International Niemann-Pick Disease Registry (INPDR)

Survey Report:
Niemann-Pick Diseases - Establishing the Learning Needs of Health Professionals

Background:

The International Niemann-Pick disease registry (INPDR) has been co-funded under the 2012 EU Health Programme by the Directorate General for Health and Consumers (DG-SANCO) via its Executive Agency for Health and Consumers. The INPDR is a collaborative disease registry owned by patient groups, clinicians, scientists and researchers involved in the care of patients with Niemann-Pick disease (NPD).

The registry collects clinical, genetic, diagnostic and outcome data from patients as well as a patient reported dataset designed for patients and family members to complete. The registry will enable progress in the field of NPD by allowing authorised access to anonymised clinical data, helping to identify and recruit patients to clinical trials and coordinate research efforts globally.

The purpose of this survey and the resulting report is to facilitate the development of educational applications that will support and enable the continued training of physicians and other healthcare professionals in relation to NPD. The survey has identified a number of areas in which the learning needs of health care professionals may be further supported and therefore impact positively upon the care and treatment of patients.

This report addresses specific objective 5 of the INPDR project:

To address the information needs of 60 patients/ carers (20 for each disease) to inform the development of FAQ's and the needs of 30 healthcare professionals to develop to educational material for NP diseases"

Method:

Healthcare professionals within 12 countries worldwide were invited to participate in the survey via email.

Participating Countries:

- UK
- Germany
- Czech Republic
- France
- Ireland
- USA
- Spain
- Canada
- Argentina
- Italy
- Netherlands
- Brazil

Those invited to participate were part of the wider networks of the International Niemann-Pick Disease Alliance and the Associate and Collaborating partners of the INPDR.

The survey was developed by the Niemann-Pick Disease Group (UK) and based on their knowledge of current educational opportunities and relevant literature relating to NPD plus existing gaps in provision. Because the purpose of the survey was exploratory, survey reliability and validity were not examined. The survey was reviewed by external sources from different disciplines and settings, and questions and response options were reworded to provide clarity.

A multiple-choice format was used for 6 items: experience of NPD, self-assessment of skills, training interest (3 items), barriers to learning and use of additional knowledge. The remaining 2 items were open-ended questions that addressed each of the following: obstacles in the application of new skills and knowledge, plus additional comments, needs and concerns.

In addition and in tandem, patients and family members affected by Niemann-Pick diseases were invited to take part in focus group sessions aimed at establishing their learning needs by understanding their 'frequently asked questions'.

The outcomes of these sessions, which took place at the 2014 Niemann-Pick UK Annual Family Conference and involved 64 directly or indirectly affected family members, will facilitate the development of resources to support and inform the NPD patient and family community. Participation of family members included representation of each disease type and eight countries worldwide:

- UK
- France
- Brazil
- Norway
- Spain
- Switzerland
- Canada
- Belgium

Results

The survey was answered by 36 respondents from a wide range of healthcare backgrounds; 61%, identified themselves as working in community/clinical practice, 30% in an academic setting and 25% as science/research professionals (summarised in Table 1).

Table 1. Professional/Educational status*:

Category	Respondents
Health Professional in community/clinical practice	22
Health Professional in academic practice/setting	11
Science/Research Professional	9
Student	0
Other:	1
*Please note; a number of healthcare professionals indicated their involvement and experience in more than one area.	

The professional status of the participants is summarised in Table 2. We expected that learning needs would differ amongst professional groups as a function of the training they received in their respective disciplines, therefore including professionals already practising provided a broad scope,

Table 2. Profession/Role/Specialty*:

Profession	Respondents
Specialist Consultant	6
Professor	4
Nurse/Nurse Specialist/Nurse Practitioner	13
Academic	2
Neurologist	2
Physiotherapist	4
Dietitian	3
Neuropathologist	1
Researcher/Scientist	3
Physician-Scientist	1
Paediatrician	3
Pharmacist	1
*Please note; a number of healthcare professionals indicated their professional involvement in more than one area.	

