

Spanish mental health residents' perspectives about residency education on the genetics of psychiatric disorders: A cross-sectional survey

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Abstract

Despite compelling evidence that some patients with a psychiatric diagnosis could benefit from genetic assessment, genetic testing for psychiatric patients is underutilized. Few studies have reported psychiatric genetics training for mental health specialists, and such research is especially lacking in Spain. We aimed to gather the opinions of Spanish mental health residents, including resident intern nurses (RINs), doctors (RIDs) and psychologists (RIPs). A short survey was prepared by an expert team and distributed to all mental health residency centers in Spain during the first semester of 2021. Of the 2028 residents, 18% responded. Participants were mainly females (71%), in their first year of residency (37%) and within the 27–31-year age range. While participants received little theoretical (13.4%) and practical (4.6%) training on average, RIDs had the most affirmative responses. Notably, RINs and RIDs were interested in genetics during residency (>40%) and strongly believed (85.0%)

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that genetics training using both theoretical and practical methodologies should be incorporated into residency training. However, RIPs were less interested (20%), and only 60% believed that genetics training should be incorporated. Spanish mental health residents, although interested in genetics in psychiatry, receive little training on this topic. They strongly believe that genetics training using theoretical and practical methodologies should be incorporated.

KEYWORDS

education, genetics, mental health, psychiatry, residents, survey

1 | INTRODUCTION

Some psychiatric disorders, such as autism, schizophrenia and bipolar disorder, have a strong genetic component. The heritability of these disorders is approximately 80%, a figure much higher than that of other complex diseases such as diabetes or coronary heart disease. Other psychiatric disorders (such as depression, anxiety, and eating disorders) have lower genetic components.¹

Currently, it is accepted that the genetic component of such disorders is derived from common and rare genetic variants. Several international consortia, including the Psychiatric Genomics Consortium, have used genome-wide association analysis (GWAS) based on single nucleotide polymorphisms (SNPs, with a frequency >1%) to identify some of the common variants associated with each psychiatric diagnosis. For instance, the largest GWAS for schizophrenia identified 120 genes associated with the disorder, with an SNP-based heritability of 0.24 (24% of the genetic variance is explained by these SNP variants).² Similar studies on other severe psychiatric disorders have yielded similar results, identifying hundreds of SNPs in each psychiatric condition and confirming a polygenic model.³ With information from these studies, a new parameter can be calculated for each individual to infer his or her genetic risk for a psychiatric diagnosis: the polygenic risk score (PRS). PRSs, by itself, are not useful as a diagnostic element, but if combined with other risk factors, they can be a useful tool both for diagnostic processes and for prognosis and treatment evaluations.⁴ For instance, while PRSs have poor predictive power in general psychiatry, they have positive predictive value for some individuals, such as carriers of 22q11.2 deletions.⁵ Genome sequencing studies allowed us to identify rare (frequency <1%) single nucleotide variants (SNVs) with a high phenotypic impact, which together can explain 1%–2% of the genetic variability in the case of schizophrenia.⁶ In addition, a considerable amount of the genetic variance can be explained by another type of rare variant known as copy number variants (CNVs), which are deletions or duplications of stretches of DNA (from 50 kb to hundreds of Mb). Some of these CNVs are currently known as neuropsychiatric CNVs because they are recurrent and highly associated with different psychiatric conditions.⁷ The relatively common (1/2000 births) CNV, 22q11.2 deletion, has been exhaustively studied in terms of phenotypic expression, and guidelines on clinical treatment and education and lifestyle recommendations exist.^{8,9} It has been estimated that carriers of 22q11.2

deletions and duplications (known as 22q11.2DS) have a 60%–80% risk of developing a psychiatric disorder.¹⁰ Altogether, rare variants (CNVs+SNVs) can explain up to 40% of intellectual disability, 25%–35% of autism spectrum disorders and 10% of schizophrenia.^{5,6} Currently, chromosome microarray, the first-tier test to detect CNVs, is not expensive. Genetic studies, if clearly indicated, should be performed in specialized genetic counseling units.¹¹

