

Forgotten Conditions:

misdiagnosed and
unsupported,
how patients are
being let down.

Mark Weston
Julia Manning
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1 Introduction

This paper is a report on a roundtable held by 2020health on 12 June 2012 in Portcullis House, London, on “Forgotten Conditions”. These are health conditions and diseases that due to their low prevalence mean patients often get neglected or overlooked. The event, hosted by Andrew George MP and chaired by Lord Clement-Jones, brought together 15 health policy-makers, GPs, academics, public relations professionals and representatives of third sector organisations to discuss how to ensure that the rarer conditions that have traditionally been neglected by the National Health Service (NHS) are allocated the investment and attention necessary to ensure prompt diagnosis and appropriate treatment for the many individuals they affect.

The following is a short report of proceedings with the background context for the discussion. A list of participants, the agenda, and a further reading list are provided in appendices.

2 Background

Rare conditions are defined by the European Union (EU) as those which affect fewer than 5 in 10,000 of the population. There are more than 5,000 of them, with about five new conditions described in the medical literature every week.

According to the Rare Disease UK Alliance (RDUK), 1 in 17 people will be affected by a rare condition in their lifetime. A single rare condition might affect up to 30,000 people, although most affect far fewer. Eighty percent of rare conditions have a genetic component, with thirty percent of those with rare diseases not reaching their fifth birthday.

Although today there are 5,000 rare diseases, in the future this number is likely to mushroom. Advances in genomic sequencing will mean that conditions that are currently common will be broken up into many sub-types. When a single disease is subdivided into hundreds or thousands of smaller diseases, as one roundtable participant observed, 'almost everything will be a rare condition.' This will present a major challenge to health services, which rather than focusing resources on a handful of major illnesses will need to develop effective systems for diagnosing and treating an array of conditions that affect only very small numbers of people.

There are some uncommon diseases which are neglected by health policy-makers even though they do not meet the EU's definition of rare conditions. An example which was discussed in depth at the roundtable along with motor neurone disease, psoriatic arthritis and Huntington's disease, was ankylosing spondylitis (AS). AS affects 2-5 adults per 1,000 in the UK, causing sometimes lifelong spinal pain and restriction and limiting affected individuals' ability to work, rest, socialise and exercise. Recent surgical advances and drugs in the shape of TNF alpha blocking agents offer promise in reducing suffering, but access to them is often hampered by delayed or wrong diagnosis, late referral to the right specialists, and uneven rheumatology and drug provision across the UK resulting in a "postcode lottery" as to whether one receives the appropriate treatment. Debbie Cook, Director of the National Ankylosing Spondylitis Society (NASS), pointed out in her presentation to the roundtable that although twice as many people suffer from AS as multiple sclerosis, the latter is by far the better known condition. As an indication of the neglect surrounding AS, she added that the average delay between onset of the disease and correct diagnosis is 8.5 years, although in some cases the delay can be up to 30 years or more. If diagnosis is very late,

she added, the spine can fuse, leading to a sharp reduction in mobility – one patient Cook had met recently reported having to lie on the ground if he ever wanted to see the stars. Decreased mobility can in turn lead to having to leave work and ultimately depression, thereby further reducing quality of life.

NASS has argued that the low profile of AS is at the root of the failure to provide high quality care to all those with the disease. As a consequence, it explained in a recent report, 'people who develop symptoms have no familiarity with the disease; medical professionals confronted by people who may have AS may not consider it as a possible diagnosis; and specialist services are under-developed.' As Debbie Cook reported, many AS patients are 'stuck in primary care' because GPs lack awareness of the condition or are reluctant because of cost considerations to refer patients to a specialist rheumatologist. The latter, as discussed in the next section, are problems that bedevil many forgotten conditions.

3 The problem

In its constitution, as RDUK Executive Director Stephen Nutt observed in his opening presentation to the roundtable, the NHS pledges to use its resources ‘for the benefit of the whole community, and make sure nobody is excluded or left behind.’

At present, however, those with rarer conditions are generally ill served by the health service. Stephen Nutt argued that policy frameworks are often designed to deal only with high priority conditions, while in his talk at the roundtable, Andrew George MP described rare conditions as ‘languishing in the too-difficult-to-deal-with box.’ The Chief Medical Officer (CMO) for England’s 2009 annual report acknowledged that although rarer diseases collectively account for a large part of NHS work, awareness and understanding of them are uneven, and that adult services for rare conditions are not sufficiently widely available. The CMO observed too that misdiagnoses and unnecessary treatments waste NHS resources, a point taken up by Stephen Nutt, who explained that misdiagnosis ultimately means that more expensive treatment interventions are required, and that more NHS time is taken up by having to deal with cases that have needlessly become complicated.

