Public Query Report

Cohort of Patients with Intellectual and Developmental Disabilities Across Clinical Research Networks Participating in PCORnet®, The National Patient-Centered Clinical Research Network.

Rationale for Network Query of PCORnet Data Resources:

This network query of PCORnet data resources was requested by the Patient-Centered Outcomes Research Institute® (PCORI®) in collaboration with the PCORnet Network Partners. Network queries are developed, distributed, and processed through the PCORnet® Front Door, which is the point of contact and manages all data network requests.

In 2019, the U.S. Congress passed legislation that reauthorized funding for PCORI, the founding and primary funder of PCORnet®, for 10 years, and identified intellectual and developmental disabilities (IDD) as a research priority. Current prevalence estimates for IDD-related conditions indicate that large numbers of children and adults in the US are living with IDD. Approximately 1 in every 6 children between the ages of 3 – 17 years old have at least one IDD diagnosis in the U.S.[1] In 2020, Larson et al. estimated there were 7.4 million people living with IDD in the U.S., of which approximately 5.3 million were children.[2]

A national-scale infrastructure- such as PCORnet has the capacity to support national health system research efforts on IDD. However, the prevalence of patients with IDD served by PCORnet partner networks and eligible to participate in research is unknown. PCORI requested an exploratory query of individuals with IDD to demonstrate the utility of the PCORnet data infrastructure to support future national-scale research on IDD, and to identify and characterize the cohort of patients with IDD served by health systems participating in PCORnet ("IDD cohort").

Background on PCORnet®:

PCORnet is a large, distributed "network of networks" (Figure 1) funded by PCORI to improve the nation's capacity to efficiently conduct definitive health research, particularly patient-centered comparative clinical effectiveness research (CER).

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[1] Cogswell ME CE, Tian LH, et al. Health Needs and Use of Services Among Children with Developmental Disabilities — United States, 2014–2018. 2022.

[2] Larson S TB, Sowers M, Bourne M Lou. In-Home and Residential Long-Term Supports and Services for Persons with Intellectual or Developmental Disabilities: Status and Trends Through 2017.; 2020.

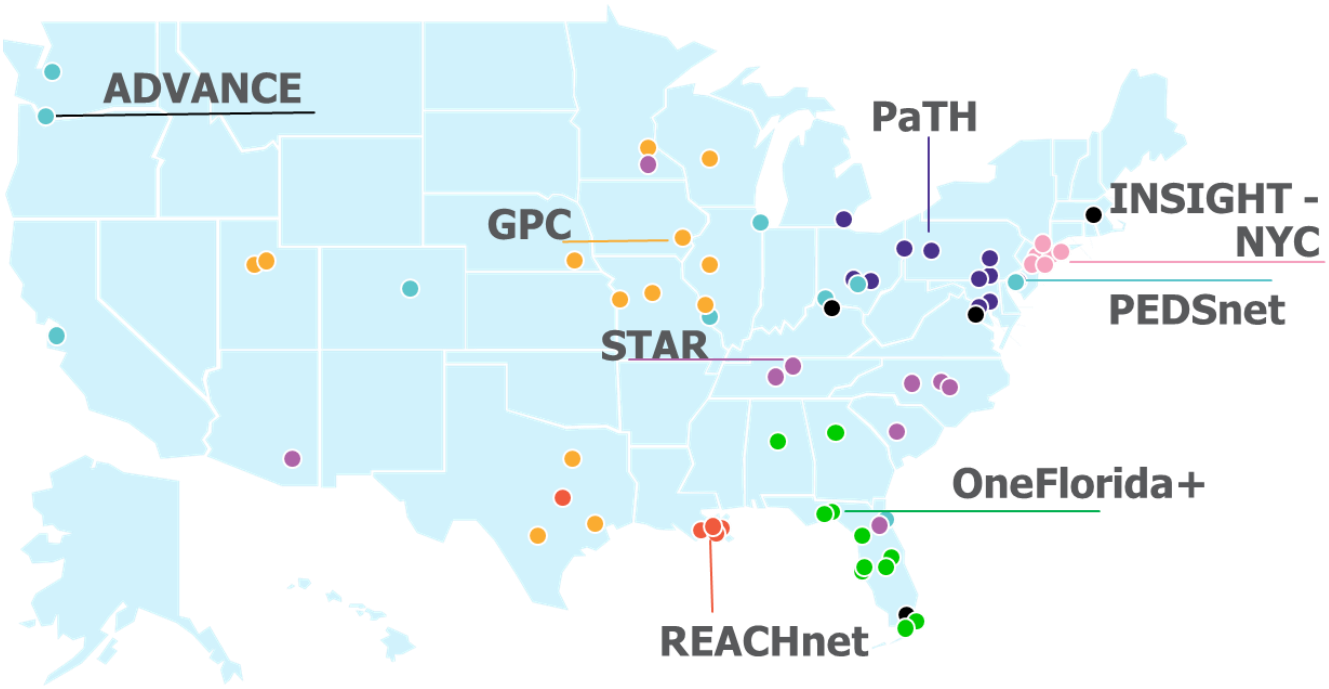

Figure 1. Clinical Research Networks participating in PCORnet, June 2023. Source: Developed by the Duke Clinical Research Institute (DCRI) with funding through a PCORI Award (RI-DCRI-01-PS3).

PCORnet consists of 63 data contributing partners[3] across eight Clinical Research Networks (CRNs), in addition to patient partners and a Coordinating Center. Collectively, CRN data-contributing partners consist of more than thirteen thousand clinical sites across the U.S., including large academic health systems, hospitals, federally qualified health centers, and community clinics.

A unique feature of PCORnet is that all data contributing partners store a version of their clinical data in the same standardized data model, the PCORnet® Common Data Model (CDM). In this distributed network, data holders (e.g., health systems, clinics) maintain physical control, use, and manage the transfer of their data to the CRNs, the Coordinating Center for PCORnet and data requestors.

