

## 5 – Diagnosis

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**Study Advisory Group questions:** *Are there delays in diagnosis? Is there variation in how the cerebral palsies are described?*

**Why is this important?** *Timely diagnosis of a cerebral palsy matters so that early interventions can be accessed and all reasonable adjustments put in place to facilitate the best possible participation in everyday activities.*

The diagnosis of a cerebral palsy is clinical, based on specific findings on medical, developmental and family history and on clinical examination. A cerebral palsy is not the same as ‘any physical disability of any cause’, but is a very precise and specific diagnosis. It is important to distinguish the cerebral palsies from other conditions that may masquerade as such, but which have very different clinical courses and implications for management. These include, progressive, neurodegenerative conditions, hereditary spastic paraplegias and situations where a child’s development arrested at a stage before motor skills were acquired and has stopped progressing further, leading to postural changes and contractures due to disuse. The Surveillance of Cerebral Palsy in Europe’s Reference and Training Manual provides clear guidance on the diagnostic assessment process to be undertaken.<sup>16</sup> In addition, red flags for other neurological conditions and risk factors for the cerebral palsies are detailed in the NICE Guideline NG62.<sup>19</sup>

### Timely diagnosis

A timely diagnosis is one that is made as early as possible in the child’s life. The majority of children with a cerebral palsy will receive their diagnosis by three years of age,<sup>20</sup> although this will vary in individual circumstances and will depend on the severity of motor impairment, with those with the most severe motor impairment being identified earliest. For some infants, for example those born prematurely, the clinician may use the term ‘probable emerging cerebral palsy’ during the period in the early months when neurological

signs can fluctuate, to avoid over-diagnosis in those whose neurological signs subside over time, but also to facilitate early interventions.

Whilst routinely collected population datasets do not record the time of diagnosis, 60% of cases of a cerebral palsy first appeared within CPRD (England HES linked) dataset before the age of five years, 38.5% before the age of two years. The North of England Collaborative Cerebral Palsy Survey data, showed that the diagnosis of a cerebral palsy was made before the age of two years in 73% (293/398) of cases. There is therefore an apparent delay between the diagnosis of a cerebral palsy recorded in routine national datasets and within the cerebral palsy registers (the latter is influenced by the rules of the register i.e. the data capture points, which can vary between registers.)

### CASE STUDY 1

A teenage patient was reviewed by a new clinician in the paediatric clinic. The diagnosis recorded in the patient’s medical record was ‘ataxic cerebral palsy’. The clinical assessment documented a changing profile of needs over time that did not fit with this and further investigations were arranged.

*The case reviewer noted that the evidence of the investigation findings was that the diagnosis was actually one of a rare group of conditions with progressive and multi-system effects that required a completely different, proactive healthcare management plan than that for a person with ataxic cerebral palsy. They commented that it is always good practice to review the evidence for, or against, any diagnostic labels and be prepared to reinvestigate in the light of new information or new diagnostic technologies.*

A delay in diagnosis was reported by the case reviewers in 19/193 (9.8%) of the case notes reviewed. Where the diagnosis was made in the last three years (n=46), lead clinicians who returned a questionnaire indicated there had been a delay in diagnosis in five patients.

### Description of tone variation and pattern of motor impairment

Precision of description of tone variation and pattern of motor impairment are well described in the Surveillance of Cerebral Palsy in Europe Reference and Training Manual<sup>16</sup> and are very important in informing accurate management across settings and ensuring the best outcomes.

Documentation of the patient's specific cerebral palsy diagnosis was recorded by the case reviewers in 430/540 (79.6%) cases, no such documentation in 110/540 cases (20.4%), unable to answer was recorded in 15/554 cases. In 150/521 (28.8%) cases reviewed the term used to describe the diagnosis was 'cerebral palsy', with no more specific detail of tone variation, whilst in a further 76/521 (14.6%) only the term 'bilateral cerebral palsy' was used, but the tone variation was not described. The diagnostic term did not include information about the specific tone variation in 297/521 (57%) cases. Table 5.1 shows whether a diagnosis was documented, by age of the patient as reported by the case reviewers.

**Table 5.1 Documentation of the patient's specific cerebral palsy diagnosis by age**

	0-4 years	5-9 years	10-14 years	15-19 years	20-25 years	Total
	n=	n=	n=	n=	n=	n=
Yes	79	116	84	84	68	<b>431</b>
No	18	5	20	21	36	<b>100</b>
<b>Subtotal</b>	<b>97</b>	<b>121</b>	<b>104</b>	<b>105</b>	<b>104</b>	<b>531</b>
Not answered	1	6	3	2	1	<b>13</b>
<b>Total</b>	<b>98</b>	<b>127</b>	<b>107</b>	<b>107</b>	<b>105</b>	<b>554</b>

### Routinely collected data about a cerebral palsy diagnosis

Challenges in identifying those with cerebral palsies from routinely collected population data included:

1. Lack of specificity of ICD-10 and Read version v2 codes used. The most common code used for the cerebral palsies was G80.9 (cerebral palsy unspecified) in CPRD (England HES linked data); analysis by a cerebral palsy type was therefore not possible. G80.9 was used for:
  - 41% of all inpatient episodes
  - 71% of outpatient attendances (for the few cases where disease coding for a cerebral palsy was available)
  - 87% of patients who died
2. For some children and young people, multiple codes were used
3. Cerebral palsies were rarely coded at every point of contact with NHS services
4. Different codes were used on different occasions for the same child or young person.