In 2010, RDUK conducted a survey of the experiences of patients with rare conditions. The survey covered people with over 100 different diseases. It found that although there are many examples of excellent service by the NHS, there are several key problems facing patients, including:

- delayed diagnoses and misdiagnoses
- a lack of information provided by the NHS on rare conditions
- a lack of co-ordination of care
- inequalities in treatments offered to patients around the United Kingdom

In concluding his talk, Stephen Nutt asked if those with rarer conditions should be prepared to accept a lower standard of treatment merely because their condition is not shared by many others. He argued that those with rare conditions should not be forgotten but ‘given the same consideration as people with better-known conditions.’ Agreeing, a specialist in AS treatment argued that ‘rarer conditions should not be forgotten conditions.’ There is strong public backing for action to be taken in this area – in an EU survey of 1,200 Britons in 2011, 98% agreed that people with rare conditions need specific NHS support, while in Europe as a whole 90% supported the development of strategies on rare diseases by member states.

In recent years there have been some efforts to rectify the situation. In 2009 the European Council recommended that each European Union state should have in place a strategy for managing rare conditions. The Department of Health is therefore conducting a ‘Consultation on the United Kingdom Plan for Rare Diseases’, whose results are due to be published by the end of 2013. The consultation’s key questions cover the themes of:

- speedy diagnosis and early intervention
- coding and classification
- research
- centres of expertise and networks
- patient information and support

Current reforms to health service delivery provide further openings for change. Specialised services in England will in future fall under the remit of the NHS Commissioning Board (NHSCB). This will in theory ensure that all patients can access equitable high quality services, regardless of where they live and which rare condition they have. The NHSCB will work with local commissioners of care, including social services, to ensure co-ordinated and joined up pathways for those with a rare condition. Working through the newly-established local Health and Wellbeing Boards will be key to this joined-up commissioning.

4 Suggestions for change

Making the case

Attendees at the roundtable agreed that a much more concerted effort is needed to address rarer conditions, but that health policy-makers are not sufficiently apprised of the latter's importance, with lobbying efforts often falling on deaf ears. Debbie Cook reported that the National Institute for Health and Clinical Excellence (NICE) has yet to produce quality standards and clinical guidelines for AS, for example, while other participants bemoaned the fact that the UK only provides screening for five childhood diseases, compared with over thirty in the United States and twenty in many European countries.

Hitherto, lobbying to raise the profile of forgotten conditions has relied on small third sector organisations such as NASS, the Huntington's Disease Association (HDA), and the Motor Neurone Disease Association (MND Association). These are often voluntary or have only very limited funding, so it is difficult for their voice to be heard when it is competing against the much greater capacity – including, often, celebrity endorsements - of groups lobbying for more widespread conditions. Lord Clement-Jones argued that 'lobbying each individual rare condition is unproductive – there is a need to address systemic issues.' He suggested that both advocates and health policy-makers should look at clusters of rarer conditions, grouping together rare musculoskeletal conditions, for example, to push for a broad response that can then be tailored to individual disorders. Andrew George agreed that lobbying on a condition by condition basis is inadequate, noting that the deficiencies of such an approach will be exacerbated as larger diseases are subdivided and the number of rare conditions soars. 'Whether a disease receives attention shouldn't come down to who can shout the loudest,' he said. 'That's no way for a health service to proceed.'

One way the health service currently proceeds is to rely on data. NICE develops its guidelines based on scientific evidence, but some conditions are so rare that it is difficult to access a sufficiently robust sample to develop sound recommendations. NICE often therefore takes a cautious stance, and delays issuing guidelines or sets higher thresholds for diagnosis or treatment than advocates would like. Two participants argued that an increase in the number of UK specialist centres for particular diseases would help tackle this problem, making it easier to pool individuals with rarer conditions and as a consequence to produce a stronger evidence base.

Recommendation 1

Clear pathways to expertise in rare and forgotten conditions are required. Development of shared care pathways, clinical guidance and centrally collated evidence, and recognition from NICE that small cohort of patients should not preclude them from new treatments.

Action: NHSCB to consider how to increase awareness of best practice for those with rarer conditions.

Who would be a GP?

The role of GPs in diagnosing and treating forgotten conditions was discussed extensively at the roundtable.

