

ICPGC Common Data Elements for Genetic Studies in CP

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Background

These common data elements (CDEs) were developed in 2019 by the International Cerebral Palsy Genomics Consortium Phenotype Working Group.

They are designed to be used for genomics studies in cerebral palsy. In addition to these CDEs to capture clinical data, we highly encourage researchers to adopt the Human Phenotype Ontology (HPO) to describe clinical traits. The HPO is a standardised vocabulary of phenotypic abnormality. For more information, please see [here](#).

There are **118 CDEs**: 8 data elements make up the Minimum Data Set (Mandatory), 50x Core, 44x Recommended and 16x Exploratory data fields

Current working version: Version 1.2, updated 8th October 2020

NB: Red changes have not yet been implemented. These will be part of Version 1.2 (late 2020)

Demographics (9)

FIELD NAME	CATEGORY	DESCRIPTION	VALUE	NOTES / LOGIC
birth_year	Mandatory	What is the individuals' year of birth?	XXXX (years)	1900+
sex	Mandatory	What is the sex of the individual?	1, Male 2, Female 3, Intersex 99, Unknown	Boolean
birth_country	Mandatory	What country was the individual born in?	ISO-3166 2-alpha code	See Excel document, more than 2 characters will generate error upon upload
country_residence	Recommended	What country does the individual currently reside in?	ISO-3166 2-alpha code	See Excel document, more than 2 characters will generate error upon upload
personal_ethnicity	Recommended	What ethnicity does the individual identify with?	free text	
genotype_ethnicity	Recommended	What is the individuals' ethnicity; as determined by genotype?	free text	
vital_status	Core	What is the individual's vital status?	1, Alive 2, Dead 3, Unknown 4, Not reported	Boolean
death_age	Recommended	If deceased, what was the individual's age at time of death?	1-99	This can only be responded if [vital_status] has a value of (2)
cause_of_death	Core	If deceased, what was the individuals primary cause of death (ICD-10 code)	3-7 alphanumeric code with a single period (.) that follows the first 3 alphanumeric characters (e.g. F91.9)	This can only be responded if [vital_status] has a value of (2) https://icd.who.int/browse10/2019/en

Diagnostics (10)

FIELD NAME	CATEGORY	DESCRIPTION	VALUE	NOTES /LOGIC
CP_phenotype	Mandatory	Does the individual have a permanent (non-paroxysmal) movement disorder?	0, No 1, Yes	
CP_onset	Exploratory	Was onset at age 2 or younger?	0, No 1, Yes	Follow up question to [CP_phenotype]
degenerative	Mandatory	Is the individual's clinical course degenerative?	0, No 1, Yes	Follow up question to [CP_phenotype]
CP_dx	Core	Has the individual been diagnosed with CP?	0, No 1, Yes	
CP_dx_year	Recommended	What year was the individual diagnosed with CP?	YYYY	
confirmed_CP_dx	Core	Was the diagnosis of CP confirmed at age 5 or older?	0, No 1, Yes 2, No, individual is younger than 5 99, Unknown	
timing_CP_injury	Recommended	During what period did the individuals' CP-related brain disturbance occur?	1, Prenatal 2, Perinatal 3, Post-neonatal 99, Unknown	
ORDO	Core	Has the individual been diagnosed with a known syndrome?	3-6 numeric code	This ontology is built into Phenotips. Comma-separated list of ORDO codes or "blank" if patient phenotype is not associated to a clinical disorder.
OMIM	Core	Has the individual been diagnosed with a genetic disorder?	6-digit integer (minimum 100000)	This ontology is built into Phenotips. Comma-separated list of OMIM codes or "blank" if patient phenotype is not associated to a genetic disorder.
ICD10	Core	Please describe any medical conditions the individual may have (ICD-10 codes)	3-7 alphanumeric code with a single period (.) that follows the first 3 alphanumeric characters (e.g. F91.9)	Comma-separated list of ICD-10 codes or "blank" if patient phenotype is unknown.

