Signposts to Change by and for the rare disease community

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Signposts to change

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by and for the rare disease community.

Each signpost directs to a source of information which people indicated as a priority for change within the Northern Ireland health and social care system.

Website/hub where all rare disease information can be accessed or linked.

Training for professionals.

Rare Disease Coordinator to make information and research available.

Public health campaign (incl. social media).

Regular local support meetings.

Strong inter-professional working between specialists & GPs.

Raise awareness and have timely reminders in the press about conditions.

Central register of those with a rare condition.

Research and accurate data gathering on those affected by rare disease.

Individualised care plans/patient-centred approach.

A survey was conducted among individuals living and working with rare diseases to evaluate communication for them within the healthcare system, how individuals access or engage with medical practitioners, and how individuals generally access information about rare diseases. Also included were questions to help prioritise improvement within, and allied with, communication and information processes of the integrated health and social care system in Northern Ireland.

Considering communication on a system level, it is important to be aware that communication can be especially challenging for those who have been diagnosed with a rare condition. For example, isolation is described as an immediate consequence of being diagnosed with a rare disease (Pelentsov et al. 2016). Due to the condition being rare who can empathise with persons living or working with the condition? Who will be able to offer suggestions or experience? And who will have accurate or reputable information? These are questions which are faced by many who are affected by a rare disease. Thus, effective support networks are vital for those living and working with rare conditions and social networks are a significant method by which communication involving rare disease occurs (van Betuw 2010).

Participants of the survey said that **professionals not listening was a serious problem for a person with a rare disease.** The participants wanted 'involvement of patients as experts', 'empathy', and 'a more open mind from consultants when we present information we have found'.

Not being patronised, not having the research they'd done taken seriously, and a lack of empathy have also been shown in the wider research to be a problem for parents who are speaking to geneticists (Anonymous 2015). Patients need to be treated as partners or at least included in decisions around the healthcare options available or the next steps.

The Internet was the most regularly accessed source of information for rare diseases. 64.9% of respondents accessed it regularly, where the next most regularly accessed source of information (charity/support groups) was accessed regularly by only 48.2% of the respondents. The Internet was also the source of information found by most respondents as very easy to access, with 58.0% of respondents finding it very easy, and a further 29.5% respondents finding it manageable. The source found by the second highest number of people to be very easy to access was social media with 34.8% saying that it was very easy, and 25.0% saying it was manageable. Given that the Internet is both the most regularly accessed and the easiest way to source information on rare disease, this highlights the problem faced by people with rare disease who find it considerably harder to access medical professionals, information events and conferences both local and international, and the medical literature, than they do to do a search engine on the Internet. What affect does this have on the **engagement between the patient and the medical professional**, and also on the wellbeing of the patient who is relying on information they are sourcing themselves with no obvious credibility or reliability?

Is there a **pathway to diagnosis** documented? Over a fifth of the responses to the question of what clinical information they would like to have available about rare diseases were about diagnosis. Diagnosis appears to be something that many of the respondents have found difficult. It is possible that there is information available that they are not aware of, or that in fact there is not information available at present about the process or complexities, and timelines involved in getting diagnosis. Neither of these possibilities are ideal. There is also **a lack of information post-diagnosis about the process to come**. This means that patients may receive a letter informing them of a diagnosis, but no information as to what the short, medium, and long term steps within the healthcare system will be.

Further to this many respondents to the survey have found that communication between services in the NHS doesn't happen successfully for rare disease patients. This was not directly addressed in any one question but was commented on in many.

References:

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A.L. Crowe[1], H. McAneney[1], A.J. McKnight[1]

[1] Centre for Public Health, Queen's University Belfast

c/o Regional Genetics Centre, Level A, Tower Block, Belfast City Hospital, Lisburn Road