Variability in Cerebral Palsy Diagnosis

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abstract

BACKGROUND: Cerebral palsy (CP) is the most common childhood motor disability. The emergence of genetic CP etiologies, variable inclusion of hypotonic CP in international registries, and involvement of different medical disciplines in CP diagnosis can promote diagnostic variability. This variability could adversely affect patients' understanding of their symptoms and access to care. Therefore, we sought to determine the presence and extent of practice variability in CP diagnosis.

METHODS: We surveyed physicians in the United States and Canada interested in CP on the basis of membership in the American Academy of Cerebral Palsy and Developmental Medicine or the Child Neurology Society Neonatal Neurology, Movement Disorders, or Neurodevelopmental Disabilities Special Interest Groups. The survey included the 2007 consensus definition of CP and 4 hypothetical case scenarios.

RESULTS: Of 695 contacted physicians, 330 (47%) completed the survey. Two scenarios yielded consensus: (1) nonprogressive spastic diplegia after premature birth with periventricular leukomalacia on brain MRI (96% would diagnose CP) and (2) progressive spastic diplegia (92% would not diagnose CP). Scenarios featuring genetic etiologies or hypotonia as the cause of nonprogressive motor disability yielded variability: only 46% to 67% of practitioners would diagnose CP in these settings.

CONCLUSIONS: There is practice variability in whether a child with a nonprogressive motor disability due to a genetic etiology or generalized hypotonia will be diagnosed with CP. This variability occurred despite anchoring questions with the 2007 consensus definition of CP. On the basis of these results, we have suggested ways to reduce diagnostic variability, including clarification of the consensus definition.

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WHAT'S KNOWN ON THIS SUBJECT: Cerebral palsy (CP) diagnostic practice variability has not been assessed but could exist because of the involvement of multiple medical disciplines in CP care, emerging genetic etiologies for CP, and/or variable inclusion of hypotonic CP in international registries.

WHAT THIS STUDY ADDS: Physician experts variably diagnose CP in the setting of genetic etiologies or hypotonia, which can contribute to inconsistent prognostication, management, and understanding of patients' medical conditions. Clarification of the CP consensus definition may help reduce diagnostic practice variability.

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Cerebral palsy (CP) is often considered to be the most common cause of motor disability in childhood, affecting 3 of every 1000 children.[1–3] Given its prevalence, CP diagnostic practice variability would have widespread implications. Potential contributing factors to diagnostic variability include the increasing number of genetic etiologies associated with a CP phenotype,[1,4,5] somewhat variable consideration of hypotonia as a CP phenotype in international registries,[6–8] involvement of multiple specialties in CP care,[9] the demonstrated value of diagnosing CP as early as possible,[10] the variable need for a CP diagnosis to gain consistent access to treatment services, and caregiver perceptions of a CP diagnosis. The most recent consensus definition of CP states that CP "is not an etiologic diagnosis, but a clinical descriptive term" without explicit description of whether motor disability predominantly due to hypotonia can constitute CP.[11] In light of the above factors, it is unclear whether this definition is interpreted uniformly between practitioners.

We hypothesized that there may be CP diagnostic practice variability between specialties, particularly regarding genetic etiologies and hypotonia as the cause of nonprogressive motor disability. Determining areas of diagnostic controversy could guide efforts to mitigate practice variability. Therefore, our objective was to survey the CP diagnostic practices of physicians in the United States and Canada who are most likely to diagnose children with CP. This investigation marks a key step toward ensuring a consistent and accurate CP diagnosis.

METHODS

This study was granted human subjects research exemption from the Washington University School of Medicine in St Louis Institutional Review Board.

We conducted a cross-sectional survey of physicians practicing in the United States and Canada who were members of the American Academy of Cerebral Palsy and Developmental Medicine (AACPDM) or the Child Neurology Society Neonatal Neurology, Movement Disorders, or Neurodevelopmental Disabilities Special Interest Groups. Members were e-mailed between September 1, 2019, and December 15, 2019, with a link to the Research Electronic Data Capture survey.

Survey development occurred via a modified Delphi process[12–14] with input from physicians and nonphysician parents of children with CP who were members of the Cerebral Palsy Research Network.[15,16] The survey contained (1) questions about respondent training and expertise, (2) the 2007 consensus definition of CP,[11] and (3) 4 hypothetical case scenarios (Table 1, Methods section of the Supplemental Information). Scenarios provided a set of hypothetical patients across which all respondents' diagnostic determinations could be comparably judged. The first 3 scenarios depicted a common CP phenotype: spastic diplegia in a 5-year-old boy after premature birth with periventricular leukomalacia on MRI. The spastic diplegia was nonprogressive in scenario 1, progressive in scenario 2, and nonprogressive but associated with a genetic etiology in scenario 3. Scenario 4 depicted a 5-year-old boy born at term gestation with a normal MRI, generalized hypotonia causing

TABLE 1 Hypothetical Case Scenarios Presented in the Survey

	Scenario Description
Shared Information for Scenarios 1–3	A 5-y-old boy presents to you for walking difficulties. He was born at 30 wk gestation after an unremarkable pregnancy, with labor precipitated by preterm premature rupture of membranes. He had a 7-wk NICU stay primarily to manage feeding immaturity. A brain MRI done at 4 y old revealed bilateral periventricular T2 hyperintensities.
Scenario 1	This child has had gross motor developmental delays but no regression and has gradually gained milestones. He began walking at 2 y old and has always used a walker to ambulate. He has spasticity and hyperreflexia in both legs on your examination.
Scenario 2	This child was walking normally and attaining age-appropriate developmental milestones until he turned 4 y old. Since then, he has had progressively increasing tone in his legs such that he now has to use a walker to ambulate. He has spasticity and hyperreflexia in both legs on your examination.
Scenario 3	The child has had speech and gross motor developmental delays but no regression and has gradually gained milestones. He is able to understand simple commands and is nonverbal but communicates with some sign language. He began walking at 2 y old and has always used a walker to ambulate. He has spasticity and hyperreflexia in both legs on your examination. His older brother has a similar phenotype with no history of prematurity. Given this, genetic testing was done, revealing that both brothers carry biallelic pathogenic mutations in *ADD3*, which has a known association with nonprogressive spasticity and intellectual disability.
Scenario 4	A 5-y-old boy presents to you for walking difficulties. He was born at 40 wk gestation after an unremarkable pregnancy and delivery. He also has a history of epilepsy. He has global developmental delay but no regression and has gradually gained milestones. He can ambulate short distances using a walker but primarily uses a wheelchair. He has diffusely low tone and hyporeflexia on your examination. A brain MRI done at 4 y old was normal. He was found to have a chromosome 1q microdeletion that has a known association with nonprogressive motor symptoms.

motor disability, and a genetic etiology for his symptoms. After each scenario, respondents were asked, "Is it possible to make a diagnosis of CP in this child at this time?" "No" responses prompted open-ended explanations of respondent rationales. Respondents were excluded if they did not respond to all 4 case scenarios.

Statistical analyses were undertaken by using SPSS (IBM SPSS Statistics, IBM Corporation, Armonk, NY) and R (R Core Team, R Foundation for Statistical Computing, Vienna, Austria). Logistic regression was used to determine which variables affected the odds of diagnosing CP in each scenario. Independent model variables were medical specialty (grouped on the basis of specialty boarding status as surgery, physical medicine and rehabilitation, neurology, or pediatrics, with self-reported subspecialties noted in Table 2), subspecialization status (yes, with fellowship training; yes, without fellowship training; or no), years in practice (<6, 6–10, or >10 years), practice setting (inpatient, outpatient, or both), practice affiliation (academic, private, or both), age of patients (children younger than 18 years, adults 18 years or older, or both), and percentage of patients with a nonprogressive motor phenotype (<25%, 25%–50%, 50%–75%, and >75%). We hypothesized that these variables might independently contribute to diagnostic practice variability. The χ^2 statistic and Wald test were used to determine the significance of the model and model terms, respectively. Adjusted odds ratios (ORs) and 95% confidence intervals (CIs) were calculated for each variable. The Hosmer-Lemeshow test was used to estimate goodness of model fit. The area under the receiver operator characteristic curve (AUC) was used to assess the model's predictive ability. Significance levels were set a priori at $P < .05$.

