Improving identification of people with a learning disability: guidance for general practice

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Introduction

1. The NHS Long Term Plan[1] commits to improve uptake of the existing annual health check in primary care for people aged over 14 years with a learning disability, so that at least 75% of those eligible have a learning disability health check each year.

2. There is also a need to increase the number of people receiving the annual seasonal flu vaccination, given the level of avoidable mortality associated with respiratory problems.

3. In 2017/18, only 44.6% of patients with a learning disability received a flu vaccination and only 55.1% of patients with a learning disability received an annual learning disability health check.[2]

4. In June 2019, NHS England and NHS Improvement announced a series of measures to improve coverage of annual health checks and flu vaccination for people with a learning disability. One of the commitments was to improve the quality of registers for people with a learning disability[3].

Clinical coding review

5. Most GP practices have developed a register of their patients known to have a learning disability. This has been developed from clinical diagnoses, from information gathered from learning disabilities teams and social services and has formed the basis of registers for people with learning disability developed for the Quality and Outcomes Framework (QOF). These registers have been used to identify patients eligible for an annual seasonal flu vaccination and an annual learning disability health check.

[1] https://www.longtermplan.nhs.uk/wp-content/uploads/2019/01/nhs-long-term-plan-june-2019.pdf
[2] https://digital.nhs.uk/data-and-information/publications/statistical/health-and-care-of-people-with-learning-disabilities/experimental-statistics-2017-to-2018
[3] Implementing the *Long Term Plan* in primary and community Services – Board meetings held in Common – 27th June 2019

6. However, some coded clinical diagnoses do not automatically result in a patient being included on the register. The most common example of this is Down's Syndrome.

7. NHS England and NHS Improvement, working with clinical and coding experts, have undertaken a review of the diagnoses that are associated with a learning disability. We have identified clinical diagnoses that should automatically ensure a patient is included on the learning disability register. The revised list of diagnoses can be found in Appendix 1. These patients will be added automatically to the learning disability register in QOF in early 2020.

8. We have also created a list of diagnoses that may or may not be associated with a learning disability (Appendix 2). Most of these conditions are very rare and it is likely that practices will already be aware of these patients and whether or not they have a learning disability. However, practices are being asked to check that patients with a clinical diagnosis in this group have had an individual assessment to determine if they should be added to the register of people with a learning disability.

9. Whilst we have identified a large number of conditions and codes in Appendices 1 and 2; as most of these conditions are rare it is likely that only small numbers of new patients, will need to be added to a register.

10. This guidance sets out what practices need to do in 19/20 and 20/21 onwards in light of this review to maximise the number of patients receiving a learning disability annual health check and being protected with influenza vaccination. Practices should ensure that they use the recommended codes to record this care in order to receive the relevant fees for completing this work.

Actions for practices

In 2019/20

12. Practices need to use the codes provided to check that all eligible patients are included on the registers for people with a learning disability and are invited for a flu vaccination and a learning disability health check. Not all these codes are currently available in all general practice clinical systems, so practices should not be concerned if they are currently unavailable on their system.

13. There are two steps practices need to take in 2019/20:

Step 1: Review and update the register to ensure that all patients with a clinical diagnosis associated with a learning disability are invited for a flu vaccination and a learning disability health check.

14. Appendix 1 contains a list of clinical diagnoses which are consistently associated with a learning disability. Please ensure all patients with these diagnoses are offered a flu vaccination and an annual learning disability health check, by reviewing the code list in appendix 1 and searching for affected patients. For these patients there is no need to make any changes to the coding of their condition or learning disability. They will be added to the QOF register automatically as your clinical system supplier implements V44 of the QOF business rules which will be complete in early 2020. These codes have already been added to the learning disability health check extraction from October 2019, which means that these patients can be identified by running the learning disability health check reports provided by your clinical system supplier.

15. Practices should note that the code 'academic developmental disorder' has been removed from the register, as this is indicative of a learning difficulty rather than a learning disability. It is unlikely that many, if any, patients will have the 'academic developmental disorder' code in their record and be the only trigger for their inclusion in the learning disability register. Any such patients should have their record reviewed as they may require recoding to ensure that they remain on the register.

16. Practices are reminded that patients with a learning disability may require additional support or 'reasonable adjustments' to enable them and their carers to access health care. These should be assessed and recorded in their medical record.

Step 2: Identify patients with conditions who may also have a learning disability, assess whether the patient should be added to the learning disability register and be offered a flu vaccination and annual learning disability health check.

17. Appendix 2 includes the codes for conditions which may or may not be associated with a learning disability. Not everyone with a recorded diagnosis in this list will have a learning disability, for example, not all patients with cerebral palsy have a learning disability, but these diagnoses are often associated with a learning disability.

18. Practices need to use the code list provided to identify these patients and make an individual assessment of the patient to determine whether they should be added to the register of people with a learning disability. In some cases, this may require a face to face assessment and discussion with the patient and/or their carer. Those added to the register should be offered an annual learning disability health check and flu vaccination.

19. We suggest practices start with the most common conditions with initial assessments being made between now and March 2020. These are highlighted in grey in Appendix 2. NHS England and NHS Improvement are working with NHS Digital and all system suppliers to develop a search tool to help with this.

20. A checklist, known as an 'Inclusion Tool', to help clinicians to determine whether a patient would benefit from being added to the learning disability register is reproduced in Appendix 4. This can also be downloaded from https://www.getcheckedoutleeds.nhs.uk/wp-content/uploads/2019/08/Inclusion-tool-Jan-2019-3.pdf.