To enable a summary of the variation in coding used, READ v2 codes used were mapped on to ICD-10 'group' codes for the GP data (Appendix 3). For the 8,965 patients with cerebral palsies identified within CPRD GP dataset,

- 77% (6,884) were coded from one group code (G80-G83 or equivalent Read code), across all contacts, the majority (94%) of which (6,472) included a G80-G83 code, of those, 68.9% (4,463) were coded exclusively with a G80 code.
- In 22% a combination of two group codes were used over time and three or more different codes were used for 1% of cases.

Of all children and young people with cerebral palsies identified in CPRD dataset, cerebral palsies were only coded at one time point in all of the person's contacts with NHS in 36.4% (3,265/8,965) (G80-83.3 or equivalent Read v2) of cases at any time during the study period, most of these cases appeared in CPRD GP data (2080 (63.7%) and 1185 (36.3%) from England HES data).

### Availability and use of magnetic resonance imaging

MRI neuroimaging is an important tool for understanding the causal pathway of a cerebral palsy and it can highlight some important conditions with different management implications that may be missed, such as developmental brain anomalies and neurometabolic conditions. Guidelines for the use of MRI have been issued by the American Academy of Paediatrics<sup>21</sup> who recommend neuroimaging for all children where a diagnosis of a cerebral palsy is being considered and NICE guidance NG62<sup>19</sup> recommends neuroimaging only when it is not clear how the cerebral palsy came about.

#### CASE STUDY 2

A teenage patient accompanied by their father was reviewed by a new clinician in the paediatric clinic. The patient's clinical signs suggested a diagnosis of unilateral cerebral palsy. An MRI scan of the patient's head revealed a significant developmental brain anomaly which fitted in with the clinical findings.

*The case reviewer noted that the clinician had documented that the patient's father walked with a stick and on enquiry into family history, this was long standing but had never been formally assessed and no diagnoses had ever been made. The father was advised to see his GP to seek neurological assessment. He was found to have the same developmental brain anomaly as his child. The reviewer noted that subsequent genetic investigations revealed the underlying cause of the unilateral cerebral palsy in both family members.*

#### CASE STUDY 3

A young child who had been born at-32 weeks, was assessed in the paediatric clinic and found to have spasticity of both lower limbs and associated clinical signs suggestive of a diagnosis of bilateral spastic cerebral palsy. An MRI head scan revealed bilateral, symmetrical signal changes that the neuroradiologist reported were NOT typical of the expected finding of periventricular leukomalacia. Further metabolic and genetic investigations were undertaken that revealed a specific diagnosis of a rare neurodegenerative disease

*The case reviewer reflected on the important new information gleaned from the MRI scan and how this dramatically changed the management of this patient, also the implications for the family, as the parents were first cousins and planning further children, with a one in four recurrence risk. Early testing in future pregnancies could have treatment implications, as stem cell transplantation could be considered, with the chance of improved outcome.*

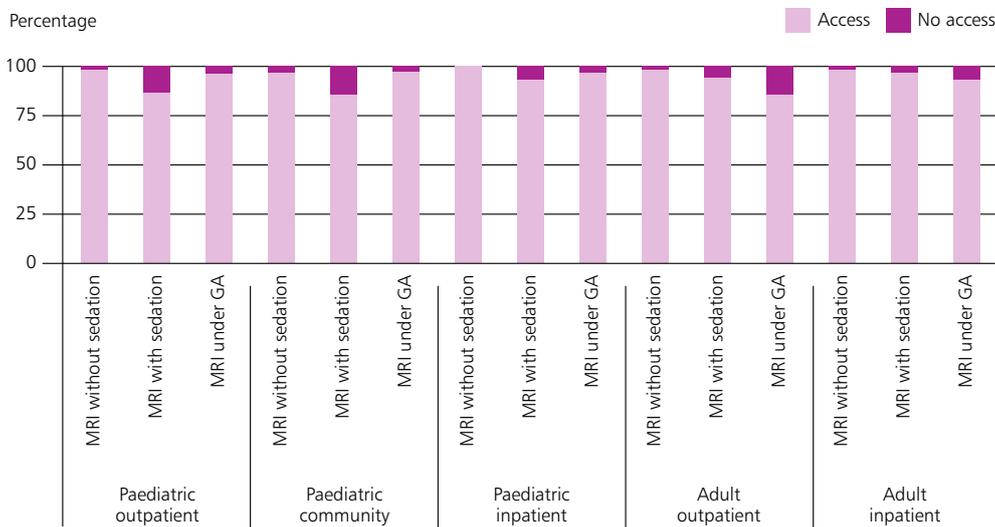
Within the population-based North of England Collaborative Cerebral Palsy Survey (NECCPS) 56% (239/429) of patients (<12 years of age and born between 1995 and 2002) with cerebral palsies had MRI neuroimaging. These data were recorded inconsistently in the Northern Ireland Cerebral Palsy Register. A review of the prevalence of MRI neuroimaging was attempted within CPRD GP and HES linked data, however a generic code for MRI was most frequently used which may have included MRI neuroimaging. The data were imprecisely coded and thus unlikely to give a true representation of the situation.

