

Screening for neuromuscular diseases: Patient organisations' achievements in European Reference Networks

F Lamy, EURO-NMD Patient Advisory Board (PAB), H Pascal, T Evangelista
ERN EURO-NMD

INTRODUCTION

The development of new genetic testing methods associated to the arrival of the first treatments that modify the natural history of the diseases have raised several questions regarding screening for neuromuscular conditions. The screening directives and access to the different existing techniques, are not uniform across Europe. The unequal degree of knowledge of the general population and clinicians about the screening situation in their own country prompted the patient advisory board (PAB) of the European Reference Network (ERN) for rare neuromuscular diseases (EURO-NMD) to conduct a qualitative study to take stock of the current state of play regarding the screening for neuromuscular diseases (NMD) in Europe and the patients' vision on this matter.

AIM

Collect data on screening for NMD around pregnancy and birth in Europe
Understand the position of patient organisations regarding screening for NMD.

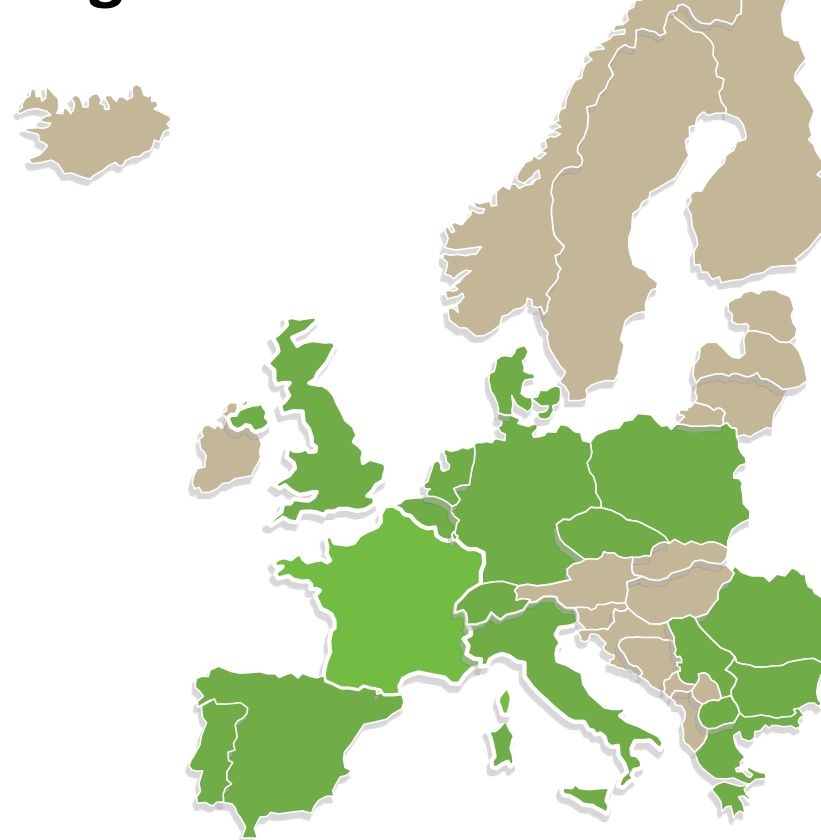
METHODS

- An online questionnaire with four main points was developed by the PAB
 - Does screening exist in your country?
 - Is it done routinely?
 - Who supports its costs?
 - What are your organization's views on screening?
- Sent to **110+** patient associations **2 emailing campaigns**: 04/06/2019 and 23/07/2019
- Survey staid open from June to October 2019
- Target: Patient organisations in European countries

DEMOGRAPHICS

We collected **30 answers** (response rate 30%) from patient associations based in 18 European countries. Of these 7 were not members of EURO-NMD at the time of the survey (Fig. 1).

Fig. 1



Legend: Countries that contributed to the survey (and number of answers per country). Belgium (1), Bulgaria (1), Czech Republic (1), **Denmark***(1), Germany (1), **Greece***(1), Italy (1), **Luxembourg***(1), The Netherlands (1), Poland (1), **Portugal***(1), **Serbia***(1), **Switzerland***(1), UK (1), France (2), **Macedonia***(2), **Romania***(2), Spain (10).

* countries that at the time were not part of EURO-NMD.

TWO MAIN CATEGORIES OF PATIENTS' ASSOCIATIONS:

- Umbrella organizations (represent all NMD): 43.3% (13/30) of the answers
- Disease specific organizations (represent a disease or thematic group of diseases): 56.7% (17/30) of answers

RESULTS

"IS PRE-IMPLANTATION DIAGNOSTIC (PGD) IN PLACE IN YOUR COUNTRY?"

YES	15/30 (9/18 countries)	Spain (5), Romania (1) , Portugal, The Netherlands, Greece, Germany, France, Bulgaria, Belgium
NO	15/30 (10/18 countries)	Czech Rep., Denmark, Italy, Luxembourg, Macedonia, Poland, Romania (1) , Serbia, Spain (5) , Switzerland

When we looked at the answers per country, we noticed inconsistencies

If yes does it include NMD? 14 yes, 1 no

"IS PRE-NATAL SCREENING IN PLACE IN YOUR COUNTRY?"