Clinical symptoms and physical signs (32)

FIELD	CATEGORY	DESCRIPTION	VALUE	
primary_motor	Mandatory	What is the predominant motor type of the individuals CP?	1, Spastic 2, Dyskinetic - Dystonic 3, Dyskinetic - Choreoathetosis 4, Ataxic 5, Hypotonic	
primary_motor_laterality	Mandatory	What is the laterality of the individuals predominant motor type?	1, Unilateral 2, Bilateral	
prim_spast_topo	Core	What is the individuals' predominant spastic topography?	1, Left hemiplegia 2, Right hemiplegia 3, Diplegia 4, Quadriplegia	This can only be responded if [primary_motor] has a value of (1).
prim_dyskinesia_type	Exploratory	If dyskinetic is predominant motor type, is the movement:	1, Focal 2, Generalised	This can only be responded if [primary_motor] has a value of (2 or 3).
second_motor	Core	What is the secondary motor type that the individual presents with?	1, Spastic 2, Dyskinetic - Dystonic 3, Dyskinetic - Choreoathetosis 4, Ataxic 5, Hypotonic	
second_motor_laterality	Core	What is the laterality of the individuals' secondary motor type?	1, Unilateral 2, Bilateral	
second_spast_topo	Core	Additional description of dyskinesia presentation	1, Left hemiplegia 2, Right hemiplegia 3, Diplegia 4, Quadriplegia	This can only be responded if [second_motor] has a value of (1).
second_dyskinesia_type	Exploratory	What is the individuals' secondary spastic topography?	1, Focal 2, Generalised	This can only be responded if [second_motor] has a value of (2 or 3).
second_NDD	Core	Does the individual have another neurodevelopmental disability?	0, No 1, Yes 99, Unknown	

autism	Core	Does the individual have autism?	0, No 1, Yes 99, Unknown	
autism_severity	Core	If the individual has an autism, what is the severity of the autism? Refer to DSM-5	1=Level 1: Requiring support 2=Level 2: Requiring substantial support 3=Level 3: Requiring very substantial support	This can only be responded if [autism] has a value of (1).
ADHD	Core	Does the individual have ADHD?	0, No 1, Yes 99, Unknown	
ADHD_type	Core	If the individual has ADHD, what type of ADHD?	1=Primarily Hyperactive-Impulsive ADHD 2=Primarily Inattentive ADHD (formerly called ADD) 3=Combined Type ADHD	This can only be responded if [ADHD] has a value of (1).
ADHD_severity	Core	If the individual has an ADHD, what is the severity of the ADHD? Refer to DSM-5	1= Mild 2=Moderate 3=Severe	This can only be responded if [ADHD] has a value of (1).
epilepsy	Core	Does the individual have epilepsy?	0, No 1, Yes 99, Unknown	
epilepsy_type	Core	If the individual has epilepsy, what type of epilepsy?	free text	This can only be responded if [epilepsy] has a value of (1).
ID	Core	Does the child have an intellectual impairment?	0, No 1, Yes 99, Unknown	
ID_severity	Core	If the individual has an intellectual impairment, what is the severity of the impairment? (ICD10 codes F70 to F73)	1. Mild (IQ50 - 69) HP: 0001256 2. Moderate (IQ 35 - 49) HP: 0002342 3. Severe (IQ 20 - 34) HP: 0010864 4. Profound (IQ < 20) HP: 0002187 5. Impairment unspecified (IQ < 50)	This can only be responded if [ID] has a value of (1).
second_NDD_other	Core	If other, please specify:	free text	

visual_impairment	Core	Does the individual have a visual impairment?	0, No 1, Yes 99, Unknown	
visual_severity	Core	If the individual has a visual impairment, please describe:	free text	This can only be responded if [visual_impairment] has a value of (1).
hearing_impairment	Core	Does the individual have a hearing impairment?	0, No 1, Yes 99, Unknown	
hearing_severity	Core	If the individual has a hearing impairment, please describe:	free text	This can only be responded if [hearing_severity] has a value of (1).
congenital_anomalies	Core	Does the individual have any congenital anomalies?	0, No 1, Yes 99, Unknown	
congenital_anomalies_type	Core	If the individual has a congenital anomaly/ies, which major anatomical system/s are involved?	1, Nervous system HP: 0000707 2, Cardiovascular system HP: 0001626 3, Respiratory system HP: 0002086 4, Gastrointestinal system HP: 0011024 5, Genital organs HP: 0000078 6, Urinary system HP: 0000079 7, Musculature system HP: 0003011 8, Skeletal system HP: 0000924 9, Integumentary system (skin) HP: 0001574 10, Eye HP: 00000478 11, Ear HP: 0000598 12, Nose HP: 0000366	This can only be responded if [congenital_anomalies] has a value of (1). Comma separated list of all systems involved.
congenital_anomalies_detail	Recommended	Congenital anomalies details [free text]	free text	This can only be responded if [congenital_anomalies] has a value of (1). Watch for personal identifiers
radiology_text	Recommended	Radiology findings [free text]	free text	Watch for personal identifiers
chronic_pain	Exploratory	Has the individual reported experiencing chronic pain?	0, No 1, Yes - self report	