Although guidelines have been established for clinical genetic testing in subjects with developmental delay, intellectual disability and autism spectrum disorder (ASD) with or without congenital anomalies, routine genetic testing in psychiatry clinics is lacking.⁵ There is an active debate, including ethical issues, regarding whether genetic testing should be offered to all psychiatric patients.¹² Most experts agree that genetic testing should at least be performed in syndromic cases, that is, in patients with more than one psychiatric condition and one or more nonpsychiatric diagnostics.^{5,6,11–13}

After a decade of clinical genetic testing recommendation for autism in Sweden, according to a survey study, <10% of patients with an ASD diagnosis were offered a referral to clinical genetic testing.¹⁴ Likewise, a Spanish survey found that genetic counseling was rarely offered to psychiatric patients, even though patients were interested in the topic.¹⁵ Mental health professionals consider it necessary to improve their knowledge of psychiatric genetics to implement genetic counseling services that account for the complexity of mental disorders.^{16–19} However, the reality is that psychiatrists lack the necessary training that would allow them to routinely consider genetic testing as part of their diagnostic tools.^{6,20} In some countries, such as the United States of America and the United Kingdom, where the need for genetic assessment has already been detected, measures have been adopted to improve training in psychiatric genetics for mental health specialists.^{17,20–23} including the proposal for rotation by genetic services.²³ However, in Spain, while training programs for residents in the field of mental health are officially stated to include training in genetics, such training does not appear to be offered actually. To address this issue, a working group was created in 2019 under the umbrella of the Spanish Network on Biomedical Research on Mental Health (CIBERSAM, www.cibersam.es). Here, we present the results of a survey to gather the perspectives of Spanish mental health residents about the importance of genetics in psychiatry and their training on this topic during residency.

2 | MATERIALS AND METHODS

2.1 | Study participants and ethical considerations

This was an observational cross-sectional study. The target population was all Spanish centers accredited by the Spanish Ministry of Health with mental health residency training programs. Some hospitals have single-specialty mental health units, while general hospitals have multispecialty residency training programs.

According to the published lists from all these units, in 2021, there were a total of 2028 residents, including 987 psychiatrists (resident intern doctor, RID), 593 clinical psychologists (resident intern psychologist, RIP) and 448 nurses (resident intern nurse, RIN) (Appendix S1, Annex 1). RID and RIP training programs last 4 years, and RIN training programs last 2 years.

The study was designed and conducted according to the Helsinki Declaration, and the project was approved by the Ethics Committee of the main investigator institution, Institut d'Investigació Sanitària Pere Virgili (CEIm-IISPV). The survey was completed anonymously, and data were collected in aggregate form to avoid participant reidentification. No signed informed consent was therefore necessary from participants.

2.2 | Data collection

2.2.1 | Questionnaire

A team of psychiatry and biology experts in psychiatric genetics who belonged to the genetics working group of the Spanish network CIBERSAM prepared the questionnaire. Five residents from the same hospitals were invited to join the project team. A questionnaire was designed to collect data from participants and to meet the following requirements: (1) web-based allowing easy access via tablet, personal computer, or mobile phone; (2) short response time; and (3) anonymous. The questionnaire consisted of two parts. The first part measuring the independent variables (specialty, age range, sex, residency city, type of residency unit) was preceded by a filtering main question to ensure that all participants were mental health residents in training. The second part consisted of five questions with categorical responses, and in some of them, an open text box was offered to add free-text comments. The questionnaire was piloted ($N = 12$ residents) in one training unit and refined according to participants' comments to improve it. Finally, the questionnaire was posted on the *Google Forms* platform (Appendix S1, Annex 2).

2.2.2 | Survey distribution

The survey was distributed between January and July 2021 through three different channels. First, an e-mail was sent to all Spanish units with mental health residency training programs as well as to several resident associations, academic colleges, RID exam prep academies,

and Spanish scientific societies related to mental health (the complete list is available in Table S1). The text first provided a short description of the project's nature and aim and then invited residents to participate and provided the link to the survey. Up to three reminders were sent to each recipient. Second, a project Twitter account (@formagenesis) was created to promote participation. Content related to the study and reminder invitations to participate were posted to this account. Finally, online informative short talks (approximately 30 min) were delivered by the team members, mainly residents, to several training resident units. During the talks, a QR code was inserted in the final presented image to allow direct access to the survey.

