Early hand preference and infantile spasms:
A case of left-sided cerebral palsy

Tanusree Dutta, Hannah Lorking, Helena Kim, Sowmya Gandham, Omar Akram, Anthony Liu, Habib Bhurawala

CASE

A girl aged 5 months was referred to a paediatric clinic because of concerns about persistent right-hand preference. She was born at term via uncomplicated vaginal delivery, with mild neonatal jaundice not requiring phototherapy and normal Apgar scores (9, 9). Her developmental milestones were appropriate for her age.

On examination, she exhibited right-hand dominance, with normal global tone and bilateral grasp reflex. There were no dysmorphic features or gross asymmetry. Early recognition of hand dominance by her general practitioner (GP), a red flag for pathology in infants aged under 12 months, prompted a structured work-up.[1] Table 1 outlines the key differential diagnoses, with cerebral palsy (CP) being a key differential when associated with motor asymmetry.[1]

QUESTION 1

What investigations/assessment are indicated in cases of early hand preference?

QUESTION 2

What are some key causes of CP?

QUESTION 3

What is porencephaly, and how does it relate to CP?

Table 1. Differential diagnosis for early unilateral hand preference

System	Differential diagnosis	Additional information
Neurological: CNS	• Perinatal stroke/haemorrhage • Periventricular leukomalacia • Congenital abnormalities (eg schizencephaly/ porencephaly) • Perinatal hypoxic–ischaemic injury • Space-occupying lesion	Many of these conditions could precipitate the development of cerebral palsy[1] MRI abnormalities in cerebral palsy:[A] • White matter damage (45%) • Basal ganglia (13%) • Congenital malformation (10%) • Focal infarcts (7%)
Neurological: PNS	• Brachial plexus injury	Often associated with complications during delivery such as shoulder dystocia. Maternal risk factors including gestational diabetes, or foetal risk factors such as macrosomia might be present
Musculoskeletal	• Trauma (dislocation/fractures) • Congenital limb abnormalities	

[A]Indicates the prevalence in children with cerebral palsy.[1,20–23]
CNS, central nervous system; MRI, magnetic resonance imaging; PNS, peripheral nervous system.

ANSWER 1

The investigations/assessment that are indicated in cases of early hand preference are:

- neuroimaging: head ultrasonography followed by magnetic resonance imaging (MRI) if aetiology is unclear.
- electroencephalography (EEG): if seizures are suspected.
- genetic testing: comparative genomic hybridisation (CGH) microarray is a technique that looks at copy number variants (loss or gain of DNA) across the entire genome at a high resolution. GPs can initiate microarray testing with appropriate pre- and post-test counselling, ideally in collaboration with or after discussion with local genetics services.[2]
- developmental assessment: allied health review (physiotherapy, occupational therapy [OT]).[1,3,4] GPs are not expected to perform formal general movements assessments (GMA); however, they play a key role in developmental surveillance, identifying red flags (eg early hand preference, abnormal tone, delayed milestones) and referring appropriately for specialist developmental assessment or GMA.

