



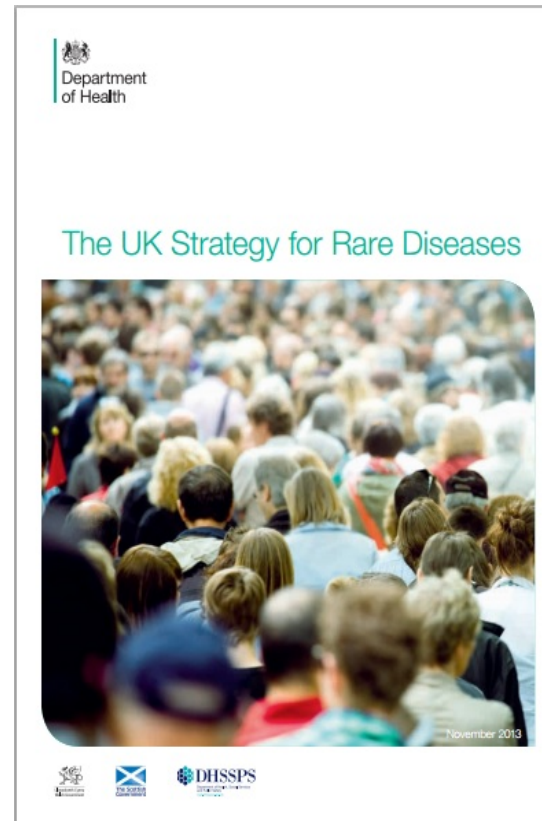
Genetic Alliance UK
Supporting. Campaigning. Uniting.

The UK Strategy for Rare Diseases: what next?

7th March 2015

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Director of Development





About Us



Genetic Alliance UK
Supporting. Campaigning. Uniting.



RARE DISEASE | UK



SWAN UK
syndromes without a name

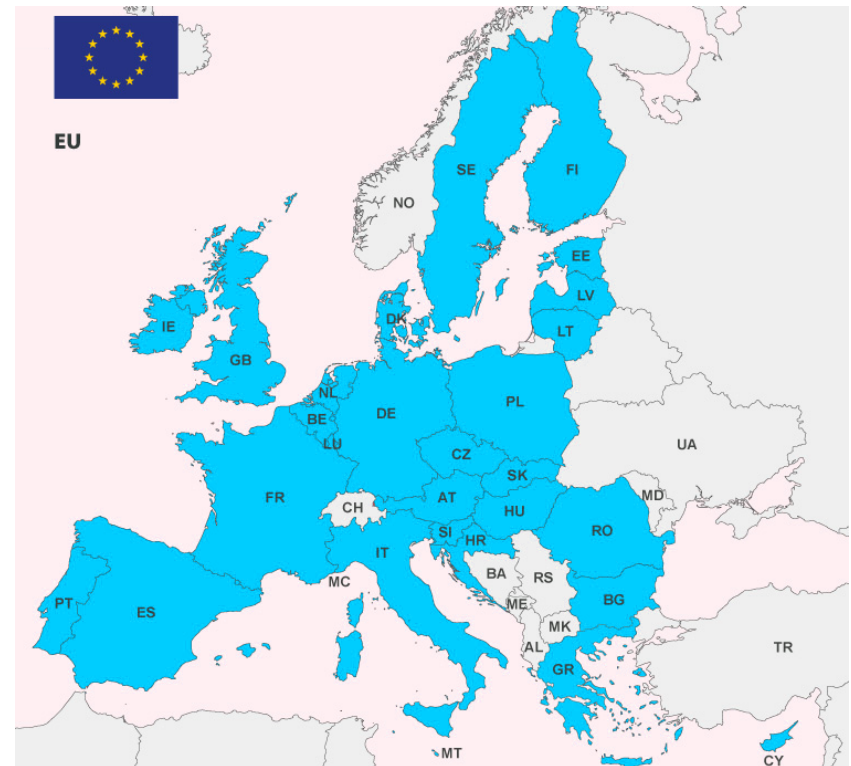
- **Genetic Alliance UK**, an alliance of over 180 patient organisations working to improve the lives of patients and families affected by all types of genetic conditions.
- **Rare Disease UK**, established in Nov 2008, the national multi-stakeholder alliance for people with rare diseases and all who support them. Its membership of over 1,700 includes:
 - Over 260 patient organisations;
 - 17 pharmaceutical companies;
 - Professional bodies, health professionals & researchers;
 - Individual patients and families.
- **Syndromes Without A Name UK (SWAN UK)** provides support and information to families of children with undiagnosed genetic conditions.



Rare Disease UK

History and origin

- A reaction to:
European Commission's Communication on Rare Diseases: Europe's Challenges
11th November 2008.
- Set out proposals for a comprehensive, EU wide, integrated strategy to support Member States on issues including diagnosis, treatment and care for rare disease patients.
- Followed by:
Council of the European Union's Recommendation on an action in the field of rare diseases
8th June 2009.
- Called on all EU member states to develop plans or strategies for rare diseases by 2013 in order to ensure universal access to high quality care.





The UK Strategy: how did we get here?

- EU Recommendation on rare diseases (June 2009) – we ensured that the UK’s health departments acted on their obligation to develop a national plan for rare diseases by the end of 2013.
- An uphill struggle:
There are “no plans to set up a national plan”. “The health and social care needs of people living with rare conditions should be met by local health bodies.”
Dawn Primarolo MP, Minister of State for Public Health
22nd May 2009
- In 2011 we published our landmark document ‘*Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy*’.





A Vision for the Strategy

- **Coordination of research**
Increase collaboration with a focus on rare conditions.
- **Prevention and diagnosis**
Raise awareness of rare disease to reduce delays in diagnosis.
- **Commissioning and planning**
Link commissioning of locally and centrally commissioned services for rare diseases.
- **Patient care, information and support**
Foster the delivery of high quality information to patients on rare diseases.
- **Delivering coordinated care**
Reduce the fragmentation of care provision in the NHS for those affected by rare diseases.





How we do our policy work

- A constructive, evidence based approach built on partnership and collaboration with our members and policy makers.
- Regular interaction with the Health Science and Bioethics Team, DH.
- Working closely with NHS England:
 - through the Rare Disease Advisory Group, Specialised Commissioning Oversight Group and Clinical Reference Groups.
 - Fiona Marley spoke at Rare Disease Day, Richard Jeavons spoke at Rare Disease Day AGM.
- Working with policy makers in the devolved nations:
 - The Northern Ireland Rare Disease Partnership, Scottish Parliament's Rare Diseases Cross Party Group and with the Welsh Health Minister's working group.
- Collaborating with NIHR to ensure effective implementation of the approx. 20 research commitments in the UK Strategy.



Supporting patient voice

Endorsed by Direct contributors

Direct contributors: Action Duchenne, Alström Syndrome UK, AT society, British Porphyria Association, Cystic Fibrosis UK Focus, Duchenne Now, Ehlers-Danlos Support UK, Familial Alzheimer's Disease Support Group, Frontotemporal Dementia Support Group, Funny Lumps, The Fragile X Society, Gauchers Association, MPS Society, Muscular Dystrophy Campaign, Niamann-Pick Disease Group (UK), Genetic Disorders UK, GIST Support UK, HAE UK, Huntington's Disease Association, Hypermobility UK, PNH Alliance, PKD charity, PID UK, Spinal Muscular Atrophy Support UK, Ichthyosis Support Group, International Brain Tumour Alliance, International Castella Syndrome Support Group, Jewish Genetic Disorders UK, Joining Jack, UK PIPS, UK Thalassemia Society, Lily Foundation, Lipoprotein Lipase Deficiency Community, Marfan Association UK, Max Appeal.

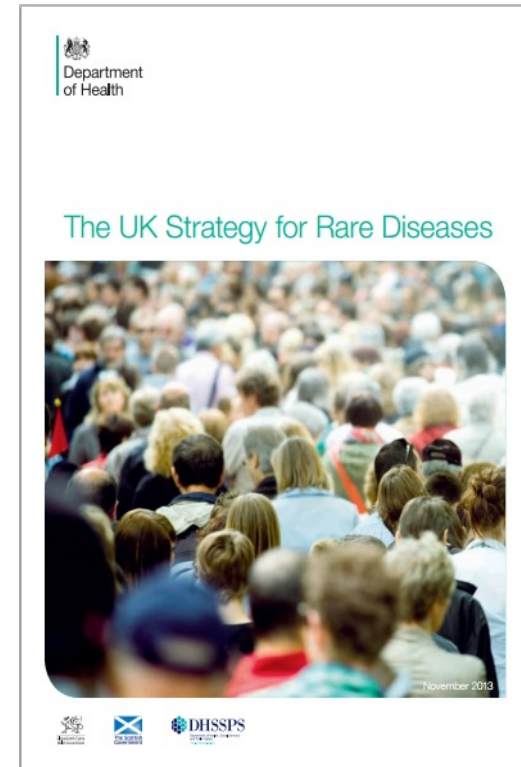
Other supporters

Other supporters: Action for Sick Children, ALDLIFE, AKU, Alpha-1 Awareness UK, Androgen Insensitivity Syndrome Support Group (AISSG), Medical Advisory Service, mnda, myrovlytis trust, Nairnbabies, NEMALINE MYOPATHY Support Group, Aniridia Network UK, Anorchidism Support Group, ARC, ANTHONY NOLAN, AGSD, PAWS-GIST, PWSA, Prader-Willi Syndrome Association UK, PSC Support, PXE Support Group, Reverse Rat, AMEND, Ataxia UK, Barth Syndrome, BDFEA, Bhatnagar Syndrome Society, RP Fighting Blindness, PWSA, Prader-Willi Syndrome Association UK, PSC Support, PXE Support Group, Reverse Rat, British Heart Foundation, Brittle Bone Society, CMA, Cavernoma Alliance UK, CGD Society, Sparks, The Children's Mitochondrial Disease Network, The Children's Mitochondrial Disease Network, THE HAEMOPHILIA SOCIETY, The Haemophilia Society, NSPKU, The National Society for Phenylketonuria, Thyroid UK, Lung Foundation, CILD Lung Foundation, Ciliopathy Alliance, CMT United Kingdom, eah, debra, TSSS, Vasculitis UK, Wilson's Disease Support Group UK, XLP Research Trust.



What's in the UK Strategy?

- UK Strategy published in November 2013
- Sets out 51 commitments across 5 areas:
 - empowering those affected by rare diseases;
 - identifying and preventing rare diseases;
 - diagnosis and early intervention;
 - coordination of care;
 - the role of research.





Things that are good...

‘...no one gets left behind just because they have a rare disease.
We want to put the patients’ needs first.’

- The 51 commitments identify who is responsible for delivering them and the timeframe for doing so.
- The first time the four UK governments worked together to produce a single comprehensive strategy for rare diseases.
- UK Rare Disease Forum to monitor how the actions outlined in the strategy are being responded to across the UK.
 - Will report progress on implementation to the health ministers of the 4 UK countries every two years.
 - Chaired by Alastair Kent OBE, Chair of Rare Disease UK and Director of Genetic Alliance UK.
- A line in the sand...



Progress: England

- ‘Statement of Intent’ published February 2014
- NHS England Taskforce - no further action taken on delivery of healthcare commitments.
- 5 Year Forward View (October 2014) refers to UK Strategy once.
- Implementation on healthcare commitments has completely stalled. Risk that implementation will not take place.
- NHS England is moving forward, slowly, on commitments related to research, data and information, and screening.





Progress: the wider UK

Scotland

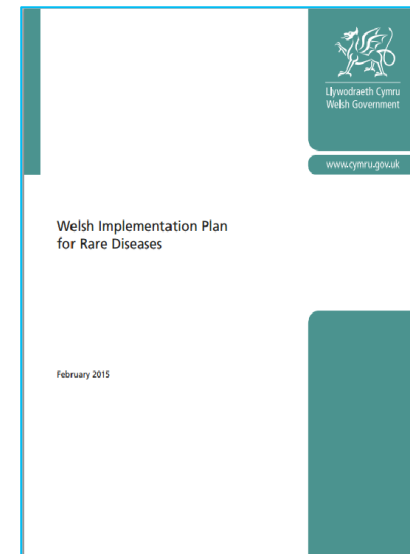
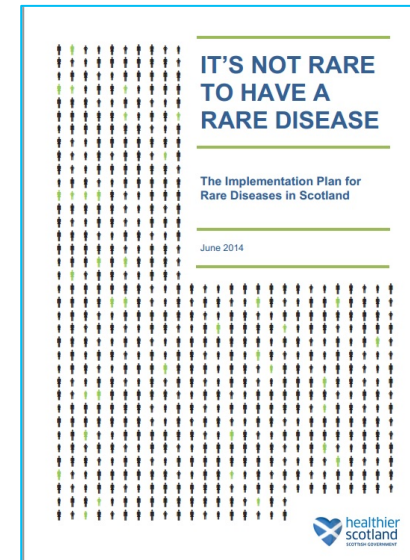
- Implementation plan published July 2014.
- Timescales and clear, deliverable actions are missing but an oversight group has been established.

Northern Ireland

- Consultation launched November 2014.
- Developed collaboratively through a Stakeholder Group chaired by Department of Health, Social Services and Public Safety.

Wales

- Success! Implementation Plan published February 2015. Contains timelines and a proper framework – a cause for celebration.
- Implementation Group established. Annual event, open to external stakeholders, will ensure update on progress.





Challenges...

- Competing priorities: targets and finance.
- Complacency and inertia: 'strategy in place? Job done'.
- Morale.
- Change: elections and NHS reorganisation.
- Compassion fatigue: the battle for attention.
- The shock of the new: genome sequencing and new technology.



What next?

- Campaign for the effective implementation of the Strategy across the UK.
- Ensure the patient voice is cohesive and represented in implementation activity through our Patient Empowerment Group.
- Raise awareness of rare diseases through:
 - Rare Disease Day 2015;
 - Contact campaigns to opinion formers;
 - Multiple parliamentary events and public affairs activity; and
 - Undertaking an in-depth study into transition of rare disease patients.





- An opportunity to focus on progress and encourage policy dialogue in advance of the 2015 general election.
- Evening receptions held:
 - Tuesday 24 February - Scottish Parliament
 - Wednesday 25 February - Houses of Parliament
 - Tuesday 3 March - Welsh Senedd





What can we all do?



- ‘Holding feet to the fire’: ongoing scrutiny to maintain commitments.
- General election – pledge card for prospective MPs.
- Developing a petition to drive action in NHS England.
- Patient empowerment group – contact us to get involved or find out more.
- Getting involved with RDUK (which is free to patients and patient groups).



Genetic Alliance UK

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Thank you!

Rare Disease UK – www.raredisease.org.uk
Genetic Alliance UK – www.geneticalliance.org.uk
SWAN UK - www.undiagnosed.org.uk

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UK GENETIC DISORDERS

LEADERSHIP SYMPOSIUM

March 6-7, 2015

Presented by Genetic Disorders UK
in partnership with Global Genes – Allies in Rare Disease

[#GDLS2015](#)