21. After assessment and appropriate discussion with the patient and/or carer, if appropriate, the patient can be added to the register by adding the relevant code "On learning disability register (finding)" (SNOMED CT code 416075005).

22. Practices should also review the use of codes listed in Appendix 3. Whilst these codes will ensure that patients are added to the registers the associated descriptions are outdated and no longer acceptable. Practices are advised that the original coding should not be removed but a new code using terminology which is more acceptable to affected individuals should be added under the original date.

23. Please note that the code lists included in appendices 1 and 2 are not exhaustive and clinicians should also ensure that other patients who, in their clinical judgement, have a learning disability are added to the register. It is recommended that this is done by using the following code: 'On learning disability register (finding)' SNOMED CT code 416075005.

From March 2020

24. **Patients with the clinical diagnoses listed in Appendix 1 (that are associated with a learning disability) will be automatically included in the** register of people with a learning disability in the Quality and Outcomes Framework **in early 2020**. This includes conditions such as Down's Syndrome.

25. Practices will need to undertake **on-going identification and review of the patients with a condition which may or may not be associated with a learning disability** (Appendix 2), **to decide if they need to be added to the learning disability register.** NHS England and NHS Improvement are working with NHS Digital and all system suppliers to develop a search tool to help with this.

Further information

26. For further information, please contact: england.pcstrategyandnhscontracts@nhs.net

Appendix 1: List of codes that indicate a learning disability

1. All these codes/diagnoses are indicative of a learning disability. These patients will automatically be included on the registers for people with a learning disability in early 2020.

2. In the interim, we would like practices to be actively searching for these patients and invite them for a flu vaccination and learning disability health check. The shaded codes have the highest prevalence and should be used to identify patients as a priority. Not all codes are currently available in all general practice clinical systems, but they are included in this list for completeness.

SNOMED CT code	**SNOMED CT preferred term**
416075005	**On learning disability register (finding)**

Codes currently included on the register[4]	
984661000000105	Mild learning disability (disorder)
984671000000103	Moderate learning disability (disorder)
508171000000105	Severe learning disability (disorder)
984681000000101	Profound learning disability (disorder)
889211000000104	Specific learning disability (disorder)

Codes that indicate a LD	
110359009	Intellectual disability (disorder)
86765009	Mild intellectual disability (disorder)
61152003	Moderate intellectual disability (disorder)
931001000000105	Significant learning disability (disorder)
40700009	Severe intellectual disability (disorder)
31216003	Profound intellectual disability (disorder)
254264002	Partial trisomy 21 in Down's syndrome
205615000	Trisomy 21- meiotic nondisjunction
205616004	Trisomy 21- mitotic nondisjunction mosaicism

[4] Whilst these codes refer to learning disability they map to learning difficulty codes in SNOMED and we therefore are advising against their continued use. Practices are advised to use the 'on learning disability register (finding)' code when adding patients to the register

719599008	19q13.11 microdeletion syndrome
17122004	4p partial monosomy syndrome
70173007	5p partial monosomy syndrome
718573009	Achalasia microcephaly syndrome
66758006	Acrodysostosis
722281001	Agammaglobulinaemia, microcephaly, craniosynostosis, severe dermatitis syndrome
763797003	Agenesis of corpus callosum and abnormal genitalia syndrome
722282008	Agenesis of corpus callosum, intellectual disability, coloboma, micrognathia syndrome
720979002	Alopecia, contracture, dwarfism, intellectual disability syndrome
734349003	Alpha-thalassaemia intellectual disability syndrome linked to chromosome 16
720982007	Alport syndrome, intellectual disability, midface hypoplasia, elliptocytosis syndrome
76880004	Angelman syndrome
720468000	Aniridia and intellectual disability syndrome
763615003	Aortic arch anomaly, facial dysmorphism, intellectual disability syndrome
720501007	Arachnodactyly with abnormal ossification and intellectual disability syndrome
720517001	Ataxia with deafness and intellectual disability syndrome
718577005	Atkin Flaitz syndrome
5619004	Bardet-Biedl syndrome
717887003	Biemond syndrome type 2
21634003	Borjeson-Forssman-Lehmann syndrome
732961003	Branchial dysplasia, intellectual disability, inguinal hernia syndrome
717945001	BRESEK syndrome
719097002	BSG syndrome
725589005	Bullous dystrophy macular type
715409005	C syndrome
726031001	CAMOS syndrome
720635002	Cerebro-facio-thoracic dysplasia
702357000	Chromosome 2q37 deletion syndrome
719808002	Chromosome Xp11.3 microdeletion syndrome
717763008	Chudley Lowry Hoar syndrome
15182000	Coffin-Lowry syndrome
56604005	Cohen syndrome
21111006	Complete trisomy 13 syndrome
51500006	Complete trisomy 18 syndrome
41040004	Complete trisomy 21 syndrome
722379001	Congenital cataract with hypertrichosis and intellectual disability syndrome
719842006	Congenital hypoplasia of ulna and intellectual disability syndrome
720748007	Cooper Jabs syndrome
732251003	Cortical blindness, intellectual disability, polydactyly syndrome
763665007	Craniodigital syndrome and intellectual disability syndrome
763320005	Craniofaciofrontodigital syndrome
764950001	Cryptorchidism, arachnodactyly, intellectual disability syndrome
59252009	Cutis laxa-corneal clouding-oligophrenia syndrome
40354009	De Lange syndrome

721087008	Deafness and intellectual disability Martin Probst type syndrome
766871009	Diencephalic mesencephalic junction dysplasia
719450007	Disorder of sex development with intellectual disability syndrome
719800009	DOORS syndrome
2593002	Dubowitz's syndrome
721208007	Ectodