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Genetic testing among Spanish pediatric neurologists: Knowledge, attitudes and practices

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ABSTRACT

Advances in genetic testing applied to child neurology have enabled the development of genetic tests with greater sensitivity in elucidating an etiologic diagnosis for common neurological conditions. The objective of the current study was to examine child neurologists' perspectives and insights into genetic testing. We surveyed 118 Spanish child neurologists, exploring their knowledge, attitudes, and practices concerning genetic tests. All of them had requested at least one genetic test in the past six months. Global developmental delay or intellectual disability in absence of a strong specific etiologic suspicion and autism spectrum disorders were the disorders for which genetic testing was most frequently requested. The most commonly requested genetic test was CGH-array. Overall, child neurologist perception of readiness for making genetic-related decisions was not bad, although many would like to have a greater support from geneticists and were interested in increasing the time dedicated to genetics within their continuing education program. These data have important implications for future practice, research, and education.

1. Introduction

Advances in clinical and molecular neurogenetics have achieved considerable success in providing a clearer understanding of genetic alterations responsible for an increasing variety of neurological disorders that affect children. New technology has allowed the development of genetic tests with greater sensitivity in elucidating an etiologic diagnosis for common conditions as intellectual disability or epilepsy [Chambers et al., 2016; Flore & Milunsky, 2012; Ream and Patel, 2015]. In child neurology the number of clinical genetic tests available is thus rapidly rising, and hence the need for increased genetic knowledge among physicians.

A limited number of studies have assessed physicians' uses and familiarity with genetic testing [Hoop et al., 2008; Klitzman et al., 2013; Lawrence and Appelbaum, 2011; Li et al., 2015; Prochniak et al., 2012] and only three previous studies have examined clinicians' perspectives on genetic testing for neurological diseases [Salm et al., 2014; Thies et al., 1993; Yoshida et al., 2013]. To our knowledge, no data have been previously published in relation to neuropediatricians' attitudes and practices toward genetic testing.

As the use of genetic tests in child neurology is expected to increase in upcoming years, understanding neuropediatricians' knowledge, attitudes, and practices regarding genetic testing and factors involved is crucial. It is critical to know what genetic tests are ordered on neuropediatric patients, how often, and for which diseases. Moreover, identification of barriers and facilitators to appropriate use of genetic tests is an important issue to design effective interventions and implement educational actions to improve detected deficiencies.

The objective of the current study was to examine child neurologists' perspectives and insights into genetic testing. To appropriate investigate and evaluate neuropediatricians' views on this topic, we performed a national survey of Spanish pediatric neurologists, asking about their self-perceived knowledge and practice regarding genetic tests, and their attitudes towards issues related to genetics.

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2. Methods

2.1. Subjects

The Spanish Pediatric Neurology Society (SENEP) was contacted regarding a membership list of 404 pediatric neurologists that was available for purchase. In Spain, most of child neurologists are members of SENEP.

2.2. Survey content

The survey was developed by the first and sixth authors. Questions targeted respondent demographics, clinical practice variables concerning to genetic testing, self-reported knowledge of genetics and genetic testing, barriers and facilitators to appropriate use of genetic tests, and perceived adequacy of training and continuing education in genetic testing. The questionnaire items were based on a comprehensive review of the literature and investigators' clinical experience.

The survey consisted of 25 questions and took approximately 15 min to complete. It was composed of dichotomous, multiple choice, and Likert scale questions. Survey questions are also reflected in the items in Tables 1 and 2 and Figs. 1–4.