2.2.3 | Independent variables

Sociodemographic data from participants included specialty (psychiatrist, clinical psychologist or mental health nurse); residency year (1st, 2nd, 3rd, or 4th for RIDs and clinical RIPs and 1st or 2nd for RINs); age range (<22, 22–26, 27–31, 32–36 and >36); gender (male, female, or other); the city where the residency was located, which was grouped by Spanish region ("comunidad autónoma"); and type of residency unit (multispecialty or mental health).

2.2.4 | Outcome variables

The research questions, which referred to participants' residency education period, were as follows:

1. Have you received theoretical training on genetics in psychiatry? If the previous answer is affirmative, specify what training you have received.
2. Have you received practical training in the application of genetics in psychiatry? If the previous answer is affirmative, specify what training you have received.
3. Have you been interested in genetic issues in psychiatry during your training as a specialist?
4. Do you think that training in genetics should be incorporated in the future?
5. Which other training methodology would you most like to receive? This question had a categorical answer: (1) Theoretical lectures, (2) Workshops of clinical cases, (3) Consultation practice with real cases, (4) The three mentioned above, and (5) Other
6. Comments you want to make to us

2.3 | Statistical analysis

Descriptive analysis was performed using frequencies and percentages for categorical variables (age, gender, region, type of residency unit, residency year). The chi-square test was used to check the difference in the resident responses according to their professional profile and/or year of residency. Statistical analysis was conducted using IBM

TABLE 1 Target population and participation according to residency specialty program.

Specialty program	N ^a	N (%)
RIN	448	87 (19.4)
RID	987	163 (16.5)
RIP	593	117 (19.7)
Total	2028	367 (18.1)

Abbreviations: RID, resident intern doctor; RIN, resident intern nurse; RIP, resident intern psychologist.

^aData obtained from the official lists of each Spanish center with mental health residency programs in 2020.

SPSS software version 21.0, and the statistical significance was set at $p < 0.05$. GraphPAD PRISM 9 was used to create graphs, and BioRender was used to create the Graphical Table of contents (BioRender.com).

3 | RESULTS

3.1 | Sample description and survey participation

During the 7 months that the survey was accessible online, 369 residents completed the survey. Two responses had to be excluded due to incompleteness. Of the 367 valid responses, 23.7% of respondents were RINs, 44.4% were RIDs and 31.9% were RIPs. The percentage of respondents was similar (range 16.5–19.7) between specialties (Table 1). Most respondents were females (70.6%), were in their 1st year of residency (36.8%), were in the 27–31-year age range (45.5%), were from Catalonia (29.2%) and were from a single-specialty mental health residency unit (64%) (Table 2). Detailed data of participants according to their specialty and residency year are shown in Table S1.

3.2 | Theoretical and practical training in psychiatric genetics

Panels A and B of Figure 1 and Table S2 show the results of the survey for the questions about the training received according to residency specialty and residency year. As shown in Figure 1A,B, 13.4% of respondents reported that they had received theoretical education on psychiatric genetics, and 4.6% reported that they had received practical education (dashed gray bars). RIDs reported higher affirmative responses (17.8%, yellow dashed bars) than RIPs (12.8%, red dashed bars) and RINs (5.7%, blue dashed bars) regarding having received theoretical education ($p < 0.05$). However, the responses of having received practical education were similar (mean 5%) among respondents with different residency specialties (Figure 1B). Regarding differences by residency year for both questions on education received, RIDs in year 4 scored higher than those in years 1–3 ($p < 0.001$). RIPs in years 3 and 4 reported that they had received less genetics education than those in years 1 and 2 ($p < 0.005$), but there

TABLE 2 Sample description according to specialty program.

	Residency program			
	RIN ^a	RID	RIP	Total
N	87	163	117	367
Year (%)				
1st	12.0	14.2	10.6	36.8
2nd	11.7	13.1	6.0	30.8
3rd	–	10.1	10.9	21.0
4th	–	6.3	5.2	11.4
Age (%)				
<22	0	0	0	0
22–26	14.7	15.3	10.1	40.1
27–31	6.0	25.3	14.2	45.5
32–36	1.4	1.6	5.2	8.2
>36	1.6	2.2	2.5	6.3
Sex (% females)	81.6	60.7	76.1	70.6
Region (%) ^b				
Andalucía	1.1	4.6	3.3	9.0
Catalonia	5.4	12.3	11.4	29.2
Comunidad Valenciana	2.5	5.2	3.5	11.2
Madrid	4.1	7.6	4.4	16.1
The Basque Country	1.9	3.3	1.1	6.3
All other	8.6	10.3	8.1	28.3
Type of education center (%)				
Mental health only	16.3	25.3	22.3	64
Mental health and other	7.4	9.5	19.1	36

Abbreviations: RID, resident intern doctor; RIN, resident intern nurse; RIP, resident intern psychologist.

