Diagnosing cerebral palsy in full-term infants

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3 Key Points

- Cerebral palsy is more common in full-term than preterm infants and can be challenging to diagnose in infants without pronounced risk factors
- Early diagnosis and intervention improve outcomes, especially for those with mild cerebral palsy
- There is increasing evidence of a genetic contribution

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Abstract

More than 50 % of infants with cerebral palsy (CP) are born at or near term, with the vast majority having pre- or perinatally-acquired CP. While some have a clinical history predictive of CP such as neonatal encephalopathy or neonatal stroke, others have no readily identifiable risk factors. Paediatricians are often required to discriminate generalised motor delay from a variety of other diagnoses including cerebral palsy. This paper outlines known causal pathways to CP in term-born infants with a focus on differential diagnosis. Early and accurate diagnosis is important as it allows prompt access to early intervention during the critical periods of brain development. A combination of clinical history taking, standard clinical examination, neuroimaging, and genetic testing should be started at the time of referral. Attention to the investigation of common co-morbidities of CP including feeding and sleep difficulties and referral to early intervention is recommended.

Introduction

Cerebral palsy (CP) is the most common physical disability of childhood and is a heterogeneous condition resulting from damage to the developing brain. The causal pathways to CP are many, and not all are known. However, the consensus definition describes CP as a group of disorders of movement due to a non-progressive lesion/abnormality that occurred in the developing brain¹.

The prevalence of CP in Australia is 1 in 500 live births and data from the Australian Cerebral Palsy Register (ACPR) indicates that the rate of cerebral palsy appears to be declining. Furthermore, there are fewer children with severe motor impairment and co-occurring intellectual disability or epilepsy². Most cerebral palsy originates in the pre- or perinatal period with only 8% of cases attributed to post-natal causes. In fact, 57% of children with cerebral palsy are born at term age, and the majority have no immediately identifiable risk factors for CP³. This lack of clinical history often leads to late diagnosis and delayed referral for intervention. A Danish register study found that ambulant children with CP are the latest to be diagnosed in particular those with bilateral spastic CP who are a mean age of 23.9 months at diagnosis⁴. In contrast, non-ambulant children typically receive their diagnosis by 6 months. A retrospective study showed referral for intervention is often delayed until the second year of life unless the child is a NICU graduate⁵. Late access to early intervention is detrimental, resulting in lost opportunities during the critical window for neuroplasticity⁶. Now that 60% of children with cerebral palsy walk independently and a further 11% use walking devices⁷ there is an ethical imperative to ensure access to cerebral palsy-specific intervention as early as possible. Children with milder CP are most responsive to early motor training and earlier diagnosis is crucial to optimise their outcomes and prevent complications.

Paediatricians are often required to distinguish between cerebral palsy and other causes of motor delay and this task can be difficult in infancy without a suspicious clinical history and no overt neurological signs such as spasticity or retained reflexes. Paediatricians may adopt a “wait and see” approach to avoid falsely labelling children and to placate parents. However, this can result in poorer parental mental health⁸ as well as delayed intervention. This paper seeks to update first contact practitioners about how to diagnose cerebral palsy in infants born at term. Case studies and a diagnostic algorithm are provided (Table 1 and Figure 1).

[Insert Figure 1]

Step 1: Take a thorough clinical history

For some infants presenting to a paediatrician with atypical motor development, a brief clinical history may highlight well-known risk factors. However, many of these children at first have a seemingly healthy pregnancy and neonatal history, but with detailed history taking, risk factors for cerebral palsy can be identified⁹. Full-term infants can have all subtypes of cerebral palsy, but usually have more severe motor impairments than preterm infants⁹. Risk factors can occur at multiple time epochs and therefore taking a good clinical history is essential. Systematic review evidence indicates pre-conception risks include: a history of previous miscarriage, stillbirth, and neonatal death; maternal age (both young and older); race; low socioeconomic status; maternal history of thyroid disease, or coagulation, disorders or seizures. Antenatal risk factors include: poly- or oligohydramnios; haemorrhage; infection; pre-eclampsia; placental abnormalities; major and minor birth defects; small and large for gestational age; plurality, multiple pregnancy and sex. Intrapartum risk factors include: length of labour; prolonged rupture of membranes; meconium-stained liquor; breech presentation; caesarean section; instrumented delivery; cord around the neck; cord prolapse; placental abruption; uterine rupture; haemorrhage; or birth asphyxia. Neonatal risk factors include: seizures; respiratory distress; hypoglycaemia; jaundice and infection⁹.