Table 1
Respondentś sociodemographic and clinical practice concerning to genetic testing.

	n (%)
Total	118
Gender	
Male	43 (36.4)
Female	75 (63.6)
Age	
<30	5 (4.2)
30–40	55 (46.6)
40–50	18 (15.3)
50–60	27 (22.9)
>60	13 (11)
Residency previous to Child Neurology training program	
General Pediatrics residency	109 (92.4)
General Neurology residency	8 (6.8)
Year of ending of the Child Neurology training program	
Before 1995	40 (33.9)
1995–2005	23 (19.5)
After 2005	55 (46.6)
Number of patients attended per week	
<10	2 (1.7)
10–19	9 (7.6)
20–29	11 (9.3)
30–39	19 (16.1)
40–49	22 (18.6)
50–59	21 (17.8)
60–69	20 (16.9)
70–79	4 (3.4)
80	9 (7.6)
Number of genetic tests ordered	
<6 tests per year	3 (2.5)
6-12 tests per year	4 (3.4)
1-2 tests per month	13 (11.0)
3-4 tests per month	32 (27.1)
1-2 tests per week	32 (27.1)
3-4 tests per week	22 (18.6)
>4 tests per week	12 (10.2)
Genetic counseling offered to patients in the past six months	
Yes	96 (81.4)
No	20 (16.9)
Preimplantation genetic diagnosis discussed with patients in the past six months	
Yes	62 (52.5)
No	56 (47.5)

n = 118. Individuals were not required to answer every question. Throughout the survey and in this table, responses may not sum to 118.

Table 2
Beliefs and attitudes among Spanish child neurologists.

	n (%)
Self-reported knowledge of genetics:	
Excellent	4 (3.4)
Good	31 (26.3)
Average	54 (45.8)
Somewhat poor	27 (22.9)
Very poor	2 (1.7)
Comfort ordering genetic tests	
Totally comfortable or comfortable	31 (26.3)
Partially comfortable	72 (61)
Uncomfortable or strongly uncomfortable	14 (11.9)
Comfort counseling patients on genetic testing	
Totally comfortable or comfortable	25 (21.2)
Partially comfortable	73 (61.9)
Uncomfortable or strongly uncomfortable	20 (16.9)
Self-referral capacity in discussing preimplantation genetic diagnosis with families	
Qualified	25 (21.2)
I need an additional training	68 (57.6)
Not qualified	24 (20.3)

2.3. Survey administration

A link to the survey was sent electronically to the 404 active members of the Spanish Pediatric Neurology Society (SENEP). The initial email notifying the members of the survey was sent on October 19, 2015, with a reminder email sent three weeks later to improve the participation rate. Responses were collected from October 2015 to December 2015. Monetary incentives were not offered to respondents.

2.4. Data analysis

SPSS version 22.0 software was used to perform statistical analyses. Chi-square tests were used to examine categorical variables. We also conducted a multiple logistic regression to explore which independent variables were most associated with physicians referred knowledge and the comfort ordering genetic tests and counseling patients on genetic testing.

3. Results

3.1. Demographics and clinical practice concerning to genetic testing of respondents

In total, 118 pediatric neurologists completed the survey, resulting in a 29.2% response rate based on the 2015 Spanish

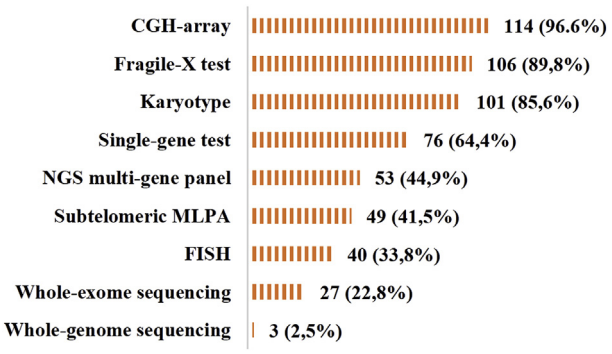

Fig. 1. Summary of most commonly requested genetic tests by Spanish child neurologists in the past 6 months. (CGH-array: array-based comparative genomic hybridization; NGS: next-generation sequencing; MLPA: multiple ligation-dependent probe amplification; FISH: fluorescence in situ hybridization).

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