^aThe RIN residency spans 2 years.

^bRegions where <5% of the total participation are grouped.

were no differences in practical training according to residency year. Among RINs, no differences were observed between those in year 1 and year 2 in either theoretical or practical training.

3.3 | Interest in psychiatric genetics and need for more training

Panels C and D in Figure 1 show the results of the survey for the questions about interest in genetics and the need to incorporate genetics training according to residency specialty and residency year. A total of 40% of respondents reported an interest in psychiatric genetics as a topic (Figure 1C, gray dashed bars). Remarkably, the percentages of RINs and RIDs who responded affirmatively (48.3 and 49.7, respectively) were significantly higher ($p < 0.0001$) than that of the RIP group (24.8%) (Figure 1, colored dashed bars in panel C). Participants responded affirmatively (82%) to the need to incorporate more education on psychiatric genetics (Figure 1D, dashed gray bar). In a comparison among the different specialty groups, RINs scored

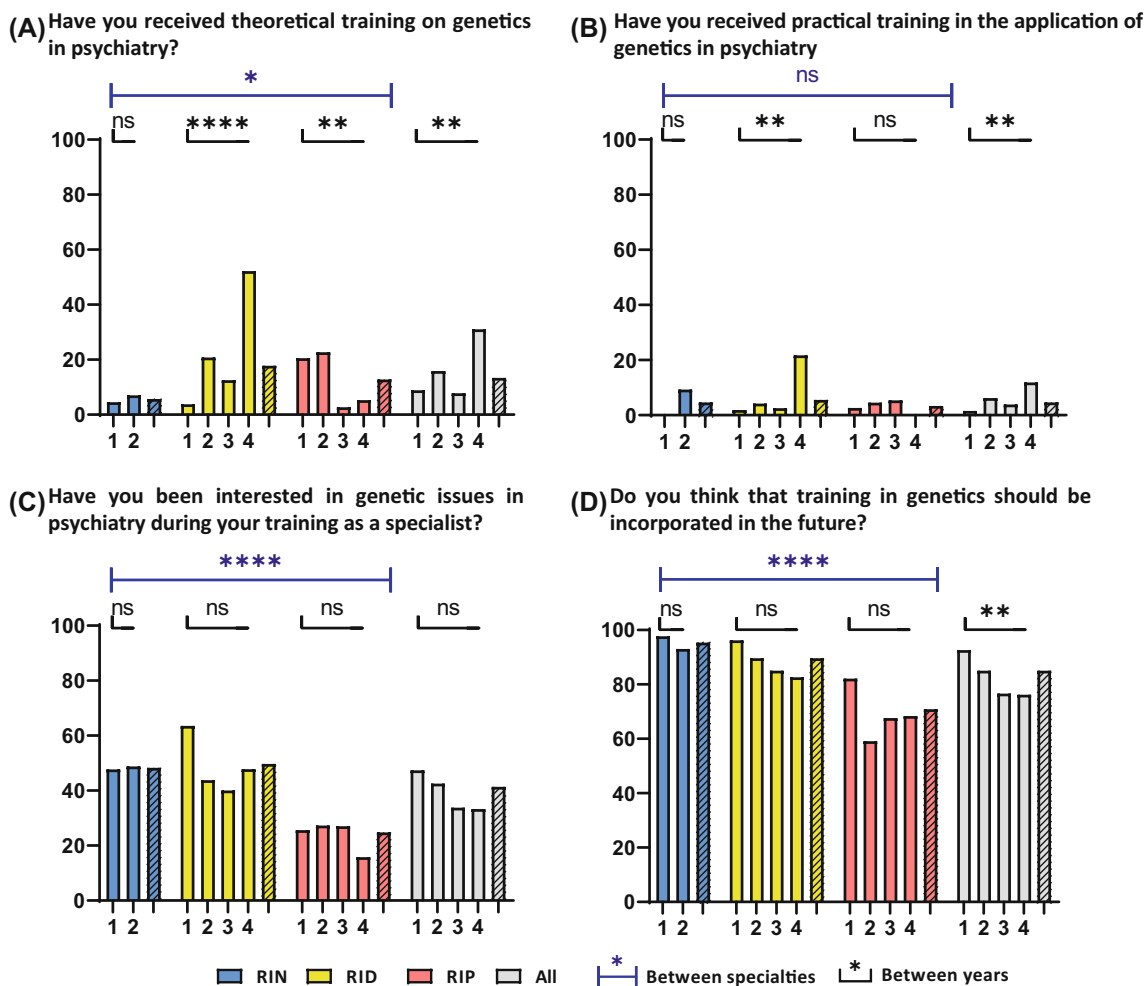


FIGURE 1 Percentage of affirmative responses to the questions about genetic training and interest during residency according to specialty and residency year. Dashed bars represent the mean for each specialty and all responders together. Chi-squared test p value: * $p < 0.05$; ** $p < 0.005$; *** $p < 0.0001$. [Colour figure can be viewed at [wileyonlinelibrary.com](https://onlinelibrary.wiley.com)]

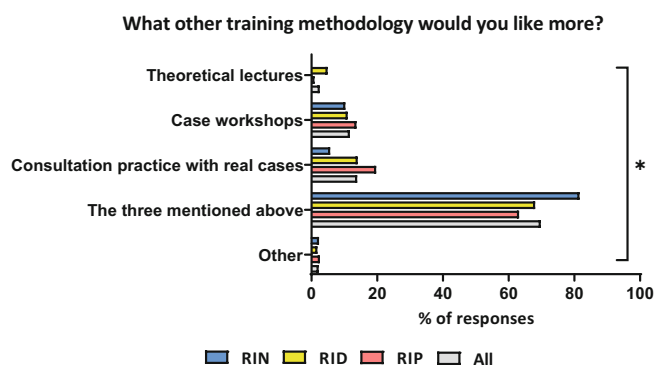


FIGURE 2 Percentage of responses to the preferred methodology question according to specialty. Chi-squared test p value: * $p < 0.05$. [Colour figure can be viewed at [wileyonlinelibrary.com](https://onlinelibrary.wiley.com)]

higher than RIDs and RIPs, and RIDs scored higher than RIPs. (95%, 89.6%, and 70.9%, respectively, $p < 0.0001$) (Figure 1, colored dashed bars in panel D). There were no differences by year of residency in the responses to either question (Figure 1C,D).

3.4 | Preferred type of training methodology

Figure 2 shows the percentage of responses to the question about the preferred training methodology according to residency specialty. On average, 69.8% of respondents chose the option “the three mentioned above”, indicating that they would like to receive training in the form of theoretical lectures, workshops and practice based on real cases. The options ‘Consultation practice with real cases’ and ‘Workshops of clinical cases’ were chosen by 13.9% and 11.7% of respondents, respectively, while ‘theoretical lectures’ was chosen by only 2.5% of residents. No differences were observed among specialties.

