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;■:■-■).

Children with global developmental delay (GDD), or significant impairment (ie, ≥2 SDs below the mean) in 2 areas of development, 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%. 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. 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. 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
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. 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.

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, $\chi^2$ tests, regression (simple linear and multinomial). We collapsed clinic wait time into binary outcome <2 months or $\geq$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,
pediatricians evaluated more children with GDD than FPs (19 vs 5, \(P = .002\)).

**Initial Management Decision: Test, Refer, Test and Refer**

When faced with a clinical vignette of a 9-month-old boy with GDD, the majority of respondents (74%) reported that their first step in a diagnostic evaluation would be to refer the child without testing, and 4% reported they would test only, and 22% reported that they would both test and refer simultaneously. Pediatricians and family practitioners did not significantly differ in their responses (\(P > .05\)). There was also no difference in respondents’ initial management decision by age, degree, sex, or mean years since graduation, number of patients with GDD seen in past year, or known travel time to nearest genetics clinic. Respondents’ workup decisions were significantly associated with how long their real life patients have to wait to see a geneticist. Specifically, those respondents’ whose real life patients had to wait longer than 2 months for an appointment with a geneticist were more likely to choose to test rather than refer in the clinical scenario (\(P < .01\); Figure 2).

**Respondents Who Chose to Refer or Refer and Test**

Of the 407 respondents who reported they would refer as part of their initial management (either refer alone or both test and refer), most chose to refer to a developmental pediatrician (58%) followed by a neurologist (27%), developmental assessment program (7%), geneticist (5%), and then “other” (3%), such as a pediatrician (from FP respondents), regional/tertiary care center, occupational therapy, or physical therapy. When examined by specialty, pediatricians were less likely than FPs to refer this child to a developmental pediatrician compared with a neurologist (OR .32, 95% CI .18-.54; Figure 3).

**Respondents Who Chose to Test or Test and Refer**

Of the 108 respondents who chose to order a test as part of the initial diagnostic evaluation of the child with GDD (either by testing alone or in combination with referral), the most commonly ordered test was general biochemical testing, followed by karyotype, and then MRI (Table II). Only 51 respondents (11%) ordered a genetic test (ie, microarray, karyotype, or targeted DNA testing), and only 3 of these did so in the context of testing without a referral to a specialist.

Among those few respondents (\(n = 16\)) who reported that they would test without referring, the most commonly ordered test was a karyotype (50%) followed by an MRI (31%), and then karyotype and “other” tests (eg, blood chemistries, complete blood count, urine test, thyroid stimulating hormone, developmental screening test; 19%, respectively). The number of respondents who chose to test alone was too small to permit a comparison with those respondents who would test and refer.

**Respondents Who Used Genetic Services**

Among all the 448 respondents, only 61 (14%) used genetic services (either order genetic test, refer to geneticist, or both) as part of their initial diagnostic evaluation of a child with GDD. Of these 61 respondents, 3 ordered genetic tests, 8 referred to a geneticist, and 50 used either of these genetic services in combination with nongenetic services. Of those who referred in combination with genetic testing, most did not refer to a geneticist: 46% referred to a developmental specialist, 28% to a neurologist, and 22% to a geneticist.
We found that when presented with a clinical vignette of a child with GDD, for their initial evaluation step most pediatric PCPs would not do testing, genetic or otherwise, and would instead refer the child. They would refer most often to a developmental pediatrician or a developmental intervention program (eg, Early On or First Steps), sometimes to a neurologist, and infrequently to a geneticist. Contrary to concerns that PCPs may be ordering genetic tests without proper training, very few PCPs would order a genetic test (11%) and of those, the most common was a karyotype. These findings address the concern that when offered the opportunity, a substantial number of pediatric PCPs will order genetic testing without consultation from a genetics specialist as part of their initial workup. In contrast, we found that even those respondents who would order a genetic test were also more likely to refer to a geneticist. It should be noted that our survey was fielded around the time that recommendations for use of chromosomal microarrays as a first line test in evaluation of children with GDD were released. In part, this might explain the relatively low utilization of genetic tests among providers.

By contrast, the test most frequently ordered by PCPs was general biochemical testing. Recent recommendations from the American Academy of Pediatrics suggest considering the use of metabolic testing in the evaluation of these patients given its wide availability and low cost. In our vignette, we did not provide history or examination findings to suggest a metabolic disorder. The fact that many providers chose this test may indicate their familiarity with such through experiences with patients through newborn screening. Given that studies have shown the yield of metabolic testing in children with GDD range from 0.2%-4.6%, this finding suggests the need for further education of PCPs about the evaluation of children with GDD.

What may be most concerning is that our findings suggest that despite a child with GDD being at increased risk for having a genetic disorder, accessing genetics services from a genetics provider were not at the forefront of pediatric PCPs’ considerations. There are a number of potential explanations for these findings. First, the choice of referring initially to a developmental pediatrician or neurologist may indicate a preference among PCPs to have a specialist offer a therapeutic intervention as well as a diagnostic evaluation. In addition, it may be a more easily accessible option by PCPs given the acknowledged relative shortage of geneticists and the slowing pipeline of genetics trainees. Although it may be optimal for GDD to undergo a genetic evaluation by a clinical geneticist, it may not be practical given location and availability of genetics clinic, workforce issues in genetics, and insurance coverage of testing. Our findings that longer wait times for an appointment with a geneticist in practice were associated with a decrease in likelihood to refer in the clinical scenario support this hypothesis and lend credence to the generalizability of our findings to practice. It should be noted, however, that the workforce of developmental-behavioral pediatrics is not much more robust than that of geneticists, with only 580 certified developmental-behavioral providers in 2012.

Second, we constructed our vignette in order not to bias the reviewers toward a specific course of action. This is not unrealistic from a clinical perspective given that 20% of children referred to a developmental or neurology clinic did not have a history or physical examination finding suggestive of a diagnosis (eg, dysmorphic features, prenatal/perinatal history of toxin exposure, psychosocial history of neglect, growth/feeding history, family history, age). However, we acknowledge that the absence of such history/physical examination findings may have biased the responders to be more cautious in their testing and referrals regarding genetics services and that their actions may represent their intent to better assess the phenotype in what they viewed as a logical, cost-efficient first step. Along these lines, the fact that 10% of respondents chose to refer to developmental assessment programs may reflect either the need to better define the phenotype or the wider availability of these programs within communities. Future studies should assess the influence of history and physical examination findings on referral and testing patterns.

Third, although we said insurance is not an issue, it is possible that difficulty with genetic testing coverage and payments may affect their daily practice and influence their responses. In addition, the time involved in precertifying genetic testing for the third party payer is time consuming and adds a layer of more complexity to already busy practices. Finally, this study was conducted close to the release of professional organizational guidelines recommending the use of chromosomal microarrays as a first line test for children with GDD. It is possible that this explains the low proportion of genomic testing, including chromosomal microarray, which we saw among respondents. However, given previous studies on clinicians’ poor adherence to guidelines and the fact that pediatric PCPs may not be aware of specialty organization guidelines, we would not expect a significant change in our findings after the release of the guidelines.

Our study has limitations that merit discussion. First, we examined responses to a clinical vignette. However, research...
has shown that physician responses to vignettes better reflect actual practice than chart abstractions.15 Although the resulting data may not be directly generalizable to every clinical case of GDD, our findings still provide helpful insights into how physicians likely act when faced with a general case of GDD in their practice. We also acknowledge that referral and testing patterns may be multifaceted and influenced by additional factors, such as institutional culture and training, which we did not measure in this study and patient characteristics (eg, physical exam, history) as previously mentioned.

In addition, we only asked respondents to provide us with their initial action in the diagnostic process. Although it is possible that they might refer to genetics at a later point in time or make multiple referrals simultaneously, we could not capture that action in this study. We did not allow respondents to indicate multiple simultaneous options because we were concerned that would encourage respondents to choose more options than they would in real life simply to appear to be more “comprehensive.” Instead, our survey allows us to identify the preferred referral among those respondents who might engage in multiple referrals. We acknowledge that this methodology may have led some respondents to have a more cautionary attitude resulting from evidence-based and cost-efficient management practices when faced with limited information and response options. In addition, this methodology limits us from fully addressing possible obstacles toward genetic evaluation in children with global development delay. Additional studies are needed to determine if, given unlimited options, PCPs would refer children with GDD for a genetics evaluation in addition to other evaluations. However, our findings do point out that when faced with limited options for an initial referral, PCPs are not inclined to refer children with GDD for a genetics evaluation.

Finally, although it is possible that the location and local practice patterns may influence diagnostic evaluation choices, we were unable to assess this because we did not have practice specific zip codes or knowledge of the providers’ clinical context and cultural influences. Despite these limitations, our study clarifies that, when evaluating a child with GDD, few PCPs would order genetic testing or refer to a genetics specialist as a first step in their evaluation. Whether or not this means that a child with GDD would ultimately receive a genetic evaluation is unclear and beyond this scope of this study. However, given their elevated risk of having a genetic condition, the extent to which children with GDD undergo an appropriate genetic evaluation, regardless of the context, is an important area for future exploration.

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References

Figure 1. Response flow chart.