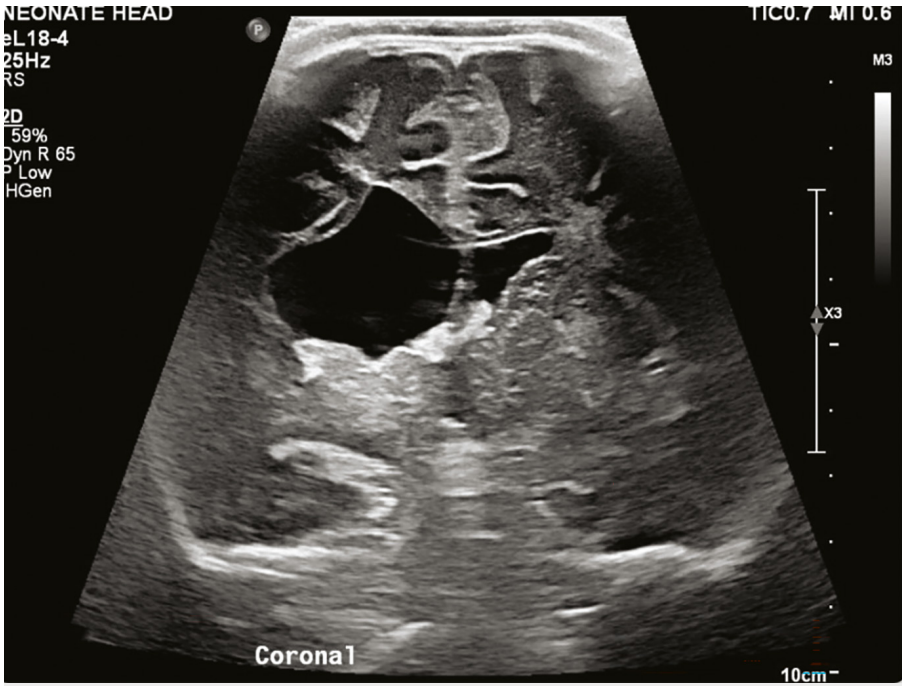

Figure 1. Head ultrasound image showing a porencephalic cyst around the frontal horn of the right ventricle.

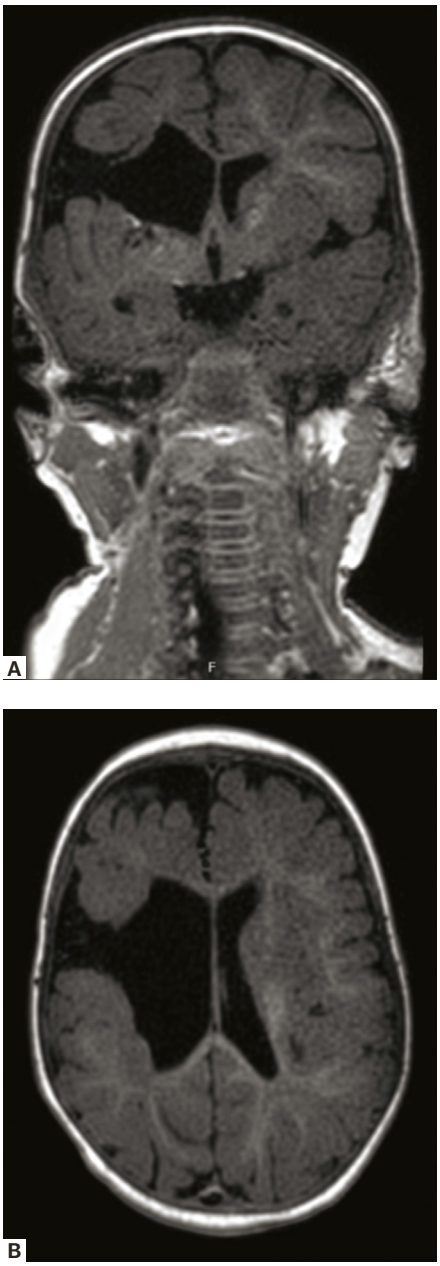

Figure 2. T1-weighted magnetic resonance imaging of this patient showing changes in the right frontal lobe in keeping with porencephaly. **A.** Coronal view. **B.** Axial view.

ANSWER 2

It can be beneficial to categorise the causes of CP as prenatal, perinatal and postnatal factors.[5,6] Prenatal factors include periventricular leukomalacia (damage to the white matter of the brain), infections caught during pregnancy (cytomegalovirus, rubella, chickenpox or toxoplasmosis) and stroke. Key perinatal differentials include antepartum haemorrhage and chord prolapse. Postnatal causes are also important to consider and include serious head injury, infections such as meningitis and hypoglycaemia.

CASE CONTINUED: INITIAL INVESTIGATIONS AND FINDINGS

Neuroimaging was arranged for the patient.