3.5 | Open comments by respondents

Forty free-text comments were obtained among all the participants (10.9%) that could be grouped into five categories. From all responses, 35% of the comments fell in the category “important issue, more training needed”, 22% lacked content and were categorized as null,

20% of the comments contained content “out of context”, 12.5% commented that the issue of the survey was “a non-priority issue” and 10% were “thanks comments”.

4 | DISCUSSION

Despite compelling evidence that some patients with a psychiatric diagnosis could benefit from genetic testing and that genetic tests are available, genetic testing for psychiatric patients is underutilized outside medical genetics units.^{5,6,11,14,24} In Spain, only a few medical genetics units for neurodevelopmental disorders exist, and the referral of psychiatric patients for genetic counseling and/or genetic testing is the exception, not the rule.^{15,17} Similar scenarios have been observed in other countries,⁶ with a few studies focused on ASD, including two studies in the USA^{25,26} and one in Europe.¹⁴ However, no studies in Spain have explored the use of genetic testing by mental health professionals. Here, we present the results of our survey made available to all residents ($N = 2028$) in the mental health field during the first half of 2021. We observed a great discrepancy between the amount of training that residents receive (very low according to their opinion) and their perspective on what they should receive (>80% of residents believed that education in psychiatric genetics should be incorporated into their resident programs, mainly through both theoretical and practical methods). These results are in agreement with previous studies showing that psychiatrists and other mental health professionals perceive that they are not trained to implement genetic testing recommendations.^{16–18,20} A positive result from our study is that year-4 residents reported more affirmative responses regarding having received genetics education, and concomitantly, they felt less need for this topic to be incorporated into their resident programs. This finding was significant specifically for RIDs. This finding could be interpreted to indicate that at least some residents received some education on psychiatric genetics during their residency.