Query Description:

This query of PCORnet data resources describes the population of patients with IDD that had healthcare encounters at a partner site. This query is the largest known, national-scale descriptive analysis of IDD populations using electronic health record (EHR) data. The query results will inform how PCORnet can be used for patient-centered CER studies and trials on IDD and to inform opportunities to enhance PCORnet data resources for IDD research. In addition, the query aims to demonstrate the utility of PCORnet to:

[3] A data contributing partner may include multiple clinics, hospitals, health networks, and other care settings.

- Describe utilization of healthcare services by patients with IDD
- Describe the extent of co-occurring chronic diseases for people with IDD diagnoses
- Support broad cohort identification of the IDD population
- Develop digital classifiers for IDD using clinical data

Query Methodology, Criteria and Engagement:

The query of PCORnet data resources includes a cohort of patients with IDD-related conditions who had an encounter at a partner site participating in PCORnet during October 2019 – October 2022. Each patient included in the IDD cohort has two or more occurrences of a relevant diagnosis (as identified by diagnosis codes billed at a healthcare encounter) within a 10-year query period (October 2012 – October 2022). Two recorded diagnosis codes are required as a minimum threshold for confirming patients were treated for a specific condition to minimize misclassification. The Coordinating Center for PCORnet programmed and distributed this descriptive query to all 63 data contributing partners.

As with all PCORI topics, the development process includes a review of the relevant literature and engagement with stakeholders such as subject matter experts and patient partners. Patient partners and subject matter experts were engaged early in the query development to help identify the range of IDD-related conditions and important characteristics to describe the IDD cohort. Literature was then used to identify the diagnosis billing codes[4] to be included in the query. Patient and clinician stakeholders reviewed the query results and public query report prior to public dissemination.

The initial cohort criteria for the IDD query were defined as patients with:

- An encounter at a partner site in the last 3 years (October 2019 – October 2022); and
- Two or more occurrences of a relevant diagnosis within the last 10 years (October 2012 – October 2022)
- Relevant diagnoses restricted to one or more of the following 11 groups[5]:
    - Attention-Deficit/Hyperactivity Disorder (ADHD),
    - Autism Spectrum Disorder (ASD),
    - Congenital Malformation of the Brain,
    - Cerebral Palsy,
    - Down Syndrome,
    - Fetal Alcohol Syndrome,

[4] Key literature to identify scope of conditions included but was not limited to 1) Reichard A, Haile E, Morris A. Characteristics of Medicare Beneficiaries with Intellectual or Developmental Disabilities. *Intellect Dev Disabil*. 2019 Oct;57(5):405-420. doi: 10.1352/1934-9556-57.5.405.; 2) Lin E, et al. Using administrative health data to identify individuals with intellectual and developmental disabilities: a comparison of algorithms. *J Intellect Disabil Res*. 2013 May;57(5):462-77. doi: 10.1111/jir.12002.; 3) Landes SD et al. Risk Factors Associated With COVID-19 Outcomes Among People with Intellectual and Developmental Disabilities Receiving Residential Services. *JAMA Netw Open*. 2021 Jun 1;4(6):e2112862. doi: 10.1001/jamanetworkopen.2021.12862.

[5] Code sets for all IDD-related condition groups are provided in Appendix A.

- Fragile X Syndrome,
- Inborn Metabolic Disorders presenting with Intellectual Disability,
- Intellectual Disability as specified in ICD-9 (317-319) and ICD-10 (F70-F79) [6],
- Spina Bifida, and
- Other Conditions presenting with Intellectual Disability.

The final patient cohort was filtered to include only patients with a visit during October 2019 – October 2022 in ambulatory, emergency, in-patient or telehealth care settings, and no record of death.

The query also included:

- demographic characteristics (e.g., race, age, sex, socioeconomic status),
- co-occurring IDD recorded diagnoses, and
- recorded diagnoses of common chronic diseases for the IDD population and psychosocial health disorders.

Results:

Sixty-one data contributing partners (96.8%) participating in PCORnet responded to the query request.

Table 1 provides details of the counts and demographic characteristics of the IDD cohort by the 11 groups of diagnoses.[7]

The most frequently recorded IDD-related condition within the IDD cohort was ADHD (n=1,074,213). The second most frequently recorded diagnosis of an IDD-related condition was ASD (n=321,700). The least commonly diagnosed condition within the cohort was Fragile X Syndrome (n=2,844).

Table 1 also includes details of healthcare utilization across the IDD cohort over the 3-year analysis period. Across the 11 groups in the IDD cohort, 19-34% of patient groups utilized the emergency department, 33-49% utilized telehealth services and 8-31% utilized in-patient services during the 3-year analysis period.

Table 2 provides details of unique patients records with co-occurring IDD-related conditions. Co-occurrence of IDD conditions was common. This table also includes recorded diagnoses of developmental delay and learning disabilities for the IDD Cohort

Table 3 provides details of unique patient records with IDD-related conditions and co-occurring chronic diseases, as well as psychosocial health disorders within the IDD cohort. Across all IDD conditions, diabetes and hypertension were prevalent. For example, depression was recorded for 31% of those with ADHD.

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[6] International Classification of Diseases (ICD)

[7] This query does not include a total count of patients with any IDD-related condition, but rather stratifies by specific IDD conditions.

Limitations:

Data and analyses presented are descriptive and derived from diagnosis codes collected during healthcare encounters in the EHR. Rows and percentages may not round due to missing values and or if counts are less than 10 they are reported as <10 to protect patient privacy and risk of identification from aggregate values as outlined in the Data Privacy Statement for PCORnet®.

No inferential testing was conducted to compare populations or test hypotheses, as these are descriptive data only. Limitations with any EHR data analysis are applicable to this data, such as the possibility for misclassification due to imperfect algorithms and lack of consistent definition of enrollment to define cohorts. Results should be interpreted with these limitations in mind.