A head ultrasound revealed a right hypoechoic area (28 × 28 mm), potentially representing ventriculomegaly or a porencephalic cyst (Figure 1). A subsequent MRI brain scan confirmed right frontal lobe porencephaly, likely secondary to perinatal haemorrhage rather than congenital malformation (eg schizencephaly) (Figure 2).

The child was referred to a CP early diagnosis clinic, where the diagnosis of left-sided spastic hemiplegic CP was made.

ANSWER 3

Porencephaly is a cystic cavity in the brain parenchyma, often resulting from vascular insults during the perinatal period. It can disrupt motor pathways and cause spastic hemiplegia.[7]

CASE CONTINUED

At 7 months of age, the child's fine motor function on the left side lagged, with a significantly weaker grasp strength. Early intervention services were continued: physiotherapy, OT, speech therapy and enrolment in the National Disability Insurance Scheme (NDIS; www.ndis.gov.au).

At approximately 9 months of age, the child's mother recorded episodes of eye-rolling and loss of postural tone. A video EEG confirmed frequent right-sided epileptic discharges and infantile spasms.

The diagnosis of infantile spasms, a severe form of epileptic encephalopathy, prompted urgent treatment with high-dose corticosteroids and then vigabatrin and

clobazam. Seizure frequency reduced substantially. A repeat EEG showed ongoing epileptiform discharges but no clinical spasms.

QUESTION 4

What are the differential diagnoses of convulsions in infants?

QUESTION 5

What are infantile spasms and their relevance in CP?

ANSWER 4

There are numerous differentials for convulsions in children, with varying aetiologies. These include epilepsy syndromes, central nervous system (CNS) infections, neurocutaneous syndromes and non-epileptic convulsions. There are many different epilepsy syndromes that present at different ages. Syndromes that often present in the neonatal period include benign familial neonatal epilepsy and early myoclonic encephalopathy. Genetic epilepsy with febrile seizures plus (GEFS+), Dravet syndrome (severe myoclonic epilepsy of infancy) and West syndrome often present in infancy. Lennox–Gastaut syndrome presents in childhood, whereas juvenile absence epilepsy can present at variable ages.

CNS infections that can lead to convulsions include bacterial/viral meningitis or encephalitis. Neurocutaneous syndromes can also cause convulsions in children; these include Sturge–Weber syndrome, tuberous sclerosis complex and neurofibromatosis. It is also vital to consider non-epileptic causes. Key differentials include hypoglycaemia or other metabolic disorders, breath-holding spells, reflex anoxic seizures and febrile seizures.[8,9]

ANSWER 5

Infantile spasms present between ages 1 and 24 months with sudden muscle contractions, eye deviation and sometimes developmental regression. They are associated with structural brain abnormalities, including those seen in CP. Hospital admission is advised with neurological evaluation and early treatment to prevent complications.[10]

CASE CONTINUED

At age 12 months, the child was making steady developmental progress, which included:

- sitting independently
- babbling and playing interactively
- improved left-hand use with therapy and splinting
- no developmental regression, which is a positive sign of treatment response
- continuation of antiepileptic medications, with seizure frequency down to 3–4/day, with ongoing neurology follow-up.

The child underwent genetic testing, which identified a *COL4A1* mutation. A timeline of events is shown is Figure 3.

QUESTION 6

How does this mutation relate to CP?

QUESTION 7

In this child, what other investigations should be conducted to diagnose the comorbidities of Gould syndrome?

QUESTION 8

How is CP diagnosed early?

QUESTION 9

What is the appropriate management approach for infantile spasms and CP?

ANSWER 6

Both *COL4A1* and *COL4A2* gene mutations can cause neurological defects including porencephaly and perinatal intracerebral haemorrhage, which can evolve into CP. The features of these mutations are collectively referred to as Gould syndrome, a very rare multisystem disorder that can also cause myopathy, nephropathy and ophthalmological disorders.[11,12] Investigating genetic causes for CP allows better management of the disease and prevention of complications arising from the underlying syndrome.