			2, Yes - proxy report 99, Unknown	
chronic_pain_age	Exploratory	At what age was chronic pain reported?	XX	Integer
chronic_pain_measure	Exploratory	What validated outcome measure was used to assess chronic pain?	free text	
positive_HPOs	Core	Please list positive HPO traits	HP:0100277	Comma-separated list of all observed HPO traits in the individual. Minimum of 3, preferably 5.
negative_HPOs	Exploratory	Please list phenotypic traits that are NOT present	HP:0012447	Comma-separated list of clinically important HPO traits that are absent in the individual.

CP-Specific Assessments (13)

FIELD NAME	CATEGORY	DESCRIPTION	VALUE	NOTES / LOGIC
GM	Recommended	Was a General Movements Assessment performed at 12 weeks of age, and if so what was the score?	0, No 1, Yes - normal 2, Yes - Abnormal fidgety 3, Yes - Absent fidgety	
HINE	Recommended	Was the Hammersmith Infant Neurological Examination (HINE) performed?	0, No 1, Yes	
HINE_age	Recommended	If the HINE was performed, at what age was the assessment performed?	MM	This can only be responded if [HINE] has a value of (1). Corrected age in months between 2-24 (integer)
HINE_outcome	Recommended	If the HINE was performed, what was the outcome of the assessment?	0-78	This can only be responded if [HINE] has a value of (1). Global score between 0-78 (Integer)
GMFCS	Mandatory	Gross Motor Functioning Classification System (GMFCS)© Score	1, GMFCS I 2, GMFCS II 3, GMFCS III 4, GMFCS IV 5, GMFCS V 99, Unknown	
BFMF	Recommended	Bimanual Fine Motor Function (BFMF) ©Score	1, BFMF I 2, BFMF II 3, BFMF III 4, BFMF IV 5, BFMF V	
MACS	Recommended	Manual Ability Classification System (MACS)© Score	1, MACS I 2, MACS II 3, MACS III 4, MACS IV 5, MACS V	

CFCS	Recommended	Communication Function Classification System (CFCS)© Score	1, CFCS I 2, CFCS II 3, CFCS III 4, CFCS IV 5, CFCS V
VFCS	Exploratory	Visual Function Classification System (VFCS)© Score	1, VFCS I 2, VFCS II 3, VFCS III 4, VFCS IV 5, VFCS V
MRICS	Core	Predominant Brain Pattern (Expanded MRI Classification System)	1, (A) Maldevelopments 2, (A.1) Maldevelopments - Disorders of cortical development (proliferation and/or migration and/or organisation) 3, (A.2) Maldevelopments - Other (ex: holoprosencephaly, Dandy-Walker formation, corpus callosum agenesis, cerebellar hypoplasia) 3, (B) Predominant White Matter Injury (PWMI) 4, (B.1) PWMI - PVL (mild/severe) 4, (B.2) PWMI - Sequelae of IVH or PVH infarction 5, (B.3) PWMI - Combination of PVL and IVH sequelae 6, (C) Predominant Grey matter Injury (PGMI) 7, (C.1) PGMI - Basal ganglia/thalamus lesions (mild/moderate/severe) 8, (C.2) PGMI - cortico-subcortical lesions only (watershed lesions in parasagittal distribution/multicystic encephalomalacia) not covered under C3 9, (C.3) PGMI - Arterial infarctions (middle cerebral artery/other) 10, (D) Miscellaneous changes (ex: cerebellar atrophy, cerebral atrophy, delayed myelination, ventriculomegaly not covered under B, haemorrhage not covered under B, brainstem lesions, calcifications) 11, (E) Normal 99, (Z) Unknown
swallowing	Recommended	Participant eating/drinking/swallowing	1, Standard mean for age - no modifications required 2, Requires modified diet 99, Unknown

VIKING	Recommended	Viking Speech Scale© Score	1, Score I 2, Score II 3, Score III 4, Score IV	
ICF	Recommended	WHO Disability Assessment Schedule 2.0	0-100	Summary disability profile score according to WHODAS2.0 (integer)

Family History (15)

