**Intellectual disabilities Algorithm**

**Phenotype definition**

Case inclusion criteria:

 -Must be 731 days old or older

-Two or more relevant ICD-9 or ICD10 codes for intellectual disabilities (Table 1) in two or more in-person visits.

Case exclusion criteria:

 - Any ICD9-ICD10 codes shown in Table 2

Covariates:

-Age at diagnosis

-Gender

-Race

-Ethnicity

-Other relevant diagnosis codes shown in Table 3

**Controls definition**

**-**Must be 731 days old or older

-No diagnosis codes for intellectual disabilities (Table 1)

-No diagnosis codes shown in Table 4.

-Must have two or more in person visits after 731 days old

 Visits must be for well child (pediatrics)

Covariates:

-Gender

-Race

-Ethnicity

-Other relevant diagnosis shown in Table 3.

**Table 1**. Case inclusion codes

|  |  |
| --- | --- |
| **ICD 9** | **Description** |
| 317 | Mild intellectual disabilities |
| 318.0 | Moderate mental retardation |
| 318.1 | Severe mental retardation |
| 318.2 | Profound mental retardation |
| 319 | Unspecified intellectual disabilities |
| **ICD10** | **Description** |
| F70 | Mild intellectual disabilities |
| F71 | Moderate intellectual disabilities |
| F72 | Severe intellectual disabilities |
| F73 | Profound intellectual disabilities |
| F78 | Other intellectual disabilities |
| F79 | Unspecified intellectual disabilities |
| R41.83 | Borderline intellectual functioning |

**Table 2.** Case exclusion codes

|  |  |  |
| --- | --- | --- |
| **ICD9** | **ICD10** | **Description** |
| 046.x | A81.81, A81.01, A81.00, A81.09, A81.1, A81.2, A81.82, A81.83, A81.89, A81.9 | Slow virus infection of central nervous system |
| 047.x | A87.0, A87.8, A87.9 | Meningitis due to enterovirus |
| 191.x | C71.0-C71.9 | Malignant neoplasm of brain |
| 270.x | E70, E71, E72 | Disorders of amino-acid transport and metabolism |
| 271.x | E74 | Disorders of carbohydrate transport and metabolism |
| 272x | E78, E88.1, E77 | Disorders of lipoid metabolism |
| 277.1-277.9 | E85, E76 | Other disorders of purine and pyrimidine metabolism  |
| 279.11 | D82.1 | DiGeorge syndrome |
| 323.6 | G04.00, G04.01, G04.30, G04.31, G04.39, G05.4 | Postinfectious encephalitis |
| 323.9 | G04.90, G04.91 | Encephalitis, unspec. |
| 330.8 | F84.2 | Rett’s syndrome |
| 348.1 | G93.1 | Anoxic brain damage |
| 348.4 | G93.5 | Compression of brain |
| 348.5 | G93.6 | Cerebral edema |
| 349.8x | G96.9 | Other specified disorders of nervous system |
| 357.x | G61.0, G61.81, G62.81, G61.82, G61.9, G63, E08.42, E09.42, E10.42, E11.42, E13.42, G62.1, G62.0, G62.2 | Inflammatory and toxic neuropathy |
| 758.x | Q90-Q99 | Chromosomal anomalies |
| 759.5 | Q85.1 | Tuberous sclerosis |
| 759.81 | Q87.1 | Prader-Willi syndrome |
| 759.83 | Q99.2 | Fragile X syndrome |
| 760.x | P00, P04 | Fetus or newborn affected by maternal conditions which may be unrelated to present pregnancy |
| 761.x | P01 | Fetus or newborn affected by maternal complications of pregnancy |
| 762.x | P02 | Fetus or newborn affected by complications of placenta cord and membranes |
| 768.x | P84, P19, P91 | Intrauterine hypoxia and birth asphyxia |
| 779.5 | P96 | Drug withdrawal syndrome in newborn |
| 799.0x | R09.01 | Asphyxia |
| 799.1x | R09.2 | Respiratory arrest |
| 995.81x | T74 | Adult physical abuse |
| V42 | Z94 |  Organ or tissue replaced by transplant |