ANSWER 7

As Gould syndrome is a very rare disorder, there is a lack of official guidance regarding its management. A multidisciplinary team approach involving nephrologists, neurologists, ophthalmologists, GPs and other specialists will be essential to address the multisystem manifestations of this disease. The age of onset and severity of symptoms can vary from patient to patient.

Foetal presentations of the cerebrovascular manifestations of Gould syndrome include hydrocephalus, haemorrhage, schizencephaly or porencephalic cysts. Older patients might present with diffuse cerebral small vessel disease. Patients will need neuroimaging for diagnosis and regular monitoring thereafter.

Ophthalmological manifestations such as congenital cataract or Axenfeld–Rieger syndrome (anterior segment dysgenesis) might be present from birth. Patients will initially require ophthalmoscopy and a slit lamp examination, with further exams as indicated.

Musculoskeletal symptoms such as muscle cramps often begin in early childhood and might warrant a baseline measurement of serum creatinine kinase.

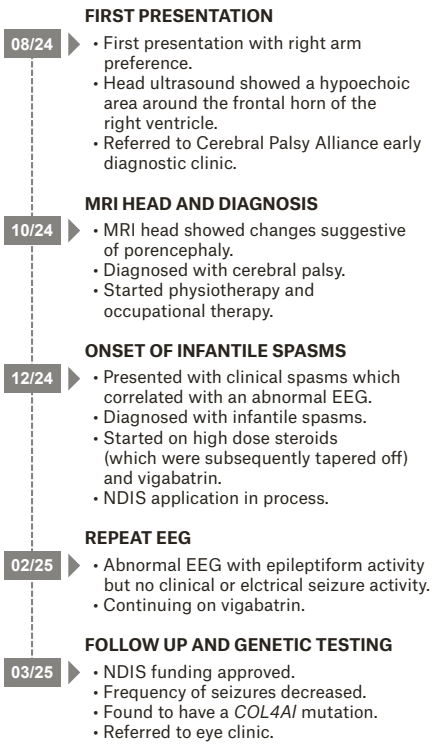

Figure 3. Timeline of key clinical events, investigations and interventions for this case study.

EEG, electroencephalography; MRI, magnetic resonance imaging; NDIS, National Disability Insurance Scheme.

A baseline electrocardiogram and echocardiogram will likely be required to screen for cardiovascular complications such as arrhythmia and congenital heart disease.

Renal manifestations can include renal cysts and haematuria. Relevant initial investigations include baseline kidney and bladder ultrasonography, urinalysis, and measurement of serum creatinine level and estimated glomerular filtration rate.[13–16]

ANSWER 8

Early diagnosis relies on clinical signs such as motor asymmetry and abnormal tone/reflexes. Neuroimaging is then required to show evidence of perinatal insult. There is also a role for structured tools, including the General Movements Assessment and Hammersmith Infant Neurological Exam.[17] Paediatric specialist review is needed in an early diagnosis clinic. GPs are instrumental in early suspicion of CP and timely referral.[3,17]

ANSWER 9

A holistic multidisciplinary approach is vital in the management of infantile spasms and CP. Seizure management is key, including the use of vigabatrin and corticosteroids (under neurology guidance). Early intervention will involve physiotherapy, OT and speech therapy for children. Another key aspect of holistic management is parental support, which involves NDIS access, care coordination and parental counselling. For these children, it is essential to monitor for developmental milestones, changes in vision and seizures. GPs should also highlight to their patients the importance of seizure safety. This involves parental education, water safety and consideration of helmets only in the rare case of frequent atonic seizures/drop attacks, with guidance from a paediatric neurologist.[13]

GPs play a central role in coordinating community follow-up, monitoring and family support.[1,3,10] A summary of key resources for GPs can be found in Table 2. With early intervention, functional outcomes can be significantly improved.[18] Therapy should be tailored to the patient's specific needs and preferences.[19]

Key points

- Persistent hand preference before age 12 months is a red flag.
- Infantile spasms are a medical emergency; early treatment alters long-term prognosis.
- GPs are central to early detection, referral and care planning for children with developmental conditions.
- Multidisciplinary early intervention improves developmental outcomes.