FIELD NAME	CATEGORY	DESCRIPTION	VALUE	
mat_age	Core	Maternal age at time of birth	YY	Age at birth of proband (integer)
mat_cob	Recommended	Maternal Country of Birth	ISO-3166 2-alpha code	See Excel document
mat_edu	Recommended	Maternal Education	1, Primary school only 2, High school graduate or equivalent 3, Occupational/Technical/Vocational 4, Undergraduate university qualification 5, Postgraduate university qualification 6, Never attended 99, Unknown	
pat_age	Core	Paternal age at time of birth	YY (age at birth of proband)	
pat_cob	Recommended	Paternal Country of Birth	ISO-3166 2-alpha code	See Excel document
pat_edu	Recommended	Paternal Education	1, Primary school only 2, High school graduate nor equivalent 3, Occupational/Technical/Vocational 4, Undergraduate university qualification 5, Postgraduate university qualification 6, Never attended 99, Unknown	
consanguinity	Core	Is any parental consanguinity reported?	0, No 1, Yes 99, Unknown	
CP_family_hx	Core	Is there a family history of CP?	0, No 1, Yes 99, Unknown	

CP_family_hx_detail	Recommended	If there is a family history of CP, please select all members that have been diagnosed with CP?	<p>comma separated list of all:</p> <ul style="list-style-type: none"> 0, None 1, Daughter 2, Granddaughter 3, Grandson 4, Half-brother 5, Half-sister 6, Maternal Aunt 7, Maternal cousin 8, Maternal grandfather 9, Maternal Grandmother 10, Maternal Uncle 11, Brother 12, Father 13, Mother 14, Sister 15, Nephew 16, Niece 17, Paternal Aunt 18, Paternal Cousin 19, Paternal Grandfather 20, Paternal Grandmother 21, Paternal Uncle 22, Son 99, Unknown 	<p>This can only be responded if [CP_family_hx] has a value of (1).</p> <p>Any value between 23-98 or text should generate an error</p>
neuro_family_hx	Core	Is there a family history of neurological disorders?	<ul style="list-style-type: none"> 0, No 1, Yes 99, Unknown 	

neuro_family_hx_detail	Recommended	If there is a family history of neurological disorders, please list all family members that have been diagnosed with a neurological disorder:	0, None 1, Daughter 2, Granddaughter 3, Grandson 4, Half-brother 5, Half-sister 6, Maternal Aunt 7, Maternal cousin 8, Maternal grandfather 9, Maternal Grandmother 10, Maternal Uncle 11, Brother 12, Father 13, Mother 14, Sister 15, Nephew 16, Niece 17, Paternal Aunt 18, Paternal Cousin 19, Paternal Grandfather 20, Paternal Grandmother 21, Paternal Uncle 22, Son 99, Unknown	This can only be responded if [neuro_family_hx] has a value of (1). Comma separated list of all family members with relevant neurology history. Any value between 23-98, >100 or text should generate an error.
neuro_family_hx_detail_tet	Recommended	If yes, please describe all relevant family history.	Free text	This can only be responded if [neuro_family_hx] has a value of (1).
siblings	Recommended	Does the individual have sibling/s?	0, No 1, Yes 99, Unknown	
sib_number	Recommended	If the individual has sibling/s, how many siblings?	XX	This can only be responded if [siblings] has a value of (1) [integer].
sib_phenotype	Recommended	Are the sibling's phenotype(s) typical or atypical? (May be both)	1=Typical 2=Atypical	This can only be responded if [sibling] has a value of (1). Can be a comma-separated list with both 1, 2 if both phenotypes are present

Antenatal and Neonatal Details (39)

FIELD NAME	CATEGORY	DESCRIPTION	VALUE	NOTES /LOGIC
mat_ht	Exploratory	What was the maternal height at beginning of pregnancy with this individual?	XXX	Metric [numeric].
mat_wt	Exploratory	What was the maternal weight at the beginning of the pregnancy with this pregnancy?	XXX	Metric [numeric]
gravidity	Exploratory	Total number of confirmed pregnancies prior to this individual	XX	0+ [integer]
preterm_birth	Core	Was there a preterm birth prior to this individual?	0, No previous preterm birth 1, Yes, there was preterm birth 99, Unknown	
perinatal_death	Core	Was there a perinatal death prior to this individual? (includes more than 20 completed weeks, still births and death during neonatal period)	0, No previous perinatal death 1, Yes, there was perinatal death 99, Unknown	
conception	Core	Was the individual a product of assisted conception?	1, No 2, Yes - Unknown 3, Yes - IVF 4, Yes - ICSI 5, Yes - GIFT 6, Yes - AI 7, Yes - Ovulation Stimulation only 8, Yes - Other 99, Unknown	
first_an_visit	Recommended	Total number of completed weeks of the pregnancy at first antenatal visit	XX	1-39 completed weeks [integer]
mat_principal_morbid	Recommended	Pre-existing maternal morbidity, principal diagnosis	free text	
mat_add_morbid	Recommended	Pre-existing maternal morbidity, additional diagnoses	free text	
gestation_principal_morbid	Recommended	Maternal morbidity (pregnancy - and birth-related), principal diagnosis	free text	

