

**Sharing best practices on integrative approach
to rare diseases in different countries**



United Kingdom

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In collaboration with

EUROPLAN

European Project for Rare Diseases National Plans Development



Strategies for rare diseases

- *No definition of RD – concept is ‘specialised services’*
- *Planning guidance to National Health Service*
- *‘Specialised’ = for populations of 1m – 10m*
- *Highly specialised = for populations of 15m – 50m*
- *Devolution – England, Scotland, Wales, Northern Ireland*

Funding of actions, regarding rare diseases

- *All health care funded by National Health Service*
- *No co-payments**
- *Social care funded by local government*



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Provision of information for rare diseases

- *Website for Specialised Commissioning*
- *'Contact a family' Help Lines*
- *Many disease-specific help lines and web sites*

- *No official list of RDs*



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Research on rare diseases

- *No research programme specifically for RDs*
- *Active contribution to EU programmes e.g.*
 - *Neuromuscular*
 - *Bone*
 - *Mitochondrial*
 - *Dysmorphologies and dysplasias*



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Prevention

- *Folic acid – no fortification of food*
- *Clinical PIGD for some disorders*
- *Routine neonatal screen for*
 - *PKU, hypothyroid*
 - *MCADD, thalassaemia*

National screening committee – www.nsc.nhs.uk



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Empowerment of patients organisations

- *Strong government policy for PPI – patient and public involvement throughout the NHS*
- *Active representation and consultation of patient organisations in highly specialised services*

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Specialised social services

- *Respite Care Services are available throughout UK but vary in extent*
- *Therapeutic Recreational Programmes are also provided often as Occupational Therapy*
- *Services aimed at the integration of patients in daily life*