Table 3 relates to participants' self-assessment of their knowledge and skills. 47% of participants rated their knowledge of NPD as limited. 41% rated their knowledge as good and 12% as excellent.

Table 3. Please rate your personal knowledge of Niemann-Pick diseases:

Level of Knowledge	Number of Respondents
No Knowledge	0
Limited Knowledge	17
Good	15
Excellent	4

As reported below in Table 4, interest in further training was high despite competing interests, lack of administrative support, and time constraints (see Table 7). All respondents were interested in learning more about NPD, with 83% very interested and 17% somewhat interested.

Table 4. Please rate your personal interest in learning more about Niemann-Pick diseases:

Level of Knowledge	Number of Respondents
Not interested	0
Somewhat Interested	6
Very Interested	30
Not Applicable	0

Additional Comments:

- Would like to understand the critical aspects of the disease that patients and families face
- Develop new insights into disease pathogenesis and the identification of therapeutic targets
- Niemann-Pick is only a very small case with in my work load
- More information regarding novel therapies and genotype-phenotype correlations
- Would like to improve knowledge base; possible networking and sharing ideas to develop best practice

In Table 5, the biggest barrier to learning more about NPD is seen as time limitations and workload, followed by a lack of funding support. Some institutions were very supportive of further learning in this area, with three respondents telling us that they did not perceive any barriers to their learning. A key point was a perceived lack of appropriate learning materials.

Table 5. What do you see as the barriers to learning more about Niemann-Pick disease?

A lack of work place support	5
Time limitations/ workload	19
Lack of Funding	9
Travel limitations	5
Not a priority	4

Other (please explain):

- Lack of information regarding new diagnostic tests (2)
- Unclear information about symptoms at diagnosis (1)
- A lack of suitable learning material (4)
- No barriers at our institution (3)

Table 6 shows that additional knowledge of NPD was seen as valuable and most useful in providing information to patients and their families. Additional knowledge could also improve and enhance research and enable better patient care.

Table 6. How would you use additional knowledge about Niemann-Pick diseases?

Provide information to patients	19
Enhance patient care	17
Enhance clinical and diagnostic skills	13
Make practice-based improvements	15
Keep up-to-date	14
Other (please explain)	3

- To improve basic and translational research (1)
- To enhance research programmes (1)
- No explanation given (1)

We asked respondents to explain, using free text, what they saw as the obstacles to applying new knowledge into practice:

- Not easy to bother busy professional colleagues to find info on testing/clinical advice etc. Could information of this nature be made more widely available?
- Most colleagues tend to be in somewhat "siloed" environments (geographically, with regards to professional background and patient spectrum dealt with)
- The obstacles encountered in applying new knowledge into practice are: 1)finding grants; 2)poor interest for rare diseases
- Dissemination of knowledge to practitioners and availability of resources to implement
- The obstacles consist in the small number of patients and in the lack of international clinical guidelines
- Limited patient numbers and large other work load
- Dissemination of information and education of medical professionals
- Time constraints in clinical practice and acquiring specific skills with scoring systems used to assess patients with NPC
- I would favour a centralised (scientific) centre collecting all new knowledge, which should be verified before being transferred to clinical practice
- Colleagues/ peers with differing levels of knowledge; long standing workplace "culture"
- A lack of work place support

In addition we gave respondents the opportunity to list any needs or concerns that were not addressed in this survey. Only one response was received to this question:

In regard to data entry in specific countries - it is difficult to find a data manager for the compilation of the registers so that is done by medical doctors who have time limitations and workload.

Table 7, along with additional comments from Table 4 (reproduced below), addresses the educational and learning needs of professionals and summarises respondents preferred learning areas. The majority of respondents were interested in learning aspects relating to clinical practice, with opportunities to learn about clinical trials, improving diagnosis and new diagnostic techniques also seen as valuable. However, the individual needs of each different learning group must be recognised when considering the development of NPD specific educational resources.

Table 7. Which of the following learning areas do you consider to be the most important and which would be of interest to you?