Table 3. Covariates for cases and controls

|  |  |  |  |  |
| --- | --- | --- | --- | --- |
| **ICD9** | **ICD10** | **Description** | **Cases** | **Controls** |
| 048.x | A88.0 | Other enterovirus diseases of central nervous system | x |  |
| 049.x | A87.2, A87.1, A85.0, A85.1, A85.8, A88.8, A86, A89 | Other non-arthropod-borne viral diseases of central nervous system | x |  |
| 070.x | B15-B19 | Viral hepatitis | x |  |
| 094 | A52.0, A52.1, A52.2, A52.3, A52.7, A52.8, A52.9  | Neurosyphilis | x |  |
| 192.x | C72.0-C72.9 | Malignant neoplasm of other and unspecified parts of nervous system | x |  |
| 290.x | F01, F02, F03 | Dementias | x | x |
| 291.x | F10 | Alcohol induced dementia | x | x |
| 292.x | F11-F19 | Drug induced mental disorders | x | x |
| 293.x | F05, F06 | Transient mental disorders due to conditions classified elsewhere | x | x |
| 294.0 | F04 | Amnestic disorders in conditions classified elsewhere | x | x |
| 294.1x | F02.80, F02.81 | Dementia in conditions classified elsewhere | x | x |
| 294.2 | F03.90, F03.91 | Dementia, unspecified | x | x |
| 299.x | F84 | Pervasive developmental disorder | x |  |
| 310.0 | F07.0 | Frontal lobe syndrome | x |  |
| 310.1 | F07.0 | Organic personality syndrome | x |  |
| 310.2 | F07.81 | Postconcussional syndrome | x |  |
| 315.x | F80, F81, F82, F89 | Specific delays in development | x |  |
| 332.x | G20, G21 | Parkinson’s disease | x |  |
| 333.1 | G25.0 | Essential tremor | x |  |
| 334.x | G11 | Spinocerebellar disease | x |  |
| 335.x | G12 | Anterior horn cell disease | x |  |
| 336.x | G95, G32.0, G99.2 | Other diseases of spinal cord | x |  |
| 341.x | G35, G36, G37 | Other demyelinating diseases of central nervous system | x |  |
| 342.x | G81 | Hemiplegia and hemiparesis  | x |  |
| 343.x | G80 | Infantile cerebral palsy | x |  |
| 344.x | G82, G83 | Other paralytic syndromes | x |  |
| 345.x | G40 | Epilepsy and recurrent seizures | x |  |
| 347.x | G47 | Cataplexy and narcolepsy | x |  |
| 348.3x | G93.4 | Other and unspecified encephalopathy | x |  |
| 356.x | G60 | Hereditary and idiopathic peripheral neuropathy | x |  |
| 358.0 | G70 | Myasthenia gravis | x |  |
| 359.x | G71, G72.2, G72.3, G72.41, G72.49, G72.81, G72.89, G72.9 | Muscular dystrophies and other myopathies | x |  |
| 430-437x | I60-I69 | Cerebrovascular disease | x |  |
| 438.x | I69 | Late effects of cerebrovascular disease | x |  |
| 663.1 | O69.1 | Cord around neck with compression complicating labor and delivery | x |  |
| 647.x | O98.x | Infectious and parasitic conditions in the mother | x | x |
| 648.x | O24 | Other current conditions in the mother classifiable elsewhere | x | x |
| 649.3 | O99.119 | Coagulation defects complicating pregnancy, childbirth, or the puerperium | x | x |