Variation in MRI scan reporting matters when considering neuroimaging in children and young people with cerebral palsies. If accurate information is to be gleaned from the imaging about likely causation of the cerebral palsy, correct identification of any clues to timing of the disruption to the developing brain as well as an accurate description of the pattern of brain disruption are essential.<sup>22</sup> MRI neuroimaging was reported in the organisational surveys to be offered as either routinely or selectively depending on clinical assessment (Table 5.2). There was also wide variation in access to neuroradiological expertise for neuroimaging reporting, where it existed, with a split between routine provision and ad hoc provision with 133/193 (68.9%) providing routine provision.

**Table 5.2 Provision of MRI for patients suspected of having a cerebral palsy**

	Paediatric outpatients	Paediatric community
Routinely	43	50
Selectively depending on clinical assessment	37	27
<b>Subtotal</b>	<b>80</b>	<b>77</b>
Not answered	3	1
<b>Total</b>	<b>83</b>	<b>78</b>

Figure 5.1 shows access and lack of access to Magnetic Resonance Imaging without sedation, with sedation and under general anaesthetic as reported by organisational leads for different pathways of care. Often the default position was to use general anaesthetic.



**Figure 5.1 Availability of MRI neuroimaging by use of sedation or general anaesthesia**

### Key Findings – questionnaire, case note review and organisational data

- The patient's specific cerebral palsy diagnosis was not documented in the case notes in 110/540 (20.4%) cases reviewed
- In 150/521 (28.8%) cases reviewed, the term used to describe the diagnosis was 'cerebral palsy' with no more specific detail of tone variation. In a further 76 cases (14.6%) the term 'bilateral cerebral palsy' was used but there was no further documentation of tone variation. The diagnostic term did not include information on specific tone variation in 297/521 (57%) cases reviewed
- Where specialist expertise was in place, this was available to interpret neuroimaging on an 'ad hoc' basis in a third of organisations (paediatric outpatient care, 23/74; community paediatrics, 25/74; adult outpatient care, 12/45)
- Where undertaken, MRI neuroimaging was offered on a routine basis in 43/82 organisations providing paediatric outpatient care and 50/77 organisations providing paediatric community care. There was variation in whether organisations offered MRI under sedation or general anaesthetic. Paediatric services were less likely to offer MRI under sedation and adult services less likely to offer MRI under general anaesthetic.

### SEE RECOMMENDATIONS 1•2•3•6

### Key Findings – routine national data

- Cerebral palsies, although chronic conditions, are not coded at every contact point with NHS services. This illustrates a problem with inconsistent coding of a chronic health condition in routinely collected healthcare data
- The variation between ICD-10 and Read v2 codes recorded both within and between individual children and young people with a cerebral palsy impairs complete and accurate case ascertainment from routinely collected healthcare datasets
- The specific type of cerebral palsy was identified at some point in 79.6% of case notes (in the case notes review). The missing data and lack of consistent documentation in case notes over time would impair the ability to code cases according to type within healthcare datasets and, a 'generic' code for a cerebral palsy was used in the majority of cases
- The absence of coding by a cerebral palsy type and the absence of a system to record the level of impairment in a patient with cerebral palsy affects the ability to use routinely collected data to analyse whether healthcare utilisation is proportionate to need or disease severity. It was not possible to analyse routinely collected data by cerebral palsy subtype or by motor function
- The inaccuracy of coding of MRI within routine healthcare datasets precluded an accurate evaluation of the prevalence of MRI neuroimaging in patients with cerebral palsies. These data were more consistently recorded within designated cerebral palsy registers
- Data accuracy should be improved with a wider adoption and recording of the same classification system and SNOMED CT codes across the UK which may facilitate data comparisons from different countries and regions in the UK, highlight variations and drive up quality of care, however the introduction of SNOMED CT varies and is at different stages across the UK. The transition to SNOMED CT is likely to have a positive impact on the analysis of routine healthcare data.

### SEE RECOMMENDATIONS 1•2