gestation_add_morbid	Recommended	Maternal morbidity (pregnancy- and birth-related), additional diagnoses	free text	
G_smoke	Recommended	Did the mother smoke during pregnancy?	0, No 1, Yes 99, Unknown	
G_alcohol	Recommended	How many standard alcoholic drinks did the mother consume per week during pregnancy?	XX	integer
G_drug	Recommended	During the first trimester, did the mother use any recreational drugs?	0, No 1, Yes 99, Unknown	
teratogen	Recommended	Please describe any teratogen exposure prior to, or during pregnancy	free text	
fetal_abnormality	Core	Was a fetal abnormality identified by ultrasound during pregnancy?	0, No 1, Yes 99, Unknown	
fetal_abnormal_detail	Recommended	If yes, please describe	free text	This can only be responded if [fetal_abnormality] has a value of (1).
birth_facility	Exploratory	What facility was the individual born in?	1, Hospital 2, Birth centre attached to hospital 3, Birth centre free standing 4, Home birth planned 5, Home birth unplanned 6, Born before arrival at hospital 7, Born outside home/hospital without medical assistance 8, Other 99, Unknown	
mag_sulphate	Exploratory	Was magnesium sulphate administered to the mother during pregnancy?	0, No 1, Yes 99, Unknown	

labour_onset	Recommended	Onset of labour during this individual's birth	1, Spontaneous 2, Induced 3, No labour 99, Unknown	
labour_hyperthermia	Exploratory	Did the mother treat with hyperthermia during labour?	0, No 1, Yes 99, Unknown	
birth_presentation	Recommended	Presentation at birth	1, Vertex 2, Breech 3, Face 4, Brow 5, Transverse 99, Unknown	
delivery	Recommended	Method of delivery	1, Vaginal non-instrumental 2, Vaginal instrumental 3, Emergency c-section 4, Elective c-section 99, Unknown	
ga	Core	Gestational age (completed weeks)	20-46	Integer
bw	Core	Birth weight (grams)	200-5000	Metric (integer)
head_cir	Core	Head circumference of individual at time of birth (centimetre)	10-60	Metric (numeric)
plurality	Core	Birth plurality	1, Singleton 2, Twin 3, Triplets 4, Quadruplets 5, Quintuplets 6, Sextuplets 7, Other 99, Unknown	

birth_order	Recommended	Birth order if a multiple birth	1, First of multiple 2, Second of multiple 3, Third of multiple 4, Fourth of multiple 5, Fifth of multiple 6, Sixth of multiple 7, Other 99, Unknown	This can only be responded if [plurality] has a value of (2+).
zygotic	Core	If this was a multiple birth, was it:	1, Monozygotic 2, Dizygotic 3, Other 99, Unknown	This can only be responded if [plurality] has a value of (2+).
amniotic	Core	If this was a multiple birth, was it:	1, Monoamniotic 2, Diamniotic 3, Triamniotic 4, Other 99, Unknown	This can only be responded if [plurality] has a value of (2+).
chorionic	Core	If this was a multiple birth, was it:	1, Monochorionic 2, Dichorionic 3, Trichorionic 4, Other 99, Unknown	This can only be responded if [plurality] has a value of (2+).
newborn_care	Core	Did the individual receive more than routine newborn care?	0, No 1, Yes, NICU or special care 99, Unknown	
phototherapy	Recommended	Did the individual receive phototherapy?	0, No 1, Yes	
hypothermia	Core	Did the individual receive therapeutic hypothermia?	0, No 1, Yes 99, Unknown	
ventilation	Core	Did the individual receive mechanical ventilation?	0, No 1, Yes 99, Unknown	

neo_infection	Core	Was infection present during the neonatal period?	0, No 1, Yes 99, Unknown	
placenta_wt	Recommended	Placental weight (trimmed of extra placental membranes and umbilical cord) (grams)	10-3000	Metric [numeric]
gross_placental	Exploratory	Gross placental abnormalities detected	free text	
histo_placental	Exploratory	Histological placental abnormalities detected	free text	