YES: 25 + 1 (Portugal)/only possible if a family history, or in very symptomatic cases
NO: 4 (2 countries: **Spain** and Switzerland/18)

When we looked at the answers per country, we noticed inconsistencies (Spain 7 yes PNS is available and 3 no)

"IS NEWBORN SCREENING IN PLACE IN YOUR COUNTRY?"

YES: 24
NO: 6 [France (1), Macédoine (2), Serbie (1), Espagne (2)]

IF YES, does it include NMD?

NO: 14
YES: 10 [Belgium (1), Bulgaria (1), Greece (1), Italy (1), Romania (1), Spain (5)]

Belgium: ongoing pilot for SMA

Italy: starting pilot for SMA / amendment for neuromuscular diseases in 11/2018

Netherlands: screening program for SMA to be started within 2 years

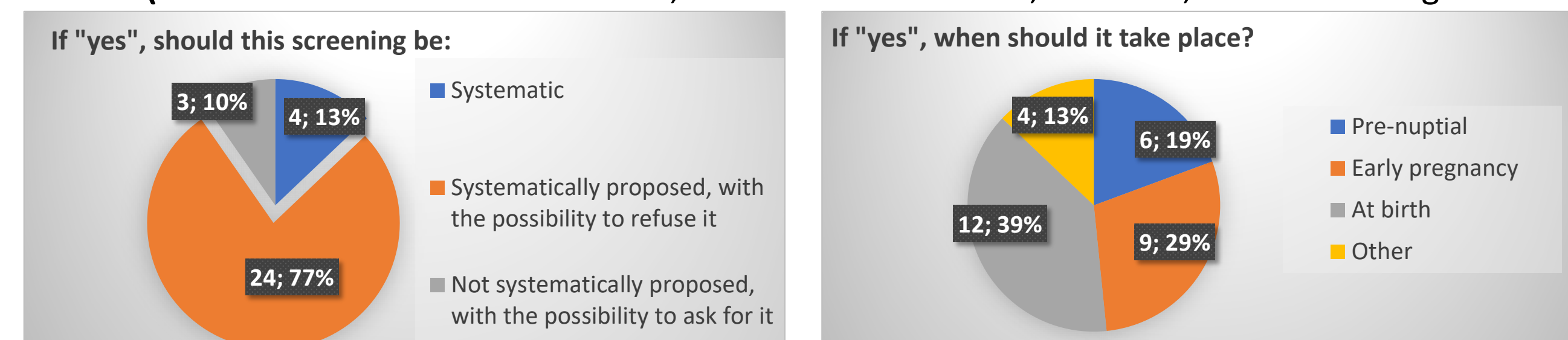
Germany: ongoing pilot for SMA

PATIENTS ORGANIZATIONS' OPINION REGARDING SCREENING

1. Are you in favor of screening for the condition(s) which are relevant to your organization?

YES: 28

NO: 2 (Reasons: Lack of reimbursement, Absence of treatment, Personal, cultural or religious belief)



Answers by diseases groups

	ALL	NMD	SMA	DMD	OTHER
Not systematically proposed, with the possibility to ask for it	11%	17%	0%	0%	20%
Systematically proposed, with the possibility to refuse it	75%	58%	100%	83%	80%
Systematic	14%	25%	0%	17%	0%

Nb of POs: 28, 12, 5, 6, 5

The opt-out option is preferred regardless the pathology

	ALL	NMD	SMA	DMD	OTHER
birth	43%	42%	100%	17%	20%
early pregnancy	25%	17%	0%	33%	60%
pre-conception	21%	17%	0%	50%	20%
it depends	11%	25%	0%	0%	0%

Nb of POs: 28, 12, 5, 6, 5

For SMA: screening at birth. Otherwise before birth techniques gain interest.

17. If [you are in favour of screening], for what reasons?

Please select the relevant reasons and rank them (1 being the most important). If you have other reason(s) please specify in question 18

	Shorter diagnostic	early access to treatments	inclusion in clinical trials	preventive care	genetic counseling
1	50%	69%	33%	44%	48%
2	27%	19%	25%	36%	26%
3	12%	4%	17%	12%	15%
4	8%	8%	0%	8%	7%
5	4%	0%	21%	0%	4%
6	0%	0%	4%	0%	0%

S [1-3] rank [1]: 88%, 92%, 75%, 92%, 89%

All data N= 28 patients organisations

Priority is given to treatment access

Then comes with equal importance:
- Shorter diagnostic
- Preventive care
- Genetic counseling

Closely after inclusion in the clinical trials

CONCLUSION

Current situation regarding screening

It seems that many POs are not precisely aware of the screening system in place in their country. In some cases this knowledge is limited to the diseases they are related to.

Opinion regarding screening

POs are massively in favour of screening. When to screen and for what reason depends on the pathology.

The presence of a disease modifying treatment seems determinant. In the absence of treatment preventive care and genetic counselling would be the priority for POs.