Making life better: improving health and care for individuals with rare diseases

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Rare Diseases

- Affect at least $^{1}/_{17}$ individuals in NI; ~6% of population
  - Many adults live with >1 rare disease
- Collectively represent a major public health problem
- Worldwide problem; implications for local policy
- No cohesive structure developed specifically to help

Early interventions may substantially improve life

Approach

- Invitations via social media, voluntary groups, online, and direct emails to previous participants
- Experiences on living everyday with rare disease
- Simple online survey with a free text comment box (www.surveymonkey.com), more complex surveys incorporating micro-narratives using SenseMaker® Suite, one-to-one semi-structured interviews, focus groups, and a series of open meetings in 2014/2015

HSC Public Health Agency | Northern Ireland Rare Disease Partnership | HSC Health and Social Care Board | Department of Health, Social Services and Public Safety www.dhsspsni.gov.uk

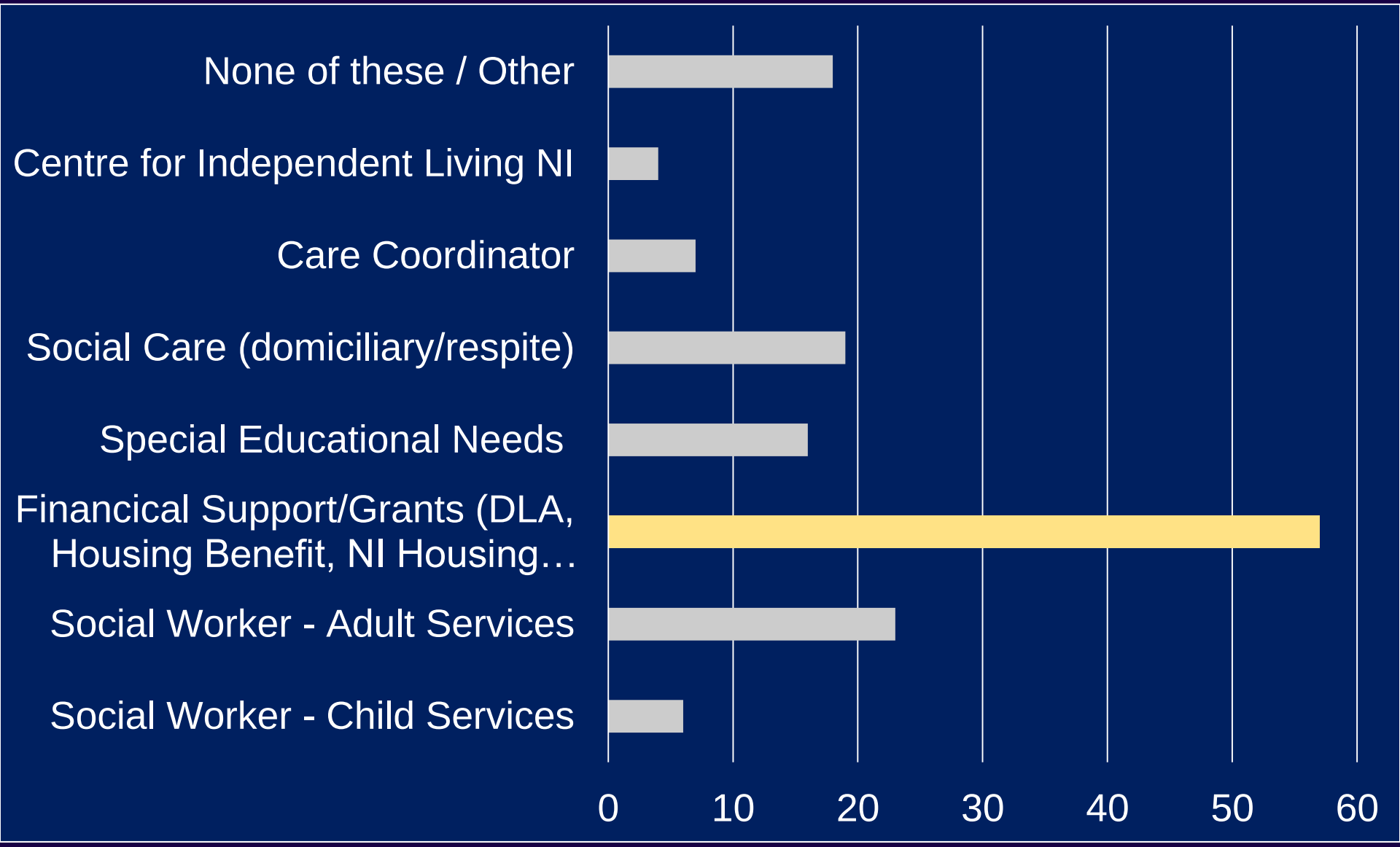
Overview

- Majority of respondents were female, >30 years old
- 56% said accessing expert medical care is difficult
- Respondents attended >40 *vs* 0 expert consultations
- ‘Expert patients’: 36% of individuals felt they were strongly involved in planning their medical care
- UK studies suggest up to 40% costs could be saved with improved preventative care

Important Details

- Most respondents informed by support groups
- >50% of respondents had more than 10 GP/practice nurse appointments in the past year
- ~$^{1}/_{3}$ people wait 1-5 years for accurate diagnosis
  - >$^{1}/_{10}$ wait >10 years; ½ receive ≥ 1 misdiagnosis
- 15% of persons see 5-10 doctors; $^{1}/_{20}$ see >10 doctors
- Individual requirements; not one-size-fits-all

Agency Engagement in Last 5 Years



Respondent Comments

- >½ narratives were self-described as having negative emotional intensity

- Common themes were consistently identified:
  - *'feeling isolated'*
  - difficulty finding details for health and social care contacts
  - challenges accessing information in suitable formats
  - improving diagnosis, treatments, accessing rare drugs