| 649.4 | 099.355 | Epilepsy complicating pregnancy, childbirth, or the puerperium | x | x |
| 678.x, 679.1 | O35, O26.889 | Other maternal and fetal complications | x | x |
| 734 | M21.40 | Flat foot | x | x |
| 735.x | M20 | Acquired deformities of fingers and toes | x | x |
| 736.x | M21 | Other acquired deformities of limbs | x | x |
| 737.x | M40, M41, M96 | Curvature of spine | x | x |
| 738.x | M95 | Other acquired musculoskeletal deformity  | x | x |
| 740.x-757.x | Q00-Q89 | Congenital malformations, deformations and chromosomal abnormalities | x | x |
| 760.x | P00, P04 | Fetus or newborn affected by maternal conditions which may be unrelated to present pregnancy |  | x |
| 761.x Except 761.6 | P01 | Fetus or newborn affected by maternal complications of pregnancy |  | x |
| 762.xExcept 762.3 | P02 | Fetus or newborn affected by complications of placenta cord and membranes |  | x |
| 763 | P03 | Fetus or newborn affected by other complications of labor and delivery | x | x |
| 764.x | P05 | Slow fetal growth and fetal malnutrition  | x | x |
| 767.x | P10-P14 | Birth trauma | x | x |
| 769.x | P22 | Respiratory distress syndrome in newborn | x | x |
| 770.x | P24 | Other respiratory conditions of fetus and newborn | x | x |
| 771.0-771.2 | P35-P37 | Infections specific to the perinatal period | x |  |
| 772.x | P50, P51, P52 | Fetal and neonatal hemorrhage | x |  |
| 773.x | P55 | Hemolytic disease of fetus or newborn, due to isoimmunization | x |  |
| 776.0x | P53 | Hemorrhagic disease of newborn | x |  |
| 776.2x | P60 | Disseminated intravascular coagulation in newborn | x |  |
| 779.0 | P90 | Convulsions of newborn | x |  |
| 779.1 | P91.811, P91.819, P91.88, P91.9 | Other and unspecified cerebral irritability in newborn | x |  |
| 779.2 | P91.4, P91.5 | Cerebral depression, coma, and other abnormal cerebral signs in fetus or newborn | x |  |
| 779.85 | P29.81 | Cardiac arrest of newborn | x |  |
| 780.0x | R40.20, R40.4, R40.3, R40.0 | Alteration of consciousness | x |  |
| 780.3x | R56.00, R56.01, R56.1, R56.9 | Convulsions | x |  |
| 780.93 | R41.2, R41.3 | Memory loss | x |  |
| 781.x | R25, R26, R27 | Symptoms involving nervous and musculoskeletal systems | x |  |
| 798.x | R99 | Sudden death | x |  |
| 800-804 | S02 | Fracture of skull | x |  |
| 851-854 | S06 | Intracranial Injury, excluding those with Skull Fracture | x | x |
| 950-954 | S04, S14 | Injury to Nerves And Spinal Cord | x | x |
| 960-979 | T36-T50 | Poisoning by, adverse effect of and underdosing of drugs, medicaments and biological substances | x | x |
| 980-989 | T51-T65 | Toxic Effects Of Substances Chiefly Nonmedicinal As To Source | x | x |
| V18.4 | Z81.0 | Family history of intellectual disabilities | x |  |