Learning Area	Not important	Somewhat important	Very important	Not applicable
Diagnosis/Diagnostic techniques	2	9	12	0
Clinical practice	0	3	19	0
Clinical trials	1	8	15	0
Basic science research	1	11	10	0
Translational research	2	8	10	0
Please use this space to identify the topics you personally feel a need to learn more about :	• How to best develop and apply therapies for NP-C and other genetic disorders that indeed work. • Nutrition / nutritional requirements / disease progression in relations to the GI tract • Symptom Management • Pathogenesis			

Table 4 Additional Comments:

- Would like to understand the critical aspects of the disease that patients and families face
- Develop new insights into disease pathogenesis and the identification of therapeutic targets
- Niemann-Pick is only a very small case with in my work load
- More information regarding novel therapies and genotype-phenotype correlations
- Would like to improve knowledge base; possible networking and sharing ideas to develop best practice

Table 8 summarises the preferred learning format of respondents; the majority held a preference for the development of disease specific literature, with web-based resources and disease specific conferences also highly rated. Responses received will inform the INPDR dissemination plan. The importance of applicability of training and follow-through in the workplace was noted and the opportunity for open case discussion was also valued.

Given the variable responses, we conclude that learning formats and content should be provided in multi-media form and adapted to suit the institution/participants wherever possible. However, maximum impact could be gained via the provision of workshops/seminars, web-based on-line resources, the development of disease specific literature and disease specific conferences.

Table 8. Please tell us your preferred learning format:

Method	Not preferable	Somewhat preferable	Most preferable	Not applicable
Workshop / seminar	2	9	**11**	0
Web-based (online) resources	3	**14**	7	0
Training opportunity such as a university course	7	10	3	1
Disease specific literature	1	7	**15**	0
Grand Rounds	7	5	5	4
Disease Specific Conferences	2	10	**12**	0
Open case discussion	1	**13**	7	1
Panel discussions	7	8	5	0
Visual or Audio resources i.e. online videos/tools	4	**11**	7	0
Patient demonstrations	4	9	7	2
Other (please identify and explain)	No comments received			
No response provided	1			

Table 9 shows patient and family members most frequently asked questions, as determined during focus group sessions. This information will inform the development of educational literature for the NPD patient and family community and the healthcare professionals who support them. The sessions involved 64 family members, each of whom had been, or were, directly or indirectly affected by Niemann-Pick disease type A, B or C.

Table 9. What questions do patients and family members most frequently ask their healthcare professional?

How is Niemann-Pick disease inherited?	How is Niemann-Pick disease diagnosed?
What is the life expectancy of a person affected by NPD?	Is there a cure for NPD?
Is there any research is being carried out?	Where can I go for support and information?
How do I cope with the symptoms of NP-C/NP-B?	What treatments and therapies are available?
What are the symptoms or consequences of being a carrier?	Can my relatives be identified as high / low risk?
Can I have pre-natal testing?	Will all my children be affected?
How many people in the world have NPD?	How many different types of NPD are there?

Conclusion:

Participants were clearly interested in the development of further learning opportunities in NPD and generally saw this as important to their continuing professional development and to achieving best practice.

In addition, it was noted that learning opportunities are not necessarily financially supported in all work place settings and that time limitations present the biggest barrier.

Overall, all participants reported similar experiences, knowledge, and training interest. The few differences that emerged may be due to differences in professional disciplines/training, workplace expectations and available resources,

The obstacles to applying knowledge in practice were related to small patient numbers in NPD and other rare diseases, a general lack of interest in rare diseases and the need for a widely available, comprehensive and reliable professional information resource.

In addition, there is a need to develop informative resources that can support healthcare professionals in communicating information about NPD to patients and their families.

As stated previously, given the variable responses, we conclude that learning formats and content should be provided in multi-media form and adapted to suit the institution/participants wherever possible.

Finally, whilst not relevant to this exercise, it should be noted that one comment received referred to the difficulty of data entry in countries where there was no data entry support. This is considered as a possible risk to the project and consideration should be given to adding this concern to the risk register.

If you would like to discuss this survey or learn more about the INPDR, please contact:

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