To ensure PCORnet data resources are of high quality for research, activities in preparation for research (e.g., network query requests), and to mitigate the limitations above, all PCORnet-accessible data resources undergo rigorous quality curation and screening as part of quarterly coordinated data quality assessment.

Conclusion:

This query of PCORnet data resources is the largest known, national-scale descriptive analysis of IDD populations using EHR data. The results presented in this report provide researchers and patient/caregiver partners with information about the capacity of the PCORnet infrastructure to contribute to future studies on individuals with IDD. Results presented in this Public Query Report are informative to the public in a variety of ways, such as the extent of healthcare utilization by the IDD population or prevalence of common chronic diseases co-occurring with IDD.

Disclaimer:

PCORnet® is intended to improve the nation's capacity to efficiently conduct patient-centered health research, particularly CER, by providing a large, highly representative network of health data, research expertise, and patient insights. PCORnet has been developed with funding from the Patient-Centered Outcomes Research Institute® (PCORI®).

Network queries that return only aggregate or limited data sets are covered by the PCORnet® Master Data Sharing Agreement (version 4.0), and site-level blanket Institutional Review Board approvals.

The statements presented in this report do not necessarily represent the views of PCORI or other organizations participating in, collaborating with, or funding PCORnet.

For questions, comments or suggestions related to this PCORnet® Front Door query or other PCORnet queries, please contact the PCORnet® Front Door at frontdoor@pcornet.org.

Tables

Table 1. Demographic characteristics of the intellectual and developmental disabilities cohort across data-contributing partners participating in PCORnet (October 2019 – October 2022).

	ADHD[1]	Autism Spectrum Disorder	Inborn Metabolic Disorders presenting with ID[1]	Intellectual Disability, Coded	Cerebral Palsy	Congenital Malformations of the Brain	Down Syndrome	Other Conditions presenting with ID[1]	Spina Bifida	Fetal Alcohol Syndrome	Fragile X Syndrome
Number of unique patient records[2]	1,074,213	321,700	126,235	118,631	98,716	77,534	51,373	49,813	44,563	4,561	2,844
Mean Age (SD)	24.65 (14.04)	15.17 (10.09)	46.44 (21.91)	32.88 (16.77)	25.03 (16.16)	18.56 (16.90)	18.75 (14.46)	19.45 (15.93)	28.75 (17.97)	18.59 (10.30)	25.80 (16.50)
Female	42%	24%	56%	42%	45%	50%	47%	50%	60%	46%	28%
Race											
White	75%	64%	74%	64%	64%	62%	67%	69%	71%	64%	73%
Black or African American	12%	14%	10%	20%	17%	16%	11%	9%	11%	19%	10%
American Indian or Alaska Native	1%	1%	0%	1%	0%	1%	0%	0%	1%	4%	0%
Asian	1%	4%	2%	2%	2%	3%	3%	3%	3%	1%	1%
Native Hawaiian or Other Pacific Islander	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%	0%
Multiple Races[3]	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA	NA
Other[3]	11%	18%	13%	13%	15%	18%	19%	17%	15%	12%	16%
Hispanic											
Yes	10%	17%	11%	14%	15%	18%	20%	16%	15%	10%	14%
No	81%	74%	77%	80%	79%	76%	72%	78%	78%	85%	76%
Other	9%	9%	12%	7%	7%	6%	7%	7%	7%	5%	10%
Area Deprivation Index (ADI)[4]											
SES Q1	34%	30%	33%	23%	26%	27%	31%	34%	28%	29%	31%
SES Q2	19%	17%	19%	18%	17%	17%	17%	17%	19%	19%	18%
SES Q3	19%	19%	21%	24%	22%	21%	19%	19%	23%	20%	18%
SES Q4	13%	14%	13%	19%	16%	16%	14%	13%	15%	12%	12%
Missing	16%	20%	14%	16%	18%	18%	19%	17%	15%	20%	20%
Healthcare Utilization in the Past 3 Years											
Ambulatory	95%	94%	97%	95%	95%	95%	94%	96%	95%	94%	94%
Emergency Department	27%	26%	29%	34%	31%	34%	22%	28%	31%	35%	19%
Inpatient	8%	10%	19%	19%	23%	31%	18%	25%	27%	15%	11%
Telehealth	47%	47%	45%	49%	45%	47%	33%	47%	42%	46%	47%

[1]ADHD = Attention-Deficit Hyperactivity Disorder; ID = Intellectual Disability

[2]Patients may have multiple conditions and be represented more than once across conditions.

[3]For this particular query “Multiple Race” is included in “Other”. For all future queries, “Multiple Race” will be disaggregated from “Other”

[4]Area Deprivation Index (ADI): Patient 5-Digit Zip Codes are mapped to socioeconomic status by normalized Area Deprivation Index (ADI) value (0-100). Lower values are associated with lower deprivation and higher values are associated with higher deprivation. A ranking of 1 indicates the lowest level of "disadvantage" within the nation and an ADI with a ranking of 100 indicates the highest level of "disadvantage". In this table, values are grouped into quartiles using the count of zip codes. Quartile 1 (SES Q1) represents the lowest range of ADI values and Quartile 4 (SES Q4) represents the highest range of ADI values (Q1=0-38, Q2=39-43, Q3=44-49, and Q4=50-100). For additional information regarding the ADI index, see the Neighborhood Atlas here: https://www.neighborhoodatlas.medicine.wisc.edu/. Note that the Area Deprivation Index (ADI) is designed for validity at the 9-digit zip or census block group level rather than the 5-digit zip level.

Table 2. Percent of patients with co-occurring intellectual and developmental disabilities conditions across data-contributing partners participating in PCORnet (October 2019 – October 2022).