High-risk for cerebral palsy:

Neonatal encephalopathy (NE): Infants born at term with neonatal encephalopathy account for 25% of term CP¹⁰. These infants typically require resuscitation at birth. Therapeutic hypothermia is now standard of care for infants with moderate to severe signs and symptoms¹¹. Cooling is recognised as a successful preventative measure for preventing death and reducing the incidence and severity of CP¹¹. Best-practice diagnostics include brain magnetic resonance imaging (MRI) with diffusion at day 7-10 of life. Infants with severe neonatal encephalopathy whose imaging reveals basal ganglia and thalamic injury most often have dyskinetic CP affecting all four limbs and trunk. These children are often diagnosed early, due to tone problems affecting volitional movement and posture, and enrolled in follow-up programmes. Less severe NE is still a risk factor for CP, and these children often have co-occurring risk factors including congenital anomalies and growth restriction³. Most but not all children with dyskinetic CP are non-ambulant. The Australian Register indicates 25% walk. Referral to early intervention is essential for optimising motor function, but also stimulating early cognitive and literacy development since many of these children have normal intelligence.

In contrast, if the infant has a seemingly normal birth history, but has clinical signs of encephalopathy in the first year of life with the MRI showing calcification, consider testing for Aicardi-Goutières syndrome (AGS), with neurotransmitters. AGS is an autosomal recessive encephalopathy presenting with acquired microcephaly, cerebral atrophy,

cerebral calcifications, leukodystrophy, lymphocytosis and raised interferon-alpha (INF-alpha) cerebrospinal fluid levels.

Stroke: Perinatal stroke is the most common type of paediatric stroke and is the leading cause of postnatally acquired CP¹². It may occur in utero from 22 weeks or up to a month after birth due to haemorrhage or ischaemia or a thrombo-embolic phenomenon.

Clinical signs of a perinatal stroke may be subtle and go unnoticed, although many present with neonatal seizures. Best practice diagnostics include placental pathology and MRI with diffusion to determine whether or not brain injury has occurred and whether the injury has affected the motor areas and tracts predicting cerebral palsy. If clinical history and MRI are suggestive, these children can be enrolled into follow-up programmes with neonatology or neurology, as children at high-risk of cerebral palsy. Some infants may present in the first few months of life with asymmetry of motor activity, most commonly in the upper limb. However the asymmetry can also present later, after 6-9 months following cortical pruning and maladaptive response to injury – thus early intervention is critical for preservation and reorganisation of motor function. Single or multiple disabilities such as hemiplegic CP, epilepsy, cognitive or behavioural disorders may be present following perinatal stroke depending on the location of lesion¹³⁻¹⁵.

Infants at the highest risk of CP are increasingly diagnosed early using the new international early diagnosis clinical practice guideline¹⁶.

Moderate risk for cerebral palsy:

Full-term born infants at moderate risk of cerebral palsy include those with congenital disabilities, septicaemia, meningitis, small for gestational age or growth-restricted infants and those who cannot sit by 9 months or who demonstrate early motor asymmetry. These children should be thoroughly screened as they are often late to be diagnosed and to receive intervention¹⁶. Sensitive and specific tools are described in the early diagnosis clinical practice guideline¹⁶.

Step 2: Organise assessments:

Neuroimaging

Brain MRI is commonly used to help establish a recognisable pattern that may contribute to an understanding of the likely aetiology in cerebral palsy and several systematic reviews associate patterns of MRI change with an estimated timing of damage¹⁷. These studies categorise changes into three major groups that broadly ascribe damage to different stages of gestation: these include those of abnormal brain development occurring mainly in the first and second trimesters, those with periventricular change relating to changes early in the third trimester in those with cortical or deep grey matter abnormalities which are

associated with changes in the late third trimester. A possibly surprising result is the rate of malformations of cortical development which occurs in approximately 10% of those with cerebral palsy and is significantly overrepresented in term compared to preterm children. Imaging is therefore recommended as part of the diagnostic workup for cerebral palsy¹⁶. It is important to remember that 9-16% of infants with cerebral palsy have no detectable changes on MRI scanning¹⁸. Those with hypotonic or ataxic forms are over-represented in this group. Therefore, a normal MRI does not exclude the clinical diagnosis of cerebral palsy¹⁸. The historical and clinical picture is important. Clinicians should be wary of progressive and or treatable conditions such as dopa-responsive dystonia¹⁹. Furthermore, MRI can help with prognostication of motor severity and likelihood of co-occurring impairments such as central vision impairment that require specialist referrals for intervention. In contrast, cranial ultrasound (cUS) is more useful in predicting CP in term than preterm infants. While some studies indicate that increased echogenicity in the basal ganglia is an indicator of risk, the value of cUS is not established in prospective studies.