GPs are often criticised for their lack of knowledge of rarer conditions and their failure to make prompt and accurate diagnoses and prescribe appropriate treatment. The pressures on GPs, however, are intense. When a patient presents with a rare condition, the GP only has a few minutes to decide what that condition is and what to do about it. It is impossible, moreover, for every GP to know about every rarer condition – a GP may only come across one case of motor neurone disease, for example, in his or her career. Expecting them in a few minutes to reach a point at which they can confidently refer a patient to the right specialist may therefore be unrealistic.

There are ways to make life easier for GPs, however. Roundtable participants emphasised the need for medical students not to be trained in every rare condition but to think widely about ranges of possible diagnoses. GPs, as a GP participant at the roundtable argued, 'cannot be trained to be expert diagnosticians, but when they realise something is wrong they should know where to turn to for specialist help.'

Recommendation 2

Training medical students to think broadly about ranges of possible diagnoses.

Action: Review of medical training by GMC and Health Education England in which meeting demands of increase in rare conditions is considered.

4 Suggestions for change

Technology can play a part here – one attendee noted that GPs faced with an unfamiliar condition can type three or four symptoms into Google and be presented with a range of possible conditions that the patient may have. He recommended that Google searches be included in record keeping documents, and suggested that ‘a smart system would on the basis of this range of possible answers tell the GP the tests and local specialised services available to help narrow down the list.’

Recommendation 3

Using technology to help narrow down to a range of possible diagnoses, and developing smart systems that, based on this range, tell GPs the tests and local specialised services available to come to a conclusion.

Action: Promotion of smart diagnostic systems such as www.isabelhealthcare.com, by the Royal Colleges, Department of Health and regulators.

This idea of establishing a range of possible diagnoses was taken up by other roundtable participants, who argued that it would allow patients to become involved in their own diagnosis and treatment. Andrew George explained: ‘If GPs tell patients a range of conditions they might have, this will help patients, who know more about their condition than anyone, to help the clinician with the diagnosis. GPs should be well-informed facilitators.’ Debbie Cook seconded this, and suggested that it should apply to treatment as well as diagnosis. Highlighting the need for health services to provide information to patients so that they can care for themselves, she said that ‘the government must support the self-care agenda.’ Technology offers promise in this area, too. One roundtable participant, noting that some mental health trusts have successfully introduced computer systems for patients’ use, asked, ‘How many GP surgeries have IT points that patients can use to help self-diagnose?’ There was some dispute over whether the responsibility for diagnosing and caring for those with rarer conditions should lie with primary or secondary care practitioners.

Recommendation 4

Primary care (community based) musculoskeletal disorder teams could include a rheumatologist trained to detect rarer MSDs, but treatment may still need to remain with specialist.

Action: Review of MSD services by new Clinical Commissioning Groups to consider the inclusion of a rheumatologist.

Some participants suggested that with better training and awareness, primary care practitioners could take on more responsibility for diagnosis and treatment of rarer conditions, so that patients would not always need to be referred to specialists. One participant argued that ‘we need to build primary care capacity for ongoing care so that the patient stays with the GP rather than with a consultant – we shouldn’t put all the money into specialised services as this would mean less money available for common conditions, elderly care etc.’ He suggested that primary care musculoskeletal disorder teams could include, for example, a rheumatologist trained to pick up rarer MSDs, but treatment may still need to remain with specialist.

Recommendation 5

Engaging patients in self-diagnosis and self-care, with the GP acting as a ‘well-informed facilitator’ along with the governments support for the self-care agenda.

Action: Higher profile given to self-care by Department of Health and GP representatives (RCGP, NAPC, NHS Alliance etc).

4 Suggestions for change

On the other hand, some participants countered that rare conditions are often too complex for primary care to deal with, and that it may be a false economy to keep patients in primary care. Mandy Ledbury of the HDA explained that care for Huntington's disease, for example, is very specialised, while an AS specialist said, 'It's not robbing Peter to pay Paul. Treatment cannot always be done in the community because GPs do not have experience of some of the newer drugs.'

Recent health service reforms mean that GPs will now take on the role of commissioning many of the services patients need. As Lord Clement-Jones pointed out, devolving commissioning to Clinical Commissioning Groups (CCGs) risks rarer conditions slipping through the cracks. 'We need to ensure that the National Commissioning Board takes sufficient account of rarer diseases when developing commissioning guidance for CCGs,' he said.

Recommendation 6

When the National Health Service Commissioning Board develops commissioning guidance for CCGs it must be clear on how CCGs can incorporate the provision for those with rare disease into their planning.