Reference IDD Condition	Unique Patient Records[2]	ADHD[1]	Autism Spectrum Disorder	Inborn Metabolic Disorders presenting with ID[1]	Intellectual Disability, Coded	Cerebral Palsy	Congenital Malformations of the Brain	Down Syndrome	Other Conditions Presenting with ID[1]	Spina Bifida	Fetal Alcohol Syndrome	Fragile X Syndrome	Developmental Delays	Learning Disabilities
ADHD[1]	1,074,213	-	9%	1%	2%	1%	1%	<1%	<1%	<1%	<1%	<1%	4%	10%
Autism Spectrum Disorder	321,700	30%	-	1%	10%	2%	2%	1%	1%	<1%	<1%	<1%	17%	33%
Inborn Metabolic Disorders presenting with ID[1]	126,235	4%	2%	-	2%	2%	1%	<1%	1%	<1%	<1%	<1%	3%	4%
Intellectual Disability Coded	118,631	21%	27%	2%	-	15%	8%	4%	4%	1%	1%	1%	15%	23%
Cerebral Palsy	98,716	6%	8%	2%	18%	-	15%	<1%	2%	2%	<1%	<1%	24%	17%
Congenital Malformations of Brain	77,534	8%	8%	2%	12%	19%	-	1%	6%	6%	<1%	<1%	27%	21%
Down Syndrome	51,373	4%	6%	1%	9%	1%	1%	-	1%	<1%	<1%	<1%	9%	17%
Other Conditions Presenting with ID[1]	49,813	8%	8%	2%	10%	4%	9%	1%	-	2%	<1%	<1%	17%	17%
Spina Bifida	44,563	6%	3%	1%	4%	4%	10%	<1%	2%	-	<1%	<1%	7%	7%
Fetal Alcohol Syndrome	4,561	50%	19%	1%	21%	5%	8%	1%	1%	1%	-	<1%	18%	27%
Fragile X Syndrome	2,844	26%	34%	1%	28%	2%	2%	<1%	3%	<1%	<1%	100%	13%	20%

[1]ADHD = Attention-Deficit Hyperactivity Disorder; ID = Intellectual Disability

[2]Patients may have multiple conditions and be represented more than once across conditions.

Table percentages are based on the Reference IDD Condition. Percentages should be read from left to right for each Reference IDD Condition. For example, there were 1,074,213 total unique patient records with a diagnosis of ADHD and 9% of these also have a recorded diagnosis of Autism Spectrum Disorder. There were 321,700 total unique patient records with a diagnosis of Autism Spectrum Disorder and 30% of these also have a recorded diagnosis of ADHD, 2% of patients with Autism also have a recorded diagnosis billing code for Cerebral Palsy.

Table 3. Percent of patients with common chronic diseases and psychosocial health conditions co-occurring with intellectual and developmental disabilities across data-contributing partners participating in PCORnet (October 2019 – October 2022).

Reference IDD Condition	Unique Patient Records[2]	Hypertension	Seizure Disorder	Congenital Heart Disease and Other Malformations of the Vascular System	Diabetes	Failure to thrive	Hip Dislocation, Displacement, or Subluxation	Torticollis	Anxiety	Bipolar	Depression	Post-Traumatic Stress Disorder
ADHD[1]	1,074,213	9%	5%	2%	3%	2%	<1%	<1%	46%	6%	31%	7%
Autism Spectrum Disorder	321,700	4%	15%	4%	2%	4%	<1%	1%	28%	3%	12%	2%
Inborn Metabolic Disorders presenting with ID[1]	126,235	41%	8%	4%	17%	3%	0%	1%	27%	3%	22%	3%
Intellectual Disability Coded	118,631	20%	37%	6%	11%	6%	2%	1%	33%	9%	19%	4%
Cerebral Palsy	98,716	12%	44%	7%	4%	11%	6%	2%	15%	2%	9%	1%
Congenital Malformations of Brain	77,534	10%	38%	14%	4%	16%	4%	2%	13%	1%	7%	1%
Down Syndrome	51,373	3%	8%	40%	4%	7%	1%	1%	8%	0%	3%	0%
Other Conditions Presenting with ID[1]	49,813	12%	16%	27%	4%	12%	2%	2%	14%	1%	7%	1%
Spina Bifida	44,563	17%	15%	6%	7%	4%	2%	1%	21%	2%	16%	2%
Fetal Alcohol Syndrome	4,561	7%	15%	8%	4%	10%	1%	1%	42%	10%	24%	13%
Fragile X Syndrome	2,844	9%	16%	3%	4%	3%	<1%	1%	33%	3%	9%	2%

[1]ADHD = Attention-Deficit Hyperactivity Disorder; ID = Intellectual Disability

[2]Patients may have multiple conditions and be represented more than once across conditions.

Table percentages are based on the Reference IDD Condition. Percentages should be read from left to right for each Reference IDD Condition. For example, there were 1,074,213 total unique patient records with a diagnosis of ADHD and 9% of these also have a recorded diagnosis of Hypertension, 5% have a recorded diagnosis of seizure disorder, etc.

APPENDIX A.

Code sets

Attention Deficit Hyperactivity Disorder Codes

ICD-9 Code	Description
314	Hyperkinetic syndrome of childhood
314.0	Attention deficit disorder of childhood
314.00	Attention deficit disorder without mention of hyperactivity
314.01	Attention deficit disorder with hyperactivity
314.1	Hyperkinesis with developmental delay
314.2	Hyperkinetic conduct disorder
314.8	Other specified manifestations of hyperkinetic syndrome
314.9	Unspecified hyperkinetic syndrome

ICD-10 Code

F90	Attention-deficit hyperactivity disorders
F90.0	Attention-deficit hyperactivity disorder, predominantly inattentive type
F90.1	Attention-deficit hyperactivity disorder, predominantly hyperactive type
F90.2	Attention-deficit hyperactivity disorder, combined type
F90.8	Attention-deficit hyperactivity disorder, other type
F90.9	Attention-deficit hyperactivity disorder, unspecified type