Genetics testing:

As many as one-third of full-term children with cerebral palsy lack traditional risk factors. In many of these, a genetic basis is suspected. Clues to a genetic contribution to the clinical syndrome of cerebral palsy include dysmorphic features and a phenotype that does not 'fit' the history. A progressive condition is inconsistent with cerebral palsy, although may be difficult to identify if it evolves slowly. Population studies 25 years ago²¹ demonstrated that one-third of cerebral palsy identified following term delivery were thought to be dysmorphic but did not have an immediately recognisable syndrome. The Australian authors postulated that these children might be more vulnerable to damage by comparatively minor hypoxic-ischaemic insult. These studies have been corroborated recently by CP registry data²² where congenital abnormalities or intrauterine growth restriction (IUGR) were identified in 48.6% of term or near-term singleton deliveries with CP. Fifteen to 40% of children with cerebral palsy in this study were reported to have a major congenital abnormality compared to a national figure of 3.6%. There is an overlap between the phenotype of cerebral palsy and syndromic diagnoses, in these instances, there is an increasing role for genetic or genomic diagnosis.

Contemporary genetics studies identify increased de novo copy number variation rates in cerebral palsy²³ in up to 20%²⁴⁻²⁵. Exome analysis can identify²⁶⁻²⁸ further genomic abnormalities cohorts of cerebral palsy. While some genes are novel, often studies ascribe a new role to a previously recognised gene. A genetic contribution may be present in as many as 34 to 45% of cases²⁵. This yield may be increased by focusing on particular groups with a distinctive phenotype.

The hypotonic phenotype of cerebral palsy is one where there may be particular interest from a genetic standpoint. Registry studies indicate that hypotonic cerebral palsy is associated with an increased rate of mortality²⁹, and of autism³⁰. Furthermore, this group has relatively higher rates of normal neuroimaging¹⁹. However, hypotonic CP is included as a registry field in only 54% of programmes surveyed and is not defined consistently³¹. Further work is needed to define the overlap in those with dysmorphic features and low tone. It is also apparent that there needs to be harmonisation between registries from different parts of the world regarding the terminology used.

Standardized Motor/Neurological Examination

Standardised motor assessment should always form part of the diagnostic process for infants with motor delay or aberrant motor behaviours such as early handedness. Paediatric allied health clinicians can assist with this part of the diagnostic workup. Most commonly used assessment tools can discriminate between normal and abnormal motor performance but do not accurately discriminate between cerebral palsy and motor delay³²⁻³³. Recent guidelines for the early detection of CP identify the most sensitive assessments for detecting CP¹⁶:

- Prechtl's Qualitative Assessment of General Movements (GMA): The GMA assesses the spontaneous movement of infants and is scored from a 3-5 minute video. Between 12 to 20 weeks post term age the GMA has the highest sensitivity and specificity for detecting CP with values of about 98% and 91% respectively during the fidgety period (9-20 weeks.) These values have also been confirmed in Australian settings and GMA is now regarded as a standard assessment in high-risk populations³⁴. Once volitional movements are more predominant around 5 months of age, general movements cannot be reliably tested³⁵.
- Hammersmith Infant Neurological Examination (HINE): From 2-24 months the HINE is one of the most sensitive tools for detecting CP¹⁷. This scoreable neurological assessment can be easily used in the clinical setting and can be learned with minimal training. Optimal scores are defined with cut offs for CP at 3, 6, 9 and 12 months³⁶ and scores less than 40 strongly predict non-ambulant CP. In the case of suspected hemiplegia, it is important to document the number of asymmetries as mildly affected children may score in the optimal range³⁷.

Assessments that focus on motor milestones are not as reliable predictors of CP unless used longitudinally. The Developmental Assessment of Young Children (DAY-C), the Alberta Infant Motor Scale (AIMS) and the Neurological, Sensory, Motor, Developmental Assessment (NSMDA) all have good sensitivity and specificity for CP when used over time^{38, 39}. It is important to note that some children with mild CP may have near normal milestone acquisition over the first year of life, especially those with mild hemiplegia. Some motor

tests allow the fine motor component to be scored on the less affected side only giving a false impression of "normality". It is therefore important to assess the quality of movement as well as the existence of asymmetry when parents express concern.

The age of the infant will determine 'best practice' choice of assessments at initial presentation but always involves a combination of assessments to triangulate findings.

Step 3: Investigate co-morbidities

Co-morbidities are common, particularly in severely affected children, and can be more disabling than the motor impairment⁴⁰. Investigating and managing these comorbidities are necessary to optimise outcomes and prevent secondary impairments. Three in 4 experience chronic pain; 1 in 2 have an intellectual disability; 1 in 3 have a hip displacement; 1 in 4 cannot talk; 1 in 4 have epilepsy; 1 in 4 have a behaviour disorder; 1 in 4 have bladder control problems; 1 in 5 have a sleep disorder; 1 in 5 have sialorrhoea; 1 in 10 have vision impairments; 1 in 15 require tube feeding; and 1 in 25 have hearing impairments⁴⁰. The most common comorbidities in infants include: pain from reflux, constipation or involuntary postures arising from the movement disorder; sleep disorders from either unmanaged pain or apnoea or vision impairments affecting sleep-onset, or seizures. Prophylactic hip x-rays to detect hip displacement should commence from one year of age. In the first year of life, regularly checking in with parents about the child's pain and sleep is an essential part of best-practice care.