Action: NHSCB

To relieve this additional pressure on GPs, several roundtable participants advocated an increase in the number of specialist centres of excellence for rarer conditions. This would extend the reach of secondary care into areas that have previously placed demands on primary care professionals. A professor who works at one of the UK's two specialist centres for ultra-rare children's diseases acknowledged that it would be impossible to have a specialist commissioning group for every rare condition, but emphasised that establishing groups that address several similar conditions would have benefits in terms of pooling expertise, strengthening knowledge networks, developing shared care pathways and clinical guidance, and providing a more robust evidence base. More of these clusters, he argued, are needed at a national level, so that GPs confronted with a rare condition have somewhere nearby to turn to.

Joined-up working

Integration of service provision was the final major theme discussed at the roundtable. Debbie Cook highlighted the importance of looking at the 'whole person' rather than just at the medical aspects of the problem. She and several other participants stressed the value to individuals and society as a whole of patients staying at or being in work, and complained that this is not a sufficiently high priority for policy-makers, and that NICE does not consider in its assessments these wider societal benefits of medical interventions. Debbie Cook called for 'better integration between health and social care, and between the Department of Health (DoH) and the Department for Work and Pensions (DWP) to help people with forgotten conditions to continue to work and to access social services.'

Such integration can result in significant cost savings. Alasdair McLeish of the MDN Association gave the example of people with motor neurone disease who may be kept in an expensive hospital bed for several days while they wait for social services to deliver reading tools; with better integration, he said, this would be unnecessary. Andrew George added that with more joined-up working between DoH and DWP, the latter could be responsible for prescribing physiotherapy or exercise to AS patients, easing the burden on the DoH and, by helping patients to return to work more quickly, reducing the costs to government and business of lost working days. In her concluding remarks to the roundtable, 2020health's Julia Manning echoed these calls for stronger integration, but bemoaned the demise of the Health, Work and Wellbeing Directorate, which she described as a step in the opposite direction.

Recommendation 7

Greater promotion of self-referral to physiotherapy including by Job Centres and re-establishing the links between DWP and DH to underpin the importance of work for health.

Action: DWP to review access to physiotherapy and with respect to promoting work as a health outcome, the links between the ESA assessment and an individual's GP support.

5 Key themes from the discussion

In conclusion, the following are the key themes and recommendations that emerged from the discussions:

- Rarer conditions are too often neglected by the health service, leading to reduced quality of life for affected individuals and increased costs for the NHS, the benefits system, employers and patients.
- As more rare conditions are discovered, and as common conditions are divided into sub-types, the impacts of deficiencies in the diagnosis and care of rarer diseases will grow. Finding systematic ways to replace the current piecemeal approach to addressing rarer diseases is likely to become an increasingly urgent task for health services. Drawing on best practice from overseas is likely to provide helpful guidance.
- Early diagnosis and prompt access to appropriate treatment are vital for rarer conditions, and save the health service money in the long-term. These rely crucially on increased awareness among health professionals, and better information for the public. They also point to the importance of a continued focus on the value of interventions to the patient and to society, rather than purely on their cost to the NHS.

It is unrealistic to expect GPs to be able during a seven-minute consultation with a patient to diagnose any of the 5,000 rare conditions that has so far been discovered. There are, however, ways to ease the pressure on GPs as detailed in our recommendations.

Better integration of service provision is needed to help those with forgotten conditions. In particular, health and social care services should work more closely together to help people to stay in work or return to work quickly. This can result not just in improved quality of life for the patient but in large cost savings for health and social services and for business.

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6 Summary of Recommendations

Recommendation 1

Clear pathways to expertise in rare and forgotten conditions are required. Development of shared care pathways, clinical guidance and centrally collated evidence, and recognition from NICE that small cohort of patients should not preclude them from new treatments.

Action: NHSCB to consider how to increase awareness of best practice for those with rarer conditions.

Recommendation 2

Training medical students to think broadly about ranges of possible diagnoses.

Action: Review of medical training by GMC and Health Education England in which meeting demands of increase in rare conditions is considered.

Recommendation 3

Using technology to help narrow down to a range of possible diagnoses, and developing smart systems that, based on this range, tell GPs the tests and local specialised services available to come to a conclusion.

Action: Promotion of smart diagnostic systems such as www.isabelhealthcare.com, by the Royal Colleges, Department of Health and regulators.

Recommendation 4

Primary care (community based) musculoskeletal disorder teams could include a rheumatologist trained to detect rarer MSDs, but treatment may still need to remain with specialist.

Action: Review of MSD services by new Clinical Commissioning Groups to consider the inclusion of a rheumatologist.

