Rare Diseases, European and UK approaches: setting the scene

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Bearing in mind...

- Rare disease 1:2000
- Very rare disease 1:100,000

In Paediatric Gastroenterology, Hepatology and Nutrition, most disease are “rare”
Why view rare diseases differently?

• Collectively, large numbers of cases of rare diseases occur (3.5 million in UK at some point in their lives)

• Advances in treatment clearly map to coordinated research

• Common themes between rare disease
European & UK initiatives

• European Commission Communication 2008. Rare Diseases : Europe’s Challenge
• Adopted unanimously by each member state June 2009
• Member States to adopt plans or strategies by 2013
• EURORDIS; summer school began July 2008, now annual, July 11-15 2016, Barcelona
  www.eurodis.org
Recommendations joint RDUK/AMRC workshop Dec 2010: Recommendation 1

- Develop clinical research networks for rare disorders
- Established research networks should support rare disease research
- Existing networks should help develop new networks
UK initiatives: patient initiatives

• RDUK November 2008 (www.raredisease.org.uk)

• James Lind Alliance (www.lindalliance.org) Aims to facilitate patient led research; rare disease initiative, bone disease, inherited anaemias, and uncertainties common to all areas.

• Good example for congenital anaemias: www.togetherwecan.uk/basic/inherited.php
UK initiatives: public/industry funded schemes

• UK Rare disease forum (5th meeting 19 June 2015)
• DoH, NHS England, PH England, INVOLVE, Genetic Alliance, Assn BPI, Reps from Wales & N Ireland
• Personal Care Plans, support specialist centres, improve education and training, promote UK as world leader in R&D in the field
• A sense of momentum!
UK initiatives: public/industry funded schemes

• NIHR sponsored Rare Disease initiative: Paediatrics cross cutting theme
  http://rd.trc.nihr.ac.uk

• Renewal bid in progress

• Industry Collaborative Call, EOI open

• Pfizer Rare Disease Consortium
Examples of good Practice: Treat-NMD network, established 2007, 5 year EU funding grant

- Produced and disseminated care standards
- Registries of patients, outcome measures
- Communications infrastructure
- Network of care and trial sites
- DNA cell and tissue biobanks

Has had a direct beneficial impact upon patients with diseases such as Duchenne’s muscular dystrophy and spinal muscular atrophy
Emerging possibilities

• Paed E-Bans

• National (UK) registry of children with intestinal failure, launched April 2015

• Complete ascertainment of all cases of Paediatric Intestinal Failure in UK
Main research message, European & UK Rare Disease Initiatives over past decade

European clinical networks are key to informing and developing research into rare diseases