RESULTS

Survey Response Rate, Participant Numbers, and Participant Demographics

A total of 695 physician members of the AACPDM and relevant Child Neurology Society Special Interest Groups were contacted. Of these, 390 responded (55% overall response rate), with 330 responding to all 4 case scenarios (47% completed survey response rate). Responses were anonymous to encourage participation, and, accordingly, the demographic characterization of nonresponders is difficult. E-mails declining to complete the survey included statements such as the following: "Not practicing, retired," "in full retirement, I don't believe I am now prepared to have a meaningful opinion about CP," and "I am currently not seeing patients." The completed survey response rate was 32% for surgeons (n = 52 of 161), 43% for physical medicine and rehabilitation specialists (n = 89 of 209), 59% for neurologists (n = 129 of 219), and 57% for pediatricians (n = 60 of 106) (Tables 2 and 3).

Respondents tended to be subspecialists (Table 2) with fellowship training in practice for >10 years who see primarily pediatric patients in an academic outpatient or mixed inpatient and outpatient setting (Table 3).

Case Scenario Characteristics Contributing to Diagnostic Practice Variability

Scenarios 1 and 2 yielded diagnostic consensus. Most respondents (96%) would diagnose CP in a child with nonprogressive spastic diplegia (scenario 1). The 4% of respondents who would not diagnose CP in scenario 1 cited the need to rule out other spastic diplegia etiologies (including spinal cord lesions) and the need for reviewing the brain imaging to confirm the presence of periventricular leukomalacia. Most respondents (92%) would not diagnose CP in a child with progressive spastic diplegia (scenario 2), with 91% explicitly noting that phenotypic progression precludes CP diagnosis (Table 4).

Scenarios 3 and 4 uncovered diagnostic practice variability. Sixty-seven percent of physicians would diagnose CP in a child with nonprogressive spastic diplegia due to a genetic etiology (scenario 3), whereas fewer (46%) would diagnose CP in a child with nonprogressive hypotonia due to a genetic etiology (scenario 4).

The most cited reason for not diagnosing CP was the presence of a genetic diagnosis (n = 88 of 330 respondents for scenario 3 and 107 of 330 respondents for scenario 4; 131 of 330 respondents overall) (Table 4, Supplemental Table 7):

- "If it's genetic, there is likely a progressive component."
- "Would you diagnose the kid with Down syndrome with hypotonic CP?"
- "I hesitate [because] I believe CP has a non-genetic connotation to some."
- "Underlying genetic dx [diagnosis] with CP phenotype."

The second most cited reason for not diagnosing CP was the presence of isolated hypotonia (n = 39 of 330 respondents for scenario 4):

- "Could still have nmd [a neuromuscular disorder]."
- "If he were ataxic or dyskinetic, that would change things."
- "Hypotonic CP is an old-fashioned diagnosis."
- "Depends on [the] definition of CP; if genetic causes/hypotonia are considered CP, then yes."

When explaining why they would not diagnose CP in scenarios 3 and 4, some respondents noted the practical benefit of a CP diagnosis (n = 7 of 330 respondents for scenario 3 and 8 of

TABLE 2 Respondent Specialties

Specialties and Subspecialties	No. Respondents[a]	Overall (N = 330), %	Percentage of Each Specialty, %
Surgery	52	16	100
Orthopedic surgery	51	15	98
Pediatrics	36	11	69
Neuromuscular disorders	1	0.3	2
No subspecialty indicated	14	4	27
Neurosurgery	1	0.3	2
Physical medicine and rehabilitation	89	27	100
Pediatrics	76	23	85
Electrodiagnostic medicine	3	1	3
Brain injury	2	1	2
Sports	1	0	1
No subspecialty indicated	13	4	15
Neurology	129	39	100
Neurodevelopmental medicine	19	6	15
Epilepsy	22	7	17
Clinical neurophysiology	7	2	5
Neonatal	6	2	5
Movement disorders	6	2	5
Genetics	5	2	4
Neuromuscular disorders	4	1	3
Headache	4	1	3
Neuroimmunology	2	1	2
Neurooncology	2	1	2
Stroke	1	0.3	1
Metabolic disorders	1	0.3	1
Neuropathology	1	0.3	1
Sleep	1	0.3	1
Brain injury	1	0.3	1
No subspecialty indicated	51	15	40
Pediatrics	60	18	100
Developmental pediatrics	30	9	50
Neonatology	3	1	5
Rehabilitation medicine	2	1	3
Orthopedics	1	0.3	2
Adolescent medicine	1	0.3	2
Internal medicine	1	0.3	2
No subspecialty indicated	22	7	37

[a] Some respondents noted >1 subspecialty.

330 respondents for scenario 4; 14/330 respondents overall):

- "I have given [a] CP hypotonia diagnosis - just to be able to get services."
- "He has CP secondary to X genetic disorder. But I would only do that for the school. Otherwise, the diagnosis is X genetic disorder."
- "For ease of understanding by schools, etc. I will often add the CP diagnosis."
- "For insurance purposes, I would diagnose CP."

In the Results section of the Supplemental Information, we list all respondent quotes considered to be representative of each of these 3 response categories.

Respondent Characteristics Contributing to the Odds of Making a CP Diagnosis

Physician specialty, subspecialization status, years in practice, practice setting, practice affiliation, age of patients treated, and percentage of patients with a nonprogressive motor phenotype who were treated were not significant predictors of CP diagnostic practices for scenarios 1, 2, or 3 (Table 5).

In the setting of a nonprogressive motor disability due to generalized hypotonia with a genetic etiology (scenario 4), a model that included all the above variables significantly improved the likelihood of correctly predicting respondents' diagnostic determinations from 52% in the null model to 69% (P = .008), with good model fit (Hosmer-Lemeshow P = .783) and predictive ability (AUC = 0.771). Only physician specialty was a significant predictor of diagnostic practices when adjusted for the other variables (P = .008). Surgeons (OR 0.56; 95% CI 0.10–3.2) and physical

TABLE 3 Respondent Training, Experience, and Practice Characteristics

Characteristic	No. Respondents	%[a]
Specialty (*N* = 330)		
Surgery	52	16
Physical medicine and rehabilitation	89	27
Neurology	129	39
Pediatrics	60	18
Subspecialty (*N* = 330)		
Yes, with fellowship	185	56
Yes, without fellowship	73	22
No	72	22
Years in practice (*N* = 330)		
<5	88	27
6–10	32	10
>10	210	64
Practice setting (*N* = 218)		
Inpatient	18	8
Outpatient	107	49
Both	93	43
Practice affiliation (*N* = 236)		
Academic	206	87
Private	24	10
Both	6	3
Patient age (*N* = 328)		
Children, <18 y old	275	84
Adult, ≥18 y old	5	2
Both	48	15
Patients with nonprogressive motor disability, % (*N* = 330)		
<25	121	37
25–50	73	22
50–75	73	22
>75	63	19

[a] Percentage is calculated relative to the total number of respondents for the indicated question.

medicine and rehabilitation specialists (OR 0.93; 95% CI 0.26–3.4) were comparable to pediatricians regarding odds of diagnosing CP in scenario 4. However, neurologists were 4.9 times more likely (95% CI 1.3–18) to diagnose CP compared with pediatricians (Table 5).

Response rates for practice setting and practice affiliation were lower (218–236 respondents) than response rates for all other variables (328–330 respondents) (Table 3). A logistic regression reanalysis that excluded these 2 variables did not change the relationships described above but resulted in lower predictive ability (AUC = 0.664) than the model that included all variables (Supplemental Table 7).

DISCUSSION

These survey results reveal CP diagnostic variability among experts in the setting of a genetic etiology and/or hypotonic phenotype. This variability is partially determined by physician specialty, with neurologists more likely to diagnose CP in the setting of nonprogressive hypotonia with a genetic etiology.

More than 90% of respondents agreed that a nonprogressive motor disability associated with hypertonia after an isolated injury to the developing brain constitutes CP (scenario 1). More than 90% of respondents also agreed that a progressive motor disability excludes a CP diagnosis (scenario 2). It can be difficult to distinguish between motor phenotype progression,[17] which rules out a CP diagnosis,[11] and motor function decline, which is commonly seen in CP.[18] These subtleties may be reflected in the lack of unanimous diagnostic consensus for scenarios 1 and 2.

Approximately 40% of respondents cited a genetic etiology as a reason to not diagnose CP. This is despite the 2007 consensus definition noting that CP is a phenotypic and not etiologic diagnosis[11] and a 2019 international consensus statement stating that "genetic or other causation should not change the clinical diagnosis of cerebral palsy."[19]

Approximately 12% of all respondents cited isolated hypotonia as a reason to not diagnose CP. Australian CP registries include

TABLE 4 Responses Regarding CP Diagnosis in 4 Hypothetical Patient Scenarios

Scenarios[a] With Responses and Rationale[b]	No. Respondents	%
S1: Nonprogressive spastic diplegia after premature birth		
Would diagnose CP	316	96
Would not diagnose CP	14	4
R1: additional imaging required	9	3
R2: alternate diagnosis offered	4	1
S2: Progressive spastic diplegia		
Would diagnose CP	25	8
Would not diagnose CP	305	92
R1: progressive phenotype	278	84
R2: alternate diagnosis offered	26	8
S3: Nonprogressive spastic diplegia with genetic etiology		
Would diagnose CP	220	67
Would not diagnose CP	110	33
R1: genetic	88	27
R2: practical diagnosis[c]	7	2
S4: Nonprogressive hypotonia with genetic etiology		
Would diagnose CP	153	46
Would not diagnose CP	177	54
R1: genetic	107	32
R2: hypotonia	39	12
R3: practical diagnosis	8	2

[a] Each scenario (S) presented in the survey (Table 1).

[b] The respondent provided rationale (R) for not diagnosing CP in each scenario.

[c] The respondent indicated they would not typically provide a diagnosis of CP but might consider providing one if it served a practical purpose for the patient (eg, explaining the diagnosis to the school district or gaining access to services).

children with "generalized hypotonia not attributable to cognitive deficiencies."[8] In contrast, European registries specify that hypotonia alone is insufficient to diagnose CP.[6,7] This discrepancy is remarkable because much of our epidemiological knowledge about CP is from Australian and European registries.[20–27]

Limitations

We surveyed content experts (physician members of organizations demonstrating an interest in CP) expected to have the greatest familiarity with CP diagnostic consensus statements. Therefore, we suspect that the practice variability among this more homogenous group of surveyed physicians is an underestimation of the practice variability that may exist overall. Reaching out to physicians via other professional organizations could be a valuable area of future research.

Our chosen survey population does not include the nonphysician practitioners who are essential for CP care. Although our original intention was to survey these practitioners (who make up 37% of AACPDM membership in the United States and Canada) (Methods section of the Supplemental Information), correspondence from many indicated discomfort with being primarily responsible for making a CP diagnosis. Therefore, their views, although critical regarding CP management, were not considered here concerning CP diagnosis.

We developed hypothetical scenarios to mimic clinical scenarios that we hypothesized would yield diagnostic consensus (scenarios 1 and 2) or diagnostic practice variability (scenarios 3 and 4). This allowed for controlled comparisons of diagnostic practices across respondents but serves only as an analog of real-world diagnostic practices. For example, some physicians who stated they would not diagnose CP in the setting of a genetic etiology or hypotonia in our survey may diagnose CP for its practical benefit in the real world.

Our completed survey response rate was 47%. Respondent anonymity was necessary to ensure respondents felt comfortable answering questions that could be interpreted as tests of clinical acumen. This anonymity precludes a detailed analysis of nonresponders. However, on the basis of isolated e-mail responses to our survey participation request, nonresponders may have been more likely to feel currently unqualified to provide an opinion on CP diagnosis. The relatively lower response rates for surgeons and physical medicine and rehabilitation specialists compared with pediatricians and neurologists could highlight a difference in specialty focus: surgeons and physical medicine and rehabilitation specialists may more typically have patients referred for management after already receiving a CP diagnosis.

Despite these limitations, the results reveal a clear lack of consensus in CP diagnosis in the setting of nonprogressive motor disability associated with genetic etiologies

TABLE 5 Respondent Characteristic Possibly Contributing to the Odds of Diagnosing CP in 4 Hypothetical Patient Scenarios

Respondent Characteristic Possibly Contributing to Odds of Diagnosing CP[a]	OR[b] (95% CI) Relative to the Reference Value for Each Characteristic			
	Scenario 1	Scenario 2	Scenario 3	Scenario 4[c]
Specialty				
Surgery	0 (—)	1.453 (0.088–23.893)	0.418 (0.076–2.304)	0.564 (0.100–3.173)
Physical medicine and rehabilitation	0 (—)	0.057 (0.003–1.123)	0.485 (0.120–1.965)	0.930 (0.256–3.377)
Neurology	0 (—)	0.040 (0.001–1.121)	1.400 (0.333–5.887)	4.947 (1.333–18.365) *
Pediatrics	Reference[d]	Reference	Reference	Reference
Subspecialty				
Yes, with fellowship	0 (—)	5.341 (0.208–137.194)	0.275 (0.072–1.055)	2.379 (0.711–7.960)
Yes, without fellowship	0 (—)	27.253 (0.547–1358.989)	0.241 (0.048–1.201)	2.370 (0.562–10.000)
No	Reference	Reference	Reference	Reference
Years in practice				
<5	0.823 (0.076–8.915)	5.017 (0.677–37.165)	0.756 (0.297–1.923)	1.628 (0.647–4.093)
6–10	0.488 (0.023–10.569)	0 (—)	0.282 (0.056–1.425)	0.602 (0.109–3.130)
>10	Reference	Reference	Reference	Reference
Practice setting				
Inpatient	0.204 (0.003–12.646)	0 (—)	0.251 (0.038–1.655)	0.127 (0.016–1.022)
Outpatient	6.595e7 (—)	0.288 (0.030–2.784)	0.817 (0.297–2.243)	0.548 (0.205–1.462)
Both	Reference	Reference	Reference	Reference
Practice affiliation				
Academic	0.858 (0.017–43.333)	0.255 (0.007–9.253)	3.028 (0.343–26.757)	1.346 (0.118–15.326)
Private	9.147e7 (—)	74.449 (0.784–7066.016)	5.432 (0.333–88.552)	2.265 (0.135–37.950)
Both	Reference	Reference	Reference	Reference
Age of patients seen				
Children (<18 y)	1.336 (0.104–17.098)	3.839 (0.153–96.017)	2.256 (0.699–7.275)	2.82 (0.762–10.437)
Adult (≥18 y)	2.968 (—)	0 (—)	1.386 (0.059–32.624)	0 (—)
Both	Reference	Reference	Reference	Reference
Percentage of patients with nonprogressive motor disability, %				
<25	0.274 (0.011–7.127)	0.652 (0.040–10.695)	0.511 (0.137–1.908)	1.361 (0.362–5.115)
25–50	6.013e7 (—)	0.423 (0.034–5.284)	1.063 (0.270–4.177)	0.828 (0.217–3.157)
50–75	0.504 (0.037–6.803)	0.766 (0.067–8.813)	1.527 (0.457–5.097)	0.993 (0.304–3.245)
>75	Reference	Reference	Reference	Reference

—, not applicable.

[a] Logistic regression analysis was used to determine if any of the surveyed respondent characteristics could contribute to the odds of diagnosing CP in each of the 4 hypothetical case scenarios (separate logistic regression model for each scenario).

[b] ORs are indicated for each respondent characteristic relative to the indicated reference value.

[c] Only the overall logistic regression model for scenario 4 significantly predicted the likelihood that a respondent would diagnose CP on the basis of the indicated respondent demographics (χ^2 statistic, $P = .008$). Of the examined demographics, only the respondent's medical specialty was a significant predictor of whether a respondent would diagnose CP in scenario 4, with neurologists almost 5 times more likely to diagnose CP than pediatricians.

[d] These ORs are adjusted for all of the indicated respondent characteristics for each scenario.

* $P = .017$ (Wald test).

and/or hypotonia. This lack of consensus should be addressed to mitigate the deleterious effects of diagnostic practice variability.

Combining a CP Diagnosis With Identification of CP Etiology: A Possible Strategy to Reduce Diagnostic Practice Variability

Practice variability in CP diagnosis may cause confusion and distress for the family.[28] As noted by some respondents, a CP diagnosis may be critical (1) for access to necessary therapy and support services, (2) for providing families with a framework to explain their child's symptoms to other laypersons, and (3) for access to a community of people with similar symptoms, which is often critical for caregiver well-being.[29] Therefore, excluding a CP diagnosis in favor of a narrower etiologic descriptor could be detrimental for children who otherwise phenotypically meet the 2007 consensus CP diagnostic criteria.

However, there is definite value in defining the specific etiologies (eg, periventricular leukomalacia) and etiologic risk factors (eg, prematurity) for any child with a CP phenotype,

noting that the phenotype may provide clues regarding the etiology.[30,31] This precision can provide additional information useful for prognosis, treatment, and family planning. Because CP is "a clinical descriptive term" and "not an etiologic diagnosis,"[11] the most thorough approach may be to both assign a diagnosis of CP (representing the clinical phenotype) and clearly state the etiologies and etiologic risk factors for CP (Table 6).

Combining a phenotypic diagnosis with identification of the etiology is an approach common to other neurologic disorders, such as autism, intellectual disability, and epilepsy.[32–34] It is widely accepted that these disorders are diagnosed on the basis of clinical phenotype but have many known etiologies, genetic and otherwise. This reflects the multiple ways via which abnormalities in the developing brain can lead to a common clinical presentation. On the basis of the 2007 consensus definition, CP should also be diagnosed with this well-established framework in mind.[11,35]

We did not explicitly assess respondent familiarity with consensus statements defining CP. However, the 2007 consensus definition of CP[11] was provided within the body of the survey. Therefore, a lack of awareness of this consensus definition is unlikely to be the main contributor to the diagnostic variability observed for scenarios 3 and 4. In addition to practitioner education and discussion regarding the optimal way the consensus definition should be interpreted, it may be additionally valuable to clarify the 2007 consensus definition to explicitly address the diagnostic controversy we have highlighted surrounding genetic etiologies and hypotonia (Table 6).[11]

Some respondents noted difficulty conceptualizing genetic etiologies as nonprogressive. This highlights how "non-progressive disturbances that occurred in the developing fetal or infant brain"[11] may be conceptually straightforward for temporally isolated injuries (such as hypoxic-ischemic encephalopathy) but conceptually murky for genetic CP etiologies for which potential lifelong neurologic effects may be currently unknown. There is growing evidence of genetic risk factors, associated copy number variants, and monogenic etiologies of true nonprogressive CP phenotypes.[4,5] These should be distinguished from genetic disorders that may mimic CP in early life but eventually yield progressive or neurodegenerative disease.[17] Therefore, it may be worth explicitly noting in the consensus definition that genetic etiologies can confer a CP phenotype.