Autism Spectrum Disorder Codes

ICD-9 Code	Description
299.0	Autistic disorder
299.00	Autistic disorder, current or active state
299.01	Autistic disorder, residual state
299.1	Childhood disintegrative disorder
299.10	Childhood disintegrative disorder, current or active state
299.11	Childhood disintegrative disorder, residual state
299.8	Other specified pervasive developmental disorders
299.80	Other specified pervasive developmental disorders, current or active state
299.81	Other specified pervasive developmental disorders, residual state
299.9	Unspecified pervasive developmental disorder
299.90	Unspecified pervasive developmental disorder, current or active state
299.91	Unspecified pervasive developmental disorder, residual state

ICD-10 Code

F84	Pervasive developmental disorders
F84.0	Autistic disorder
F84.1	Atypical autism
F84.3	Other childhood disintegrative disorder
F84.4	Overactive disorder associated with mental retardation and stereotyped movements
F84.5	Asperger's syndrome
F84.8	Other pervasive developmental disorders
F84.9	Pervasive developmental disorder, unspecified

Congenital Malformations of Brain Codes

ICD-9 Code	Description
740	Anencephalus and similar anomalies
740.0	Anencephalus
740.1	Craniorachischisis
740.2	Iniencephaly
742	Other congenital anomalies of nervous system
742.0	Encephalocele
742.1	Microcephalus
742.2	Congenital reduction deformities of brain
742.3	Congenital hydrocephalus
742.4	Other specified congenital anomalies of brain

ICD-10 Code	
Q00	Anencephaly and similar malformations
Q00.0	Anencephaly
Q00.1	Craniorachischisis
Q00.2	Iniencephaly
Q01	Encephalocele
Q01.0	Frontal encephalocele
Q01.1	Nasofrontal encephalocele
Q01.2	Occipital encephalocele
Q01.8	Encephalocele of other sites
Q01.9	Encephalocele, unspecified
Q02	Microcephaly
Q03	Congenital hydrocephalus
Q03.0	Malformations of aqueduct of Sylvius
Q03.1	Atresia of foramina of Magendie and Luschka
Q03.8	Other congenital hydrocephalus
Q03.9	Congenital hydrocephalus, unspecified

Q04	Other congenital malformations of brain
Q04.0	Congenital malformations of corpus callosum
Q04.1	Arhinencephaly
Q04.2	Holoprosencephaly
Q04.3	Other reduction deformities of brain
Q04.4	Septo-optic dysplasia of brain
Q04.5	Megalencephaly
Q04.6	Congenital cerebral cysts
Q04.8	Other specified congenital malformations of brain
Q04.9	Congenital malformation of brain, unspecified

Cerebral Palsy Codes

ICD-9 Code	Description
333.71	Athetoid cerebral palsy
343	Infantile cerebral palsy
343.0	Congenital diplegia
343.1	Congenital hemiplegia
343.2	Congenital quadriplegia
343.3	Congenital monoplegia
343.4	Infantile hemiplegia
343.8	Other specified infantile cerebral palsy
343.9	Infantile cerebral palsy, unspecified

ICD-10 Code	
G80	Cerebral palsy
G80.0	Spastic quadriplegic cerebral palsy
G80.1	Spastic diplegic cerebral palsy
G80.2	Spastic hemiplegic cerebral palsy
G80.3	Athetoid cerebral palsy
G80.3	Dyskinetic cerebral palsy
G80.4	Ataxic cerebral palsy
G80.8	Other cerebral palsy
G80.9	Cerebral palsy, unspecified

Down Syndrome Codes

ICD-9 Code	Description
758.0	Down's syndrome

ICD-10 Code	
Q90	Down syndrome
Q90.0	Trisomy 21, nonmosaicism (meiotic nondisjunction)
Q90.1	Trisomy 21, mosaicism (mitotic nondisjunction)
Q90.2	Trisomy 21, translocation
Q90.9	Down syndrome, unspecified

Fetal Alcohol Syndrome Codes

ICD-9 Code	Description
760.71	Alcohol affecting fetus or newborn via placenta or breast milk
760.77	Anticonvulsants affecting fetus or newborn via placenta or breast milk

ICD-10 Code	
Q86.0	Fetal alcohol syndrome (dysmorphic)
Q86.1	Fetal hydantoin syndrome

Fragile X Syndrome Codes

ICD-9 Code	Description
759.83	Fragile X syndrome

ICD-10 Code	
Q99.2	Fragile X chromosome

Metabolic disorders presenting with Intellectual Disability Codes

ICD-9 Code	Description
270	Disorders of amino-acid transport and metabolism
270.0	Disturbances of amino-acid transport
270.1	Phenylketonuria [PKU]
270.2	Other disturbances of aromatic amino-acid metabolism
270.3	Disturbances of branched-chain amino-acid metabolism
270.4	Disturbances of sulphur-bearing amino-acid metabolism
270.5	Disturbances of histidine metabolism
270.6	Disorders of urea cycle metabolism
270.7	Other disturbances of straight-chain amino-acid metabolism
270.8	Other specified disorders of amino-acid metabolism
270.9	Unspecified disorder of amino-acid metabolism

271	Disorders of carbohydrate transport and metabolism
271.0	Glycogenosis
271.1	Galactosemia
271.2	Hereditary fructose intolerance
271.3	Intestinal disaccharidase deficiencies and disaccharide malabsorption
271.4	Renal glycosuria
271.8	Other specified disorders of carbohydrate transport and metabolism
271.9	Unspecified disorder of carbohydrate transport and metabolism
272.7	Lipidoses

