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Primary Care Providers' Initial Evaluation of Children with Global Developmental Delay: A Clinical Vignette Study

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Objective To examine the decisions of pediatric primary care physicians about their diagnostic evaluation for a child with suspected global developmental delay (GDD).

Study design A survey was mailed to a sample of pediatricians (n = 600) and family physicians (n = 600) randomly selected from the American Medical Association Physician Masterfile. The survey contained a clinical vignette describing a 9-month-old nondysmorphic boy with GDD. Participants were asked their initial evaluation steps (test, refer, or both test and refer) and what types of referral and/or testing they would pursue. We examined bivariate associations between physician/clinical practice characteristics and participants' evaluation decision.

Results More pediatricians than family physicians completed the survey (response rates: 55% vs 38%). Almost three-quarters of the respondents (74%) reported that their first step in a diagnostic evaluation would be to refer the child without testing, 22% would test only, and 4% would both test and refer. As their initial step, most physicians referred to a developmental pediatrician (58%), and only 5% would refer to a geneticist. The most commonly ordered test was general biochemical testing (64%). The most commonly ordered genetic test was a karyotype (39%).

Conclusions When evaluating a child with GDD, few primary care physicians would order genetic testing or refer to a genetics specialist as a first evaluation step. Future studies should examine both barriers to and utilization of a genetic evaluation for children with GDD. *(J Pediatr 2015;167:1404-8)*.

Children with global developmental delay (GDD), or significant impairment (ie, ≥2 SDs below the mean) in 2 areas of development,[1] are at increased risk for a genetic disorder. Estimates place the proportion of children with GDD who have an underlying genetic condition between 17% and 47%.[2] Pediatric primary care physicians (PCPs; eg, pediatricians and family physicians [FPs] who care for children) are likely to be the first physician to identify children with GDD during their routine practice of developmental screening. As a result, PCPs are likely to be responsible for initiating an evaluation of the causes of GDD.

There are no published data regarding how pediatric PCPs proceed with a diagnostic evaluation of a child with GDD. Professional guidelines recommend that a genetics evaluation be included in the diagnostic evaluation on children with GDD.[3-5] Whether the PCP directs the genetics evaluation or refers children to a specialist, either genetic or nongenetic, depends upon a host of factors, including PCP experience and access to specialty care.[4] Examining PCPs' practice patterns in this context can help inform policy discussions regarding genetic testing and help to ensure timely and appropriate genetics evaluations for children with GDD. To this end, we surveyed a national sample of pediatric PCPs to assess their diagnostic decisions regarding children with GDD.

Methods

From June-November 2011, we conducted a mail survey of a random sample of 600 pediatricians and 600 FPs selected from the American Medical Association Masterfile, a database of all licensed physicians in the US. The sample included allopathic and osteopathic physicians who worked in office-based, direct patient care and whose board certification and whose self-described primary specialty was either pediatrics or family medicine. Per our standard practice for conducting national physician surveys, residents, federal/military, and physicians >70 years of age were excluded.[6,7]

FP	Family physician
GDD	Global developmental delay
MRI	Magnetic resonance imaging
PCP	Primary care physician

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The first mailing was sent by regular mail and included a $5 incentive. A second mailing to nonrespondents contained no incentive but was sent by priority mail and a third mailing was sent by regular mail and contained no incentive. All mailings contained a stamped business reply mail envelope for return of the completed survey. The University of Michigan Institutional Review Board approved the study (IRBMED HUM00046913).

Survey Instrument

The survey contained a clinical vignette describing a 9-month old nondysmorphic boy with GDD:

> Imagine you are seeing a 9-month-old boy with global developmental delay (not rolling over, not reaching for objects, and not babbling) in your clinic. The child is not dysmorphic, and there is no family history of developmental delay. Assuming insurance coverage is not an issue, which of the following would be your FIRST step in evaluating this child? Circle only one.

Respondents could then choose from one of the following responses: order a test, refer patient to a specialist, or simultaneously order a test and refer patient to a specialist. We chose to identify the child as a boy to raise diagnostic consideration of an X-linked cause of the GDD, such as fragile X syndrome. Our rationale for describing the child as nondysmorphic was 2-fold. First, we wished to reflect the reality that a significant number of children with developmental delay will not have dysmorphic features.[8] Second, describing the child as dysmorphic might have biased the respondents toward using genetic services and potentially away from their routine evaluation practice for this type of child with GDD.

Outcomes

The main outcome for this analysis was the respondents' initial decision-making about how to evaluate the child: referral to a specialist, order testing, or both. We then asked respondents to identify the types of specialist referral (developmental pediatrician, geneticist, neurologist, other) or testing (brain magnetic resonance imaging [MRI], general biochemical testing, DNA-based microarray test, karyotype, biochemical testing for specific disorder, DNA-based test for specific disorder, other) that they would pursue. We based response choices on clinical experience regarding referrals made for this indication and available genetic testing options.

Independent Variables

We obtained demographic data (eg, age, sex, degree, year of graduation) from the American Medical Association Masterfile. Respondents also answered questions about their practice patterns (average number of children with GDD for whom they initiate a diagnostic workup annually) and use of genetics services (number of patients referred for a genetic evaluation, genetics clinic distance and wait times, number of genetic tests ordered).

Data Analyses

We performed univariate and bivariate analyses of the demographics and our main outcome variables. For all testing, we defined the statistical significance level as $P < .05$.

We tested the association of physician and clinical practice characteristics with the initial management decision using ANOVA, χ^2 tests, regression (simple linear and multinomial). We collapsed clinic wait time into binary outcome <2 months or ≥2 months and examined the relationship with initial management decision using multinomial regression.

For those respondents who reported that they ordered tests (whether alone or in combination with a referral to a specialist), we calculated the proportion of test types ordered, including genetic tests specifically. For this analysis, we defined the following as genetic tests: karyotype, microarray (ie, chromosomal microarray), or targeted DNA test. We then tested the association between ordering of a genetic test and type of specialist referral using logistic regression. We also described the characteristics of those respondents who would order testing alone (ie, not refer).

For those respondents who reported that they would refer to a specialist, we conducted univariate analysis regarding their referral decision. Given the number of "other" responses and the proportion of those responses that indicated referral to a developmental assessment/early intervention program, post hoc we created another category for referral to an early intervention program.

Finally, we conducted univariate analysis of the initial management decision for those respondents who used genetic services (ie, referred to geneticist and/or ordered a genetics-based test) as part of their evaluation.

Results

The response rate was 55% among pediatricians and 38% among FPs (n = 448) (**Figure 1**; available at www.jpeds.com). Pediatricians who responded to the survey were more likely to be female, medical doctors, and reported more years since medical school graduation (**Table I**). There was no difference in these demographics between eligible respondents and those who did not return survey (data not shown).

In an average year, 76% of FPs and 98% pediatricians initiated a diagnostic workup for at least 1 child with GDD. Among those respondents who reported initiating a workup,

Table I. Respondent characteristics

	Pediatricians (N = 289)	FP (N = 159)	*P* value
Mean age (y)	47.9 (SD 9.8)	46.4 (SD 9.0)	.12
Female	55.7%	44.0%	.02
Years since medical school graduation	20.6 (SD 10)	18.6 (SD 8.8)	.03
Training			
Medical doctor	96.2%	83.7%	<.001
Doctor of osteopathic medicine	3.8%	16.3%	