Summary

Paediatricians have an essential role to play in the early identification of full-term infants with cerebral palsy, most of whom will not have been under the care of a neonatologist. When an infant presents with atypical movement or motor development, it is important to take a thorough clinical history and conduct investigations using sensitive and specific tools. Neuroimaging and specific motor assessments are initially used to identify potential causes and probable severity. Genetics investigations might also be required if results are confounding. The term "high risk of cerebral palsy" can be given before final confirmation of the diagnosis so that early intervention can begin. An early CP diagnosis enables diagnostic-specific early intervention. Investigations should be undertaken for concurrent impairments.

MCQ

1. What is the ONE most common co-morbidity in severe cerebral palsy?

- Author Manuscript
- A. Chronic pain
 - B. Epilepsy
 - C. Hip displacement
 - D. Intellectual disability
 - E. Sleep disorder

2. Which ONE form of cerebral palsy is associated with autism?

- A. Ataxic
- B. Athetoid
- C. Dyskinetic
- D. Hypotonic
- E. Spastic diplegia

3. Which ONE is the most common cause of post-natally acquired cerebral palsy?

- A. Child abuse
- B. Congenital malformations
- C. Genetic
- D. Meningitis
- E. Perinatal stroke

Answers to MCQ's

1. A: about 75% have chronic pain, compared with 25% with epilepsy, 33% with hip displacement, 50% with intellectual disability; and 20% with a sleep disorder.
2. D: hypotonic cerebral palsy is associated with increased risk of autism and increased mortality.
3. E: Perinatal stroke due to haemorrhage or ischaemia or a thrombo-embolic phenomenon may occur *in utero* from 22 weeks and for up to a month after birth.

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Figure 1: Algorithm for Differential Diagnosis of Term-Born Cerebral Palsy

Note this figure incorporates Kurian et al., 2011⁴¹ algorithmic decision-making support for identification of dopamine transporter syndromes. Legend: CK = Creatine Kinase; DAYC = Developmental Assessment of Young Children; MRI = Magnetic Resonance Imaging; SD = Standard Deviation; SMA – Spinal Muscular Atrophy

CASE STUDIES – DIAGNOSIS and MANAGEMENT				
CLINICAL HISTORY	PRESENTING ISSUES	INVESTIGATIONS	TREATMENTS	OUTCOME
7- month term baby	• Sleep problems –	• Sleep paediatrician	• High dose Losec	• Sleep thought

with neonatal encephalopathy, Sarnat 3	wakes hourly at night, no day sleep • Unsettled all day, needs to be held	resulting in diagnosis of GORD • OT consult	and Zantac • Sleep hygiene recommendations	night in 10 days + day sleeps • Rapid developmental progress • CP: GMFCS III, normal IQ
9-month term baby, HIE, not cooled. MRI on Day 7, damage to deep grey matter. Abnormal GMs and HINE score <40 at 3 months. Epilepsy and spasms managed with Clobazam and Keppra	• Sleep disorder – waking 6 times at night • Food refusal at 9 months due to pain although on recommended reflux meds	• Gastroenterologist referral	• Temporary NG tube • High dose reflux meds	• Reduced pain • Increased caloric intake • Improved sleep and learning • CP – GMFCS V
5-month old term baby with uncomplicated birth and pregnancy history. Developmental milestones and growth all within normal limits.	• Asymmetry of hand use, with right hand often closed	• HINE assessment with a score of 59 and 7 asymmetries	• Neurology review • MRI under GA reveals large area of encephalomalacia in LMCA territory	• Referred to physiotherapy and occupational therapy • CP – hemiplegia, GMFCS 1, MACS 2
5 month old baby born at 37 weeks, BW 2450g. Required neonatal resuscitation and had Apgar of 3 at 1 minute. Spent 3 weeks in SCN. Normal CUS and MRI. Hip dysplasia treated with Pavlik harness.	• Developmental delay, referred by allied health clinicians	• Neuro exam – full head lag • Microarray chromosome analysis was normal • 12 months – developed seizures • Repeat MRI at 12 months revealed white matter injury	• Neurology review • Medications for seizures • Early intervention	• At 2 years sibling also presented with seizures and developmental delay • Genomic analysis demonstrated pathogenic mutation in CANA1A in both siblings

TABLE 1: Clinical Scenarios

GMFCS = Gross Motor Classification Scale; GMs= General Movements; HINE=Hammersmith Infant Neurological Examination; MACS = Manual Ability Classification Scale;

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