Table 4. Control exclusion codes

|  |  |  |
| --- | --- | --- |
| **ICD9** | **ICD10** | **Description** |
| 046.x | A81.81, A81.01, A81.00, A81.09, A81.1, A81.2, A81.82, A81.83, A81.89, A81.9 | Slow virus infection of central nervous system |
| 047.x | A87.0, A87.8, A87.9 | Meningitis due to enterovirus |
| 048.x | A88.0 | Other enterovirus diseases of central nervous system |
| 049.x | A87.2, A87.1, A85.0, A85.1, A85.8, A88.8, A86, A89 | Other non-arthropod-borne viral diseases of central nervous system |
| 070.x | B15-B19 | Viral hepatitis |
| 094 | A52.0, A52.1, A52.2, A52.3, A52.7, A52.8, A52.9  | Neurosyphilis |
| 191.x | C71.0-C71.9 | Malignant neoplasm of brain |
| 192.x | C72.0-C72.9 | Malignant neoplasm of other and unspecified parts of nervous system |
| 270.x | E70, E71, E72 | Disorders of amino-acid transport and metabolism |
| 271.x | E74 | Disorders of carbohydrate transport and metabolism |
| 272x | E78, E88.1, E77 | Disorders of lipoid metabolism |
| 277x  | E84, E85, E76 | Other disorders of purine and pyrimidine metabolism  |
| 279.11 | D82.1 | DiGeorge syndrome |
| 294.8 | F06.0 | Other persistent mental disorders due to conditions classified elsewhere |
| 294.9 | F06.8 | Other specified mental disorders due to known physiological condition |
| 295.x | F20, F25 | Schizophrenic disorders |
| 297.x | F22 | Delusional disorders |
| 299.x | F84 | Pervasive developmental disorder |
| 310.0 | F07.0 | Frontal lobe syndrome |
| 310.1 | F07.0 | Organic personality syndrome |
| 315.x | F80, F81, F82 | Specific delays in development |
| 323.6 | G04.00, G04.01, G04.30, G04.31, G04.39, G05.4 | Postinfectious encephalitis |
| 323.9 | G04.90, G04.91 | Encephalitis, unspec. |
| 330.x | E75  | Cerebral degenerations usually manifest in childhood |
| 331.x | G30.9, G31, G91,  | Other cerebral degenerations |
| 332.x | G20 | Parkinson's disease |
| 333.2 | G25.3 | Myoclonus |
| 333.4 | G10 | Huntington's chorea |
| 334.x | G11 | Spinocerebellar disease |
| 335.x | G12 | Anterior horn cell disease |
| 336.x | G95, G32.0, G99.2 | Other diseases of spinal cord |
| 341.x | G35, G36, G37 | Other demyelinating diseases of central nervous system |
| 342.x | G81 | Hemiplegia and hemiparesis  |
| 343.x | G80 | Infantile cerebral palsy |
| 344.x | G82, G83 | Other paralytic syndromes |
| 345.x | G40 | Epilepsy and recurrent seizures |
| 348.x | G93 | Other conditions of brain |
| 349.8x | G96.9 | Other specified disorders of nervous system |
| 357.x | G61.0, G61.81, G62.81, G61.82, G61.9, G63, E08.42, E09.42, E10.42, E11.42, E13.42, G62.1, G62.0, G62.2 | Inflammatory and toxic neuropathy |
| 358.0 | G70 | Myasthenia gravis |
| 359.x | G71, G72.2, G72.3, G72.41, G72.49, G72.81, G72.89, G72.9 | Muscular dystrophies and other myopathies |
| 430-438x | I60-I69 | Cerebrovascular disease |
| 647.x | O98.x | Infectious and parasitic conditions in the mother |
| 648.x | O24 | Other current conditions in the mother classifiable elsewhere |
| 663.1 | O69.1 | Cord around neck with compression complicating labor and delivery |
| 740.x | Q00 | Anencephalus and similar anomalies |
| 741.x | Q05 | Spina bifida |
| 742.x | Q01, Q02, Q03, Q04, Q06 | Other congenital anomalies of nervous system |
| 756.17 | Q76.0 | Spina bifida occulta |
| 758.x | Q90-Q99 | Chromosomal anomalies |
| 759.x | Q87, Q89 | Other and unspecified congenital anomalies |
| 761.6 | P01.6 | Maternal death affecting fetus or newborn |
| 762.3 | P02.3 | Placental transfusion syndromes affecting fetus or newborn |
| 768.7x | P91.60, P91.61, P91.62, P91.63 | Hypoxic-ischemic encephalopathy |
| 771.0-771.2 | P35-P37 | Infections specific to the perinatal period |
| 772.x | P50, P51, P52 | Fetal and neonatal hemorrhage |
| 773.x | P55 | Hemolytic disease of fetus or newborn, due to isoimmunization |
| 776.0x | P53 | Hemorrhagic disease of newborn |
| 776.2x | P60 | Disseminated intravascular coagulation in newborn |
| 779.0 | P90 | Convulsions of newborn |
| 779.1 | P91.811, P91.819, P91.88, P91.9 | Other and unspecified cerebral irritability in newborn |
| 779.2 | P91.4, P91.5 | Cerebral depression, coma, and other abnormal cerebral signs in fetus or newborn |
| 779.85 | P29.81 | Cardiac arrest of newborn |
| 780.0x | R40.20, R40.4, R40.3, R40.0 | Alteration of consciousness |
| 780.3x | R56.00, R56.01, R56.1, R56.9 | Convulsions |
| 780.93 | R41.2, R41.3 | Memory loss |
| 781.x | R25, R26, R27 | Symptoms involving nervous and musculoskeletal systems |
| V18.4 | Z81.0 | Family history of intellectual disabilities |
| V42 | Z94 |  Organ or tissue replaced by transplant |