

The Irish National Rare Diseases Office and Orphanet Ireland: Forging a National Identity for Rare Diseases in a Small Country

DM Lambert, R O'Shea, R Marron, SA Lynch and EP Treacy
National Rare Diseases Office, Mater Misericordiae University Hospital, Dublin 7

NEW NATIONAL RARE DISEASE OFFICE - RESOURCES BRIDGING THE INFORMATION GAP – OUR FIRST YEAR

NATIONAL RARE DISEASE OFFICE

Opened June 2015

Rare Disease Information

- Irish Health Service Executive (HSE) initiative - based at Mater Misericordiae University Hospital
- NRDO provides reliable RD information and expert guidelines in an Irish context
- Information through Website, Free phone information line & Email
- Available to health care professionals and public

Orphanet/Orphanet Ireland

- Joint initiative of European RD-Action and the HSE
- NRDO collates and maintains Irish rare disease resources through Orphanet Ireland
- Irish arm of the largest international rare disease online portal



DISSEMINATION OF RD INFORMATION

- 23 Departmental presentations
- 5 Patient organization presentations
- 1 Hospital grand rounds
- 3 Presentations at scientific meetings
- 4 Posters at scientific meetings
- 10 Leaflet tables / Information booths
- 4 University rare disease lectures
- 1 Medical publication
- 3 Newspaper and other media publications
- 1 National Health Service-hosted webpage
- 1 Rare Disease office launch with Minister of Health
- 1 College of Physicians half day Masterclass

RESPONDING TO RESOURCE GAPS

HSE 'Have Your Say' RD public consultation 2012

- Evidence-based and trustworthy information
- Guidance on how to access specialist services
- Summary of treatments available/clinical trials

RD WIFI Irish patient study, Nicholls et al, 2015

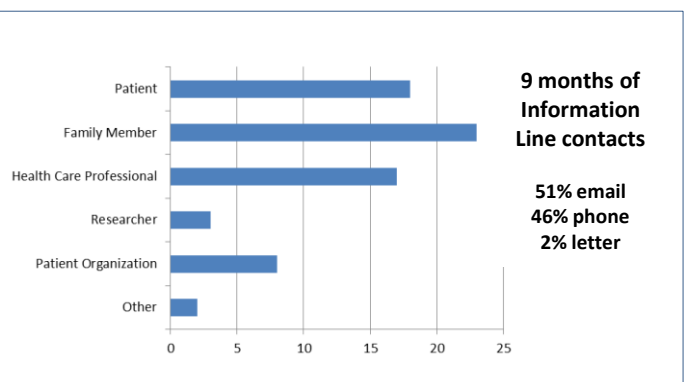
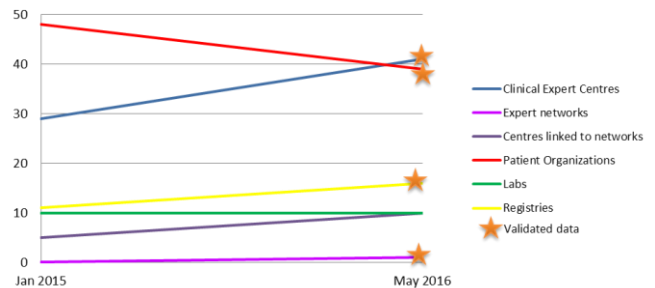
- Coordination of Care
- Research and trials
- Accessing specialists
- Managing/navigating the 'system'
- Support Groups, Benefits and Entitlements

GP survey by P. Byrne, UCD School of Medicine, 2014

Obstacles identified by GPs for RD patients:

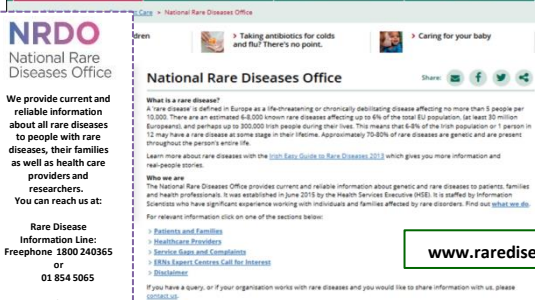
- Lack of knowledge by GP's (66%)
- Limited GP time for RD research (55%)
- Lack of support (36%)
- No national centre of expertise (32%)
- Too many diseases and not enough time, risk of de-skilling

Orphanet Ireland Activity



IN CONCLUSION

- Much achieved but still a long way to go
- Priorities to map-out resources
- ...Clinical Orphanet data for information line
- ...Expert centres for ERN linkage
- ...Diagnostic laboratories for Cross border genetic testing
- ...Registries to map out the extent of rare disease impact in our country



www.rarediseases.ie