Recommendation 5

Engaging patients in self-diagnosis and self-care, with the GP acting as a 'well-informed facilitator' along with the governments support for the self-care agenda.

Action: Higher profile given to self-care by Department of Health and GP representatives (RCGP, NAPC, NHS Alliance etc).

Recommendation 6

When the National Health Service Commissioning Board develops commissioning guidance for CCGs it must be clear on how CCGs can incorporate the provision for those with rare disease into their planning.

Action: NHSCB

Recommendation 7

Greater promotion of self-referral to physiotherapy including by Job Centres and re-establishing the links between DWP and DH to underpin the importance of work for health.

Action: DWP to review access to physiotherapy and with respect to promoting work as a health outcome, the links between the ESA assessment and an individual's GP support.

Appendix 1

Meeting Agenda

12.30pm

Lord Clement-Jones:
Welcome and introductions

12.35pm

Mark Weston, 2020health:
Setting the scene

12.40pm

Opening talks:

- **Stephen Nutt, Executive Officer, Rare Disease UK:**
In a resource-constrained policy environment, which conditions or diseases are deserving of an increase in attention and investment, and why?
- **Andrew George MP:** Given a resource-constrained environment, what progress is it realistic to expect to achieve in the coming years? What are the barriers and opportunities presented by the changing healthcare landscape?
- **Debbie Cook, Director, National Ankylosing Spondylitis Society:** Who should be responsible for pressing the case for increased attention and investment? To whom should they be directing their efforts? And how can their case be strengthened so that the importance of the conditions is understood and change implemented?

1.00pm

Plenary discussion of opening talks

1.15pm

Plenary discussion of key themes, namely:

- What impact will the Government's Health reforms have on forgotten conditions and what more do health policy makers need to do to ensure awareness and optimal outcomes for conditions such as ankylosing spondylitis and psoriatic arthritis?
- How can Government and wider healthcare policy makers ensure diagnosis and access to effective treatments is accelerated to enable eligible patients to receive treatment and ultimately better outcomes?
- What steps can Government and wider health policy makers take to ensure recommendations are implemented across the NHS from newer best practice documents such as Outside In and Looking Ahead?

1.55pm

Lord Clement-Jones:
Closing remarks from the Chair

Appendix 2

Roundtable Attendees

Lord Clement-Jones

Andrew George MP

Julia Manning,
2020health

Mark Weston,
2020health

Stephen Nutt,
Executive Director, Rare Diseases UK

Debbie Cook,
Director, National Ankylosing
Spondylitis Society (NASS)

Ross Carroll,
Government Affairs Manager, Abbott
Dr Dennis Cox, GP and Director of Cox Medicolegal Ltd

Barry Day,
Service Improvement Manager LTC, Royal Hospital
for Neuro-disability

Bobby Gaspar,
Professor of Paediatrics and Immunology, Honorary
Consultant in Paediatric Immunology, UCL

Mandy Ledbury,
Huntington's Disease Association
Dr Helena Marzo-Ortega, Leeds University and Leeds
Teaching Hospitals NHS Trust

Christian May,
Account Director, Media Intelligence Partners Ltd

Alasdair McLeish,
Acting Chief Executive,
MND Association

Farah Nazeer,
Director of External Affairs,
MND Association

Carl Packman,
2020health

Appendix 3 Further Reading

Alphabetical list of rare diseases:

http://www.orpha.net/consor/cgi-bin/Disease_Search_List.php?lng=EN

EC Communication on Rare Diseases:

http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf

Department of Health Rare Diseases Plan consultation document:

http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/documents/digitalasset/dh_132883.pdf

Rare Disease UK recommendations for a UK strategy for rare diseases:

http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/@dh/@en/documents/digitalasset/dh_132883.pdf

Experiences of Rare Diseases – 2010 RDUK patient survey:

<http://www.raredisease.org.uk/documents/RDUK-Family-Report.pdf>

NASS: Looking Ahead – Best Practice for the Care of People with Ankylosing Spondylitis:

http://www.nass.co.uk/campaigning/looking-ahead/UK_PsA_Working_Group:_Outside_In_-_Joint_Advances_in_Rheumatology_and_Dermatology:

<http://www.psoriasis-association.org.uk/silo/files/L267%20Outside%20In%20BP%20Document%20G-1.pdf>

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Mark Weston
Julia Manning
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2020health.org
83 Victoria Street
London SW1H 0HW

T 020 3170 7702
E admin@2020health.org

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The Abbott logo, consisting of a blue stylized 'A' shape.
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