  - gaining information about appropriate care pathways
  - insufficient expertise of local healthcare providers in highly specialised treatment options for rare diseases is a major problem…*this is true worldwide; networks are key*

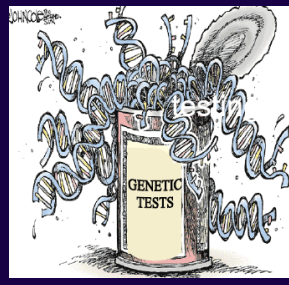
Wordle based on 91 stories from SenseMaker (2014)

Options for Consultation

- UK Strategy for Rare Diseases
- NI statement of intent & public consultation on the NI Rare Diseases Implementation Plan
- Accessibility of information needed by adults living with rare diseases; use of jargon and abbreviations; complex issues; challenging for informed response

Using an appropriate research framework to seek public opinion is essential to minimise inadvertent bias of results

Example of Genetic Testing…



Open meetings: overview & smaller discussion groups may lead to changes in an individual's or group's belief that would change their personal response to an e-consultation / online survey

Each group discussion contained different members and was conducted several times using a different facilitator who recorded results and explained terms on request to enable improved understanding

A collaborative approach that enables discussion

Suggestions

Changing Priorities Ahead ?

- Respondents felt that primarily doctors, allied health professionals, nurses and politicians should be made aware of patient views *in that order*

- Strategies should be developed to:
  - improve the diagnostic process; promote existing resources
  - strengthen collaborative partnerships; whole system framework
  - enhance communication and coordination of care
  - improve data collection & information sharing
  - increase training and network opportunities

Conclusions

- Multiple data collections have provided important information to help identify and prioritise local needs for adults living with rare diseases in NI; patient involvement, baseline data, improved information
- Empower adults by making information readily available and accessible in a variety of formats
- Enable affected individuals and families to maximise health and wellbeing, whilst streamlining health and social care, which may lead to substantial £ savings

Thank You

to everyone who contributed

HSC Public Health Agency

NORTHERN IRELAND RARE DISEASE PARTNERSHIP

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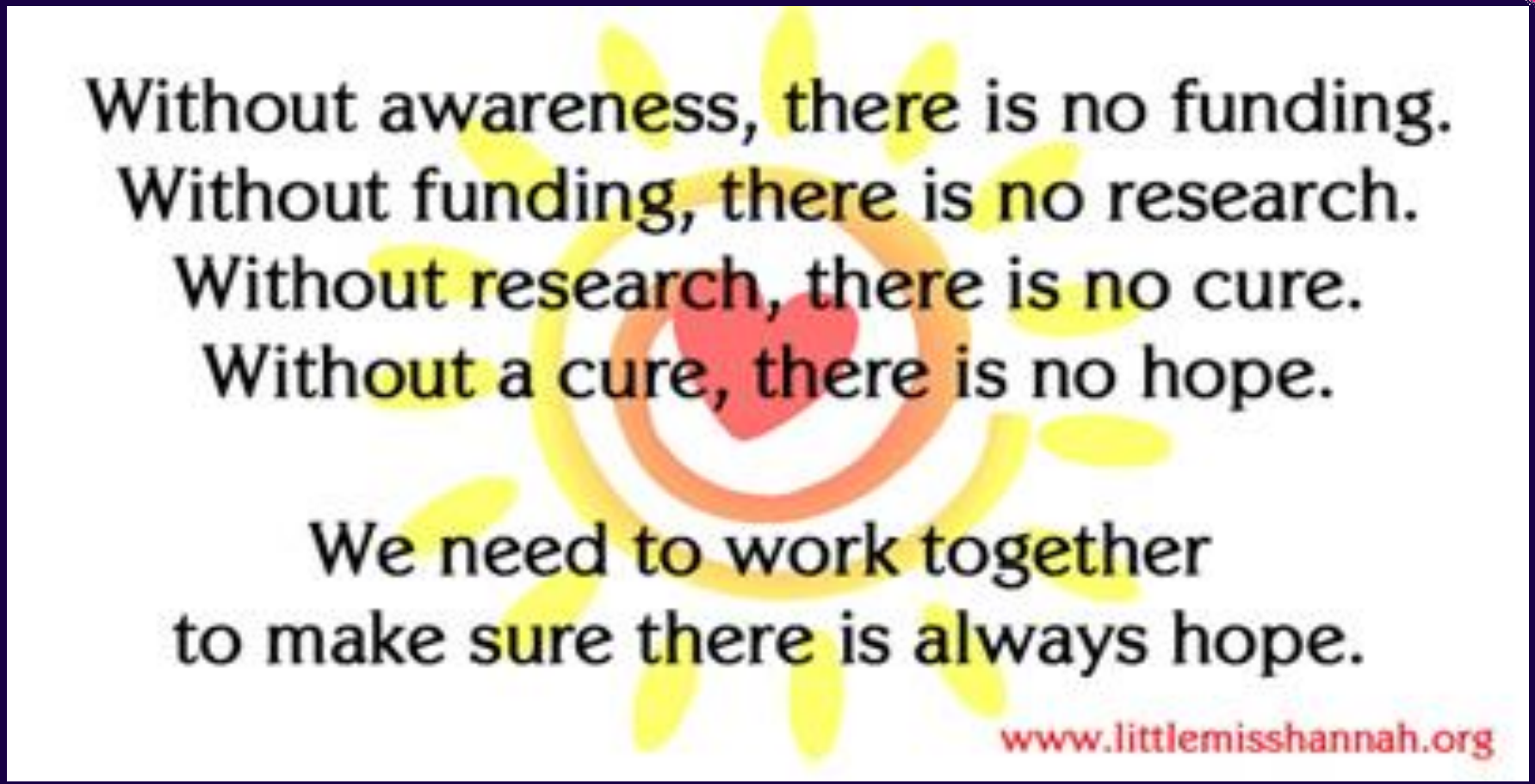

A Vision for a Centre of Expertise for Rare Diseases / Genomic Medicine Centre NI

Rare disease documents for NI include information on a vision for a Centre of Expertise for Rare Diseases in NI....all thoughts and suggestions for practical improvement very welcome.

Specifically helping with:

- Empowering those affected by rare diseases
- Identifying and preventing rare diseases
- Diagnosis and early intervention
- Co-ordination of care
- The role of research

NI has unique potential to develop a rare disease registry and information hub for patients and healthcare professionals. Local expertise and experience in epidemiology, statistics, informatics, rare disease genetics, data management, registries, existing resources, NIECR, NIADRC, and data protection are all readily available.