



'Improving Lives, Optimising Resources: A Vision for the UK Rare Disease Strategy' – Summary of the report

Coordination of Research

Current situation

Research into rare diseases is currently hindered by limited funding provision by research funding bodies, limited collaboration between researchers both nationally and internationally, a lack of basic epidemiological data on rare diseases, and the burdensome bureaucracy involved in obtaining R&D approval for proposed research projects. There are difficulties in the development of diagnostic tests for rare diseases, as well as in therapeutic and prevention research and research into effective management of rare diseases.

Recommendations

Recommendations to improve the current rare disease research situation include:

- Mechanisms to improve collaboration between researchers, including the development of clinical research networks.
- Encouraging funding bodies to support rare disease research, for example by promotion of awareness of the issue of rare diseases to large funding bodies.
- Provision of support to develop effective data collection methods such as patient registries and surveillance units.
- Appraisal and streamlining of the research and development (R&D) approval system.
- Measures to encourage the development and approval of diagnostic tests for rare diseases, including the implementation of a coordinated strategic approach to the development of rare genetic tests.
- Recognition of the different methods that have to be used to study rare disease patients due to the low numbers of affected people, including observational studies.

Prevention and Diagnosis

Current situation

There is currently a lack of professional awareness and identification of rare diseases which often results in delays in diagnosis and misdiagnosis of rare disease patients. This is further compounded by the inequity of access to the diagnostic tests that do exist for rare diseases. Following diagnosis it can be difficult for professionals and patients to access reliable, up-to-date information on the disease and who the specialists are in a particular condition. Rare diseases are also often not considered as suitable for inclusion in newborn screening programmes which can again result in delayed access to necessary services and/or treatment.

Recommendations

Recommendations to improve the prevention and diagnosis of rare diseases include:

Rare Disease UK

Unit 4D, Leroy House, 436 Essex Road,
London, N1 3QP

www.raredisease.org.uk

T: +44 (0)20 7704 3141 | F: +44 (0)20 7359 1447

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)



- Improving professional knowledge and awareness of rare diseases, for example by including a rare diseases module in medical training and providing ongoing education for qualified healthcare professionals.
- Improving linkage between specialist centres and local services to encourage the sharing of knowledge and expertise.
- Improving access to reliable sources of information on rare diseases, including through the development of an online 'portal' to information.
- An appraisal of the current criteria used by the National Screening Committee in assessing conditions for newborn screening programmes.
- Ensuring equitable access to diagnostic tests throughout the UK.
- Improving access to carrier tests for at-risk individuals and groups.

Commissioning and Planning

Current situation

There is a wide variation in the health services available for patients with rare diseases throughout the UK, resulting in inequitable levels of care and support dependent on where a patient lives and the condition they have. The 'postcode lottery' still exists whereby some patients are able to access certain drugs or services and others are not, resulting in very different experiences for patients with the same needs.

Recommendations

Recommendations to improve the commissioning and planning of services for rare diseases include:

- Implementing commissioning and planning systems and structures that ensure equitable access to health services and treatments across the UK, including ensuring that commissioning and planning of services is carried out at the appropriate planning population level.
- Coordination between what is commissioned nationally and what is commissioned at a more local level.
- The safeguarding of resources to fund specialised services for rare diseases.
- Assessment of the mechanism by which the value of medicines for rare conditions is appraised for reimbursement on the NHS.

Patient Care, Information and Support

Current situation

Patients with rare diseases too often report not being provided with sufficient information on their medical, social, financial and psychological needs throughout the progression of their condition. Information that is provided is not always patient-centred or in the most appropriate format or at a level that ensures understanding and supports informed decision making. Links between the medical and social aspects of care are often lacking or weak.

Recommendations

Recommendations to improve patient care, information and support include:

Rare Disease UK

Unit 4D, Leroy House, 436 Essex Road,
London, N1 3QP

www.raredisease.org.uk

T: +44 (0)20 7704 3141 | F: +44 (0)20 7359 1447

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)



- Providing patients with ongoing, patient-centred information on their condition and how to manage it. This would include provision of an ‘information prescription’ for newly diagnosed patients detailing the information they should expect to receive, when and from whom.
- Making sure that information is available in various formats and at various levels of scientific and medical understanding to ensure informed decision making.
- Consideration of psychological support for the whole family as an integral part of the care package.
- Ensuring access to social support for those affected by rare diseases.

Delivering Coordinated Care

Current situation

Care for patients with rare diseases is often fragmented and poorly coordinated resulting in patients having to attend multiple different appointments with all the specialists involved in their condition and making it difficult for patients and carers to continue with their normal daily routine. Communication between specialists is frequently lacking, and care is not always patient-centred and considerate of an individual’s needs.

Recommendations

Recommendations to improve the delivery of coordinated care include:

- The development of a programme of designation for centres of excellence for rare diseases in the UK, supported by networks linking into local services.
- Ensuring good communication between all professionals involved in the care of a patient, for example by implementing a ‘hub and spoke’ model of communication between centres of excellence and local services, having regular meetings between all those involved and the development of systems to enable better sharing of information between professionals.
- Ensuring care is patient-centred, for example by running specialists clinics whereby patients are able to see all specialists in one meeting and by introducing Care Coordinators to liaise between patients and specialists and to act as a point of contact for patients.

Rare Disease UK

Unit 4D, Leroy House, 436 Essex Road,
London, N1 3QP

www.raredisease.org.uk

T: +44 (0)20 7704 3141 | F: +44 (0)20 7359 1447

E: info@raredisease.org.uk

An initiative of



Genetic Alliance UK
Supporting. Campaigning. Uniting.

A charity registered in England and Wales (no. 1114195)
in Scotland (no. SC039299).

A company Limited by Guarantee (Number 05772999)