Given diagnostic practice variability regarding children with hypotonia, the field must come to a consensus across clinical practice and patient registries regarding whether generalized hypotonia due to a central etiology and resulting in motor disability should be explicitly included as a CP phenotype.[8] Inclusion of hypotonia could address the aforementioned practical values of a CP diagnosis, but could broaden the CP diagnostic sphere to include etiologies previously not considered to be CP (eg, Down syndrome or developmental coordination disorder).[8,36–38] To this end, to be considered a CP phenotype, it may be worth noting whether motor dysfunction from hypotonia should be disproportionately greater than the degree of cognitive impairment. Given the discrepancy between international CP registries regarding inclusion or exclusion of hypotonia,[6–8] addressing hypotonic CP explicitly in the CP consensus definition could be paradigm shifting. Therefore, together with reinforcing the validity of genetic CP etiologies, codifying whether hypotonia can be a CP phenotype may warrant a formal effort to revise the 2007 consensus definition of CP.

In summary, in addition to incorporating validated functional systems that are widely accepted parts of the CP diagnostic framework (such as the Gross Motor Function Classification System)[39–43] and delineating the known conditions that often coexist with CP (such as intellectual disability, epilepsy, and autism spectrum disorder),[35,44] we propose that children with CP may be best served if physicians specify the following features when making a CP diagnosis: types of tone

TABLE 6 Suggestions for Areas of Emphasis or Clarification in the 2007 International Consensus Definition of CP

	Suggestions
Emphasize	CP "is not an etiologic diagnosis, but a clinical descriptive term."[11]
	CP can exist as a diagnosis separate from definition of its etiology. It may be ideal for children to both carry a CP diagnosis and have specification of the etiology. Factors that could be explicitly stated for any child who meets clinical criteria for a CP diagnosis include the following: types of motor abnormalities (eg, spasticity, dystonia, hypotonia, ataxia, chorea), regions of the body affected by each type of motor abnormality (eg, spastic diplegia with axial hypotonia), and etiologies and etiologic risk factors for CP, noting that multiple may exist.
	Specify the known coexisting conditions for each child with CP (eg, intellectual disability, epilepsy, and autism spectrum disorder).
Clarify	Can CP be diagnosed in the setting of any nonprogressive motor disability regardless of the etiology (including genetic or other nonacquired etiologies)?
	Can CP be diagnosed in children with pure hypotonia?

abnormalities, distribution of tone abnormalities, and putative etiologies and etiologic risk factors for the CP phenotype. This description allows for the explicit provision of a CP diagnosis combined with etiologic clarity, including a definition of what tone abnormalities (potentially including hypotonia) may be a part of the CP phenotype.

CONCLUSIONS

This study highlights CP diagnostic practice variability among 330 physicians in the United States and Canada from diverse medical training backgrounds who are all interested in CP. This diagnostic practice variability, notably regarding children with generalized hypotonia and genetic etiologies of a CP phenotype, could inform a larger discussion regarding clarification of the 2007 consensus CP diagnostic criteria (Table 6). Next steps should involve surveying the field regarding how to best address this practice variability (eg, via practitioner education on uniform interpretation of the 2007 consensus definition), which will delineate the potential need to organize a formal effort to revise the 2007 consensus definition of CP.

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ABBREVIATIONS

AACPDM: American Academy of Cerebral Palsy and Developmental Medicine
AUC: area under the receiver operator characteristic curve
CI: confidence interval
CP: cerebral palsy
OR: odds ratio

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