In our study, RIPs were less interested than RIDs and RINs in psychiatric genetics. This finding is concerning, as psychologists can play a very active role in the genetic counseling process. RIPs can help the patient and his or her family members understand and adapt to the result of the genetic diagnosis. In addition, according to the treatment recommendation guides for each genetic alteration, some of the recommended interventions are often psychological. For instance, cognitive behavioral therapy can be recommended for patients carrying a genetic variant that increases the risk for psychosis since this therapy is beneficial in preventing the transition to psychosis.¹³ Another example is that clinical psychologists can monitor cognitive functions to be able to advise on educational plans and life plans in 22q11.2DS carriers.⁹ In contrast, RINs who had the lowest level of education on psychiatric genetics (5.7% in both theoretical and practical training) strongly believed (95.4%) that psychiatric genetics education should be included in their resident programs.

Our results can be the starting point for (1) a future study to collect the opinions of active professionals in the mental health care field in Spain, (2) a recommendation to the Spanish commissions

responsible for mental health resident programs to increase education (theoretical and practical training) on psychiatric genetics and (3) a recommendation to health sciences university faculties to improve their education programs in clinical genetics. Other countries have already determined this necessity,^{17,19–21} and some countries have already launched psychiatry genetics-specific programs that could be implemented in Spain.^{27,28} Interestingly, in 2007, Finn and colleagues reported a detailed and useful list of recommendations for genetics education for psychiatry residents.²³ Moreover, education tools such as the “Psychiatric Genetic Counseling for Genetic Counselors” workshop that was designed for genetic counselors²⁸ could be adapted to empower mental health professionals to do their side in implementing genetic counseling in their clinical settings.

Some limitations of our study need to be accounted for. First, the participation rate was 18% of the total target population. Despite not having a high percentage of participation, we must emphasize that the result was similar to other web-based surveys.^{29,30} Second, we cannot rule out a bias in sample participation. For instance, we had more participants from the Madrid and Catalonia regions, where a higher proportion of hospitals are primary and secondary hospitals with clinical genetics units, rather than tertiary hospitals that rarely have these services. This may have influenced awareness about genetics in the participants from these regions. Likewise, we had more participation from Catalonia, Madrid, Valencia, Andalucía and The Basque Country, which were the regions represented by a resident on the research team, and team residents were very active in promoting participation among their colleagues. In addition, we cannot rule out that survey respondents were those residents more informed about genetics. However, our results are similar to those of other studies.^{15,31}

In summary, through a survey of Spanish mental health residents in 2021, we concluded that on average, these residents received little theoretical (13.4%) and practical (4.6%) training in genetics, were interested in genetics during residency (41.4%) and strongly (85.0%) believed that training in genetics through both theoretical and practical methodologies should be incorporated into residency training. RIPs were significantly less interested in genetics than RINs and RIDs.

AUTHOR CONTRIBUTIONS

Elisabet Vilella and Iñaki Zorilla designed and supervised all study procedures. Paola Saura, Juan García-Virumbrales, Juan Pablo Carrasco, Miguel Pérez-Carazol, Lluç Colomer, and Javier Camacho-Rubio conducted the experimental procedures. Paola Saura and Juan García-Virumbrales wrote the first manuscript draft. All authors prepared the final manuscript.

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CONFLICT OF INTEREST STATEMENT

The authors declare no conflict of interest.

PEER REVIEW

The peer review history for this article is available at <https://www.webofscience.com/api/gateway/wos/peer-review/10.1111/cge.14393>.

DATA AVAILABILITY STATEMENT

Study data are available upon request to the authors.

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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