Rare Diseases, European and UK approaches: setting the scene

Dr Julian Thomas
Great North Children's Hospital
Newcastle UK

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 uncertainities 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

PEDDCReN

Paediatric European Digestive Diseases Clinical Research Network

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

PEDDCReN

Paediatric European Digestive Diseases Clinical Research Network

Main research message, European & UK Rare Disease Initiatives over past decade

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