Table 2. Resources for GPs: CP management, infantile spasms referral pathways, NDIS engagement and support for parents

Topic	Resources available
CP management	• Cerebral Palsy Alliance: Useful guides on early diagnosis and intervention for parents and care givers.[24] • Oceania Academy of Cerebral Palsy and other Childhood-onset Disabilities: Fact sheets for GPs including advice on referral pathways and the NDIS.[25] • The Royal Children's Hospital Melbourne: Clinical practice guidelines. Advice on presentation, investigation and holistic management of CP. Guidance on referral to appropriate services.[3]
Infantile spasms	• The Royal Children's Hospital Melbourne: Clinical practice guidelines. Advice on assessment, management and appropriate escalation of children presenting with infantile spasms.[10]
NDIS	• Cerebral Palsy Alliance: Provides support navigating the NDIS for patients with cerebral palsy and their families. Assists patients preparing for annual NDIS meetings by helping to identify key goals and the services, supports, therapies and equipment needed to achieve these goals.[24] • NDIS website: General information on the NDIS application process (www.ndis.gov.au).
Support/ counselling for parents	• Family Connect and Support: Can help families connect to support services. • Carers NSW: Provides online resources, supports and services, and training and education. This includes carer support groups and career pathways for carers. • Healthy Mothers Healthy Families: A website with online modules designed to help empower mothers of children with a disability.[26]

CP, cerebral palsy; GP, general practitioner; NDIS, National Disability Insurance Scheme; NSW, New South Wales.

Authors

Tanusree Dutta MBBS, BSc, Resident doctor, Department of Medicine, Imperial College London, London, UK

Hannah Lorking MBBS, DCH, BN, Provisional fellow in paediatrics, Department of Paediatrics, Nepean Hospital, Nepean Blue Mountains Local Health District, Sydney, NSW; Clinical Associate Lecturer, Sydney Medical School, Faculty of Medicine and Health, The University of Sydney, Sydney, NSW

Helena Kim MBBS, BSc (Hons), MSc, DCH, Provisional fellow in paediatrics, Department of Paediatrics, Nepean Hospital, Nepean Blue Mountains Local Health District, Sydney, NSW; Clinical Associate Lecturer, Sydney Medical School, Faculty of Medicine and Health, The University of Sydney, Sydney, NSW

Sowmya Gandham BA, MBBS, MPH, DCH, FRACP, Staff specialist paediatrics, Department of Paediatrics, Nepean Hospital, Nepean Blue Mountains Local Health District, Sydney, NSW; Associate Lecturer, Discipline of Paediatrics, Nepean Clinical School, Faculty of Medicine and Health, The University of Sydney, Sydney, NSW

Omar Akram FRACGP, MD, PhD, BSc (Hons), General Practitioner, Medical and Fitness Centre, Sydney, NSW; Visiting Medical Officer (VMO), Cessnock Hospital, Cessnock, NSW

Anthony Liu MBBS, MPH, MMedEd, DCH, FRACP, Senior Consultant Paediatrician, Department of Paediatrics, Nepean Hospital, Nepean Blue Mountains Local Health District, Sydney, NSW; Senior Lecturer, Discipline of Paediatrics, Nepean Clinical School, Faculty of Medicine and Health, The University of Sydney, Sydney, NSW

Habib Bhurawala MBBS, MD, DCH, AFRACMA, FRACP, Head of Paediatrics and Senior Staff Specialist, Department of Paediatrics, Nepean Hospital, Nepean Blue Mountains Local Health District, Sydney, NSW; Senior Visiting Medical Officer (Paediatrics), Blue Mountains ANZAC Memorial Hospital, Blue Mountains, NSW; Clinical Professor (Paediatrics), The University of Notre Dame, Sydney, NSW; Clinical Associate Professor Paediatrics, The University of Sydney, Sydney, NSW

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Correspondence to:
tanud26@gmail.com

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