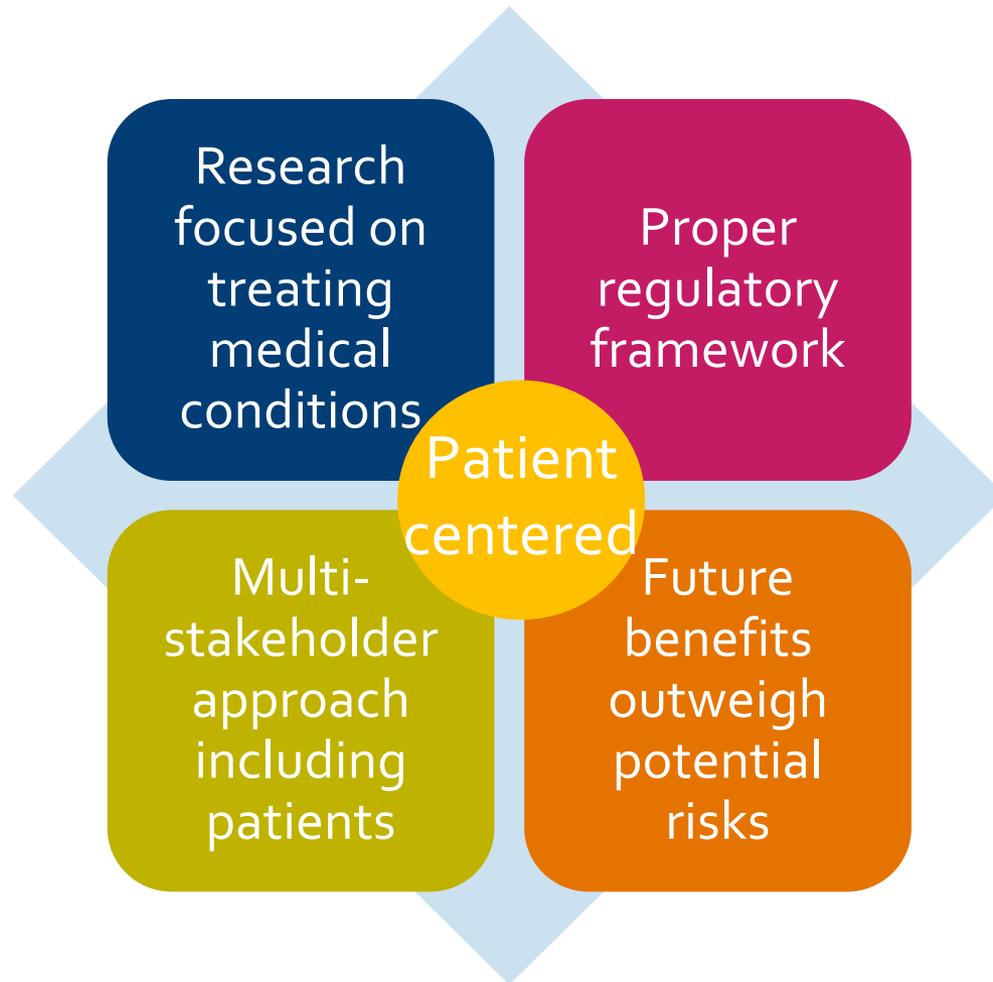
# Genetic Alliance UK initiatives

- [Genome editing: the patient perspective](#) - a report by Genetic Alliance UK on what their members think about the technology (Nov 2016)
- [Response](#) to Genomics and Genome Editing [Inquiry](#) - House of Commons Science and Technology Committee, (Feb 2017)
- [Talking about genome editing](#) - guide on what we should and shouldn't focus on when discussing genome editing (Sept 2017)
- Follow up to Genomics and Genome Editing [Inquiry](#) - House of Commons Science and Technology Committee, (Oct 2017)

# Genetic Alliance UK initiatives

- [Genome editing: the patient perspective](#) - a report by Genetic Alliance UK on what their members think about the technology (Nov 2016)
  - Asked over 150 patients, family members and carers to consider a variety of scenarios in which [genome editing](#) could be applied and give their perspectives on the ethical use and regulation of genome editing technologies in these situations.
  - Funded by EU FP7 grant

# RD Patients strongly encourage responsible research using genome editing:



# Genetic Alliance UK initiatives

- [Talking about genome editing](#) - guide on what we should and shouldn't focus on when discussing genome editing (Sept 2017) jointly with Progress Educational Trust
- The project aimed to explain genome editing to patients, parents and carers affected by genetic conditions to enable them to discuss genome editing and participate in future discussions
- It also includes 8 key recommendations, and some guidance for scientists to explain genome editing in public.
- The report recommends a focus on genome editing rather than its tools or techniques. The focus should instead be on the purpose or potential use of genome editing.
- It is also crucial to be clear about the scope of individual uses of genome editing. distinguish clearly between uses that benefit human health and other purposes (agriculture). Is genome editing being used as tool for research, or will it be part of a therapy? Would changes in the law be necessary to use genome editing in this way?
- Supported by the Wellcome Trust

# Other communication from RD Patient organisations

- AFM Telethon France: recent communication (Oct) to OPECST (committee set up to inform Parliament of scientific and technological options): clear position supporting research progress using genome editing technology but again within a tightly regulated methodological and ethical framework
- Too many uncertainties and not enough knowledge about all the consequences (German Alliance for RD ACHSE).
- Others have said that because lack of knowledge on scientific research, they have not made a statement on behalf of their organisations- highlighting the need for more accessible capacity building on genome editing

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