ICD-10 Code

E70	Disorders of aromatic amino-acid metabolism
E70.0	Classical phenylketonuria
E70.1	Other hyperphenylalaninemias
E70.2	Disorders of tyrosine metabolism
E70.20	Disorder of tyrosine metabolism, unspecified
E70.21	Tyrosinemia
E70.29	Other disorders of tyrosine metabolism
E70.3	Albinism
E70.30	Albinism, unspecified
E70.31	Ocular albinism
E70.310	X-linked ocular albinism
E70.311	Autosomal recessive ocular albinism
E70.318	Other ocular albinism
E70.319	Ocular albinism, unspecified
E70.32	Oculocutaneous albinism
E70.320	Tyrosinase negative oculocutaneous albinism
E70.321	Tyrosinase positive oculocutaneous albinism
E70.328	Other oculocutaneous albinism
E70.329	Oculocutaneous albinism, unspecified
E70.33	Albinism with hematologic abnormality
E70.330	Chediak-Higashi syndrome
E70.331	Hermansky-Pudlak syndrome
E70.338	Other albinism with hematologic abnormality
E70.339	Albinism with hematologic abnormality, unspecified
E70.39	Other specified albinism
E70.4	Disorders of histidine metabolism
E70.40	Disorders of histidine metabolism, unspecified
E70.41	Histidinemia
E70.49	Other disorders of histidine metabolism
E70.5	Disorders of tryptophan metabolism
E70.8	Other disorders of aromatic amino-acid metabolism

E70.81	Aromatic L-amino acid decarboxylase deficiency
E70.89	Other disorders of aromatic amino-acid metabolism
E70.9	Disorder of aromatic amino-acid metabolism, unspecified
E71	Disorders of branched-chain amino-acid metabolism and fatty-acid metabolism
E71.0	Maple-syrup-urine disease
E71.1	Other disorders of branched-chain amino-acid metabolism
E71.11	Branched-chain organic acidurias
E71.110	Isovaleric acidemia
E71.111	3-methylglutaconic aciduria
E71.118	Other branched-chain organic acidurias
E71.12	Disorders of propionate metabolism
E71.120	Methylmalonic acidemia
E71.121	Propionic acidemia
E71.128	Other disorders of propionate metabolism
E71.19	Other disorders of branched-chain amino-acid metabolism
E71.2	Disorder of branched-chain amino-acid metabolism, unspecified
E71.3	Disorders of fatty-acid metabolism
E71.30	Disorder of fatty-acid metabolism, unspecified
E71.31	Disorders of fatty-acid oxidation
E71.310	Long chain/very long chain acyl CoA dehydrogenase deficiency
E71.311	Medium chain acyl CoA dehydrogenase deficiency
E71.312	Short chain acyl CoA dehydrogenase deficiency
E71.313	Glutaric aciduria type II
E71.314	Muscle carnitine palmitoyltransferase deficiency
E71.318	Other disorders of fatty-acid oxidation
E71.32	Disorders of ketone metabolism
E71.39	Other disorders of fatty-acid metabolism
E71.4	Disorders of carnitine metabolism
E71.40	Disorder of carnitine metabolism, unspecified
E71.41	Primary carnitine deficiency
E71.42	Carnitine deficiency due to inborn errors of metabolism
E71.43	Iatrogenic carnitine deficiency
E71.44	Other secondary carnitine deficiency
E71.440	Ruvalcaba-Myhre-Smith syndrome
E71.448	Other secondary carnitine deficiency
E71.5	Peroxisomal disorders
E71.50	Peroxisomal disorder, unspecified
E71.51	Disorders of peroxisome biogenesis
E71.510	Zellweger syndrome
E71.511	Neonatal adrenoleukodystrophy
E71.518	Other disorders of peroxisome biogenesis
E71.52	X-linked adrenoleukodystrophy

E71.520	Childhood cerebral X-linked adrenoleukodystrophy
E71.521	Adolescent X-linked adrenoleukodystrophy
E71.522	Adrenomyeloneuropathy
E71.528	Other X-linked adrenoleukodystrophy
E71.529	X-linked adrenoleukodystrophy, unspecified type
E71.53	Other group 2 peroxisomal disorders
E71.54	Other peroxisomal disorders
E71.540	Rhizomelic chondrodysplasia punctata
E71.541	Zellweger-like syndrome
E71.542	Other group 3 peroxisomal disorders
E71.548	Other peroxisomal disorders
E72	Other disorders of amino-acid metabolism
E72.0	Disorders of amino-acid transport
E72.00	Disorders of amino-acid transport, unspecified
E72.01	Cystinuria
E72.02	Hartnup's disease
E72.03	Lowe's syndrome
E72.04	Cystinosis
E72.09	Other disorders of amino-acid transport
E72.1	Disorders of sulfur-bearing amino-acid metabolism
E72.10	Disorders of sulfur-bearing amino-acid metabolism, unspecified
E72.11	Homocystinuria
E72.12	Methylenetetrahydrofolate reductase deficiency
E72.19	Other disorders of sulfur-bearing amino-acid metabolism
E72.2	Disorders of urea cycle metabolism
E72.20	Disorder of urea cycle metabolism, unspecified
E72.21	Argininemia
E72.22	Arginosuccinic aciduria
E72.23	Citrullinemia
E72.29	Other disorders of urea cycle metabolism
E72.3	Disorders of lysine and hydroxylysine metabolism
E72.4	Disorders of ornithine metabolism
E72.5	Disorders of glycine metabolism
E72.50	Disorder of glycine metabolism, unspecified
E72.51	Non-ketotic hyperglycinemia
E72.52	Trimethylaminuria
E72.53	Primary hyperoxaluria
E72.59	Other disorders of glycine metabolism
E72.8	Other specified disorders of amino-acid metabolism
E72.81	Disorders of gamma aminobutyric acid metabolism
E72.89	Other specified disorders of amino-acid metabolism
E72.9	Disorder of amino-acid metabolism, unspecified
E74	Other disorders of carbohydrate metabolism

E74.0	Glycogen storage disease
E74.00	Glycogen storage disease, unspecified
E74.01	von Gierke disease
E74.02	Pompe disease
E74.03	Cori disease
E74.04	McArdle disease
E74.09	Other glycogen storage disease
E74.1	Disorders of fructose metabolism
E74.10	Disorder of fructose metabolism, unspecified
E74.11	Essential fructosuria
E74.12	Hereditary fructose intolerance
E74.19	Other disorders of fructose metabolism
E74.2	Disorders of galactose metabolism
E74.20	Disorders of galactose metabolism, unspecified
E74.21	Galactosemia
E74.29	Other disorders of galactose metabolism
E74.3	Other disorders of intestinal carbohydrate absorption
E74.31	Sucrase-isomaltase deficiency
E74.39	Other disorders of intestinal carbohydrate absorption
E74.4	Disorders of pyruvate metabolism and gluconeogenesis
E74.8	Other specified disorders of carbohydrate metabolism
E74.81	Disorders of glucose transport, not elsewhere classified
E74.810	Glucose transporter protein type 1 deficiency
E74.818	Other disorders of glucose transport
E74.819	Disorders of glucose transport, unspecified
E74.89	Other specified disorders of carbohydrate metabolism
E74.9	Disorder of carbohydrate metabolism, unspecified
E75	Disorders of sphingolipid metabolism and other lipid storage disorders
E75.0	GM2 gangliosidosis
E75.00	GM2 gangliosidosis, unspecified
E75.01	Sandhoff disease
E75.02	Tay-Sachs disease
E75.09	Other GM2 gangliosidosis
E75.1	Other and unspecified gangliosidosis
E75.1	Other gangliosidosis
E75.10	Unspecified gangliosidosis
E75.11	Mucolipidosis IV
E75.19	Other gangliosidosis
E75.2	Other sphingolipidosis
E75.21	Fabry (-Anderson) disease
E75.22	Gaucher disease
E75.23	Krabbe disease
E75.24	Niemann-Pick disease

E75.240	Niemann-Pick disease type A
E75.241	Niemann-Pick disease type B
E75.242	Niemann-Pick disease type C
E75.243	Niemann-Pick disease type D
E75.244	Niemann-Pick disease type A/B
E75.248	Other Niemann-Pick disease
E75.249	Niemann-Pick disease, unspecified
E75.25	Metachromatic leukodystrophy
E75.26	Sulfatase deficiency
E75.29	Other sphingolipidosis
E75.3	Sphingolipidosis, unspecified
E75.4	Neuronal ceroid lipofuscinosis
E75.5	Other lipid storage disorders
E75.6	Lipid storage disorder, unspecified
E76	Disorders of glycosaminoglycan metabolism
E76.0	Mucopolysaccharidosis, type I
E76.01	Hurler's syndrome
E76.02	Hurler-Scheie syndrome
E76.03	Scheie's syndrome
E76.1	Mucopolysaccharidosis, type II
E76.2	Other mucopolysaccharidoses
E76.21	Morquio mucopolysaccharidoses
E76.210	Morquio A mucopolysaccharidoses
E76.211	Morquio B mucopolysaccharidoses
E76.219	Morquio mucopolysaccharidoses, unspecified
E76.22	anfilippo mucopolysaccharidoses
E76.29	Other mucopolysaccharidoses
E76.3	Mucopolysaccharidosis, unspecified
E76.8	Other disorders of glucosaminoglycan metabolism
E76.9	Glucosaminoglycan metabolism disorder, unspecified
E77	Disorders of glycoprotein metabolism
E77.0	Defects in post-translational modification of lysosomal enzymes
E77.1	Defects in glycoprotein degradation
E77.8	Other disorders of glycoprotein metabolism
E77.9	Disorder of glycoprotein metabolism, unspecified

Intellectual Disability, Coded

ICD-9 Code	Description
317	Mild intellectual disabilities
318	Other specified intellectual disabilities
318.0	Moderate intellectual disabilities

318.1	Severe intellectual disabilities
318.2	Profound intellectual disabilities
319	Unspecified intellectual disabilities

ICD-10 Code

F70	Mild intellectual disabilities
F70.0	Mild mental retardation, With the statement of no, or minimal, impairment of behaviour
F70.1	Mild mental retardation, Significant impairment of behaviour requiring attention or treatment
F70.8	Mild mental retardation, Other impairments of behaviour
F70.9	Mild mental retardation, Without mention of impairment of behaviour
F71	Moderate intellectual disabilities
F71.0	Moderate mental retardation, with the statement of no, or minimal, impairment of behaviour
F71.1	Moderate mental retardation, significant impairment of behaviour requiring attention or treatment
F71.8	Moderate mental retardation, other impairments of behaviour
F71.9	Moderate mental retardation, without mention of impairment of behaviour
F72	Severe intellectual disabilities
F72.0	Severe mental retardation, with the statement of no, or minimal, impairment of behaviour
F72.1	Severe mental retardation, significant impairment of behaviour requiring attention or treatment
F72.8	Severe mental retardation, other impairments of behaviour
F72.9	Severe mental retardation, without mention of impairment of behaviour
F73	Profound intellectual disabilities
F73.0	Profound mental retardation, with the statement of no, or minimal, impairment of behaviour
F73.1	Profound mental retardation, significant impairment of behaviour requiring attention or treatment
F73.8	Profound mental retardation, other impairments of behaviour
F73.9	Profound mental retardation, without mention of impairment of behaviour
F78	Other intellectual disabilities
F78.0	Other mental retardation, with the statement of no, or minimal, impairment of behaviour
F78.1	Other mental retardation, significant impairment of behaviour requiring attention or treatment
F78.8	Other mental retardation, other impairments of behaviour
F78.9	Other mental retardation, without mention of impairment of behaviour
F78.A	Other genetic related intellectual disabilities
F78.A1	SYNGAP1-related intellectual disability

F78.A9	Other genetic related intellectual disability
F79	Unspecified intellectual disabilities
F79.0	Unspecified mental retardation, with the statement of no, or minimal, impairment of behaviour
F79.1	Unspecified mental retardation, significant impairment of behaviour requiring attention or treatment
F79.8	Unspecified mental retardation, other impairments of behaviour
F79.9	Unspecified mental retardation, without mention of impairment of behaviour

Spina Bifida Codes

ICD-9 Code	Description
741	Spina bifida
741.0	Spina bifida with hydrocephalus
741.00	Spina bifida with hydrocephalus, unspecified region
741.01	Spina bifida with hydrocephalus, cervical region
741.02	Spina bifida with hydrocephalus, dorsal (thoracic) region
741.03	Spina bifida with hydrocephalus, lumbar region
741.9	Spina bifida without mention of hydrocephalus
741.90	Spina bifida without mention of hydrocephalus, unspecified region
741.91	Spina bifida without mention of hydrocephalus, cervical region
741.92	Spina bifida without mention of hydrocephalus, dorsal (thoracic) region
741.93	Spina bifida without mention of hydrocephalus, lumbar region
756.17	Spina bifida occulta

ICD-10 Code	
Q05	Spina bifida
Q05.0	Cervical spina bifida with hydrocephalus
Q05.1	Thoracic spina bifida with hydrocephalus
Q05.2	Lumbar spina bifida with hydrocephalus
Q05.3	Sacral spina bifida with hydrocephalus
Q05.4	Unspecified spina bifida with hydrocephalus
Q05.5	Cervical spina bifida without hydrocephalus
Q05.6	Thoracic spina bifida without hydrocephalus
Q05.7	Lumbar spina bifida without hydrocephalus
Q05.8	Sacral spina bifida without hydrocephalus
Q05.9	Spina bifida, unspecified
Q07	Other congenital malformations of nervous system
Q07.0	Arnold-Chiari syndrome
Q07.00	Arnold-Chiari syndrome without spina bifida or hydrocephalus
Q07.01	Arnold-Chiari syndrome with spina bifida

Q07.02	Arnold-Chiari syndrome with hydrocephalus
Q07.03	Arnold-Chiari syndrome with spina bifida and hydrocephalus
Q07.8	Other specified congenital malformations of nervous system
Q07.9	Congenital malformation of nervous system, unspecified

Other conditions presenting Intellectual Disability Codes

ICD-9 Code	Description
758.1	Patau's syndrome
758.2	Edwards' syndrome
759.81	Prader-Willi syndrome

ICD-10 Code

E78.71	Barth syndrome
E78.72	Smith-Lemli-Opitz syndrome
Q87.1	Congenital malformation syndromes predominantly associated with short stature
Q87.11	Prader-Willi syndrome
Q87.19	Other congenital malformation syndromes predominantly associated with short stature
Q87.2	Congenital malformation syndromes predominantly involving limbs
Q87.3	Congenital malformation syndromes involving early overgrowth
Q87.5	Other congenital malformation syndromes with other skeletal changes
Q87.81	Alport syndrome
Q87.89	Other specified congenital malformation syndromes, not elsewhere classified
Q91	Trisomy 18 and Trisomy 13
Q91.0	Trisomy 18, nonmosaicism (meiotic nondisjunction)
Q91.1	Trisomy 18, mosaicism (mitotic nondisjunction)
Q91.2	Trisomy 18, translocation
Q91.3	Trisomy 18, unspecified
Q91.4	Trisomy 13, nonmosaicism (meiotic nondisjunction)
Q91.5	Trisomy 13, mosaicism (mitotic nondisjunction)
Q91.6	Trisomy 13, translocation
Q91.7	Trisomy 13, unspecified
Q93.5	Other deletions of part of a chromosome
Q93.51	Angelman syndrome
Q93.59	Other deletions of part of a chromosome
Q93.82	Williams syndrome

Developmental Delay Disability Codes

ICD-9 Code	Description
315.4	Developmental coordination disorder
315.8	Other specified delays in development

ICD-10 Code

F82	Specific developmental disorder of motor function
F88	Other disorders of psychological development

Learning Disabilities Codes

ICD-9 Code	Description
315	Specific delays in development
315.0	Developmental reading disorder
315.00	Developmental reading disorder, unspecified
315.01	Alexia
315.02	Developmental dyslexia
315.09	Other specific developmental reading disorder
315.1	Mathematics disorder
315.2	Other specific developmental learning difficulties
315.3	Developmental speech or language disorder
315.31	Expressive language disorder
315.32	Mixed receptive-expressive language disorder
315.34	Speech and language developmental delay due to hearing loss
315.35	Childhood onset fluency disorder
315.39	Other developmental speech or language disorder
784.61	Alexia and dyslexia

ICD-10 Code

F80	Specific developmental disorders of speech and language
F80.0	Phonological disorder
F80.1	Expressive language disorder
F80.2	Mixed receptive-expressive language disorder
F80.3	Acquired aphasia with epilepsy [Landau-Kleffner]
F80.4	Speech and language development delay due to hearing loss
F80.8	Other developmental disorders of speech and language
F80.81	Childhood onset fluency disorder
F80.82	Social pragmatic communication disorder
F80.89	Other developmental disorders of speech and language
F80.9	Developmental disorder of speech and language, unspecified

F81	Specific developmental disorders of scholastic skills
F81.0	Specific reading disorder
F81.1	Specific spelling disorder
F81.2	Specific disorder of arithmetical skills
F81.2	Mathematics disorder
F81.3	Mixed disorder of scholastic skills
F81.8	Other developmental disorders of scholastic skills
F81.81	Disorder of written expression
F81.89	Other developmental disorders of scholastic skills
F81.9	Developmental disorder of scholastic skills, unspecified
R48	Dyslexia and other symbolic dysfunctions, not elsewhere classified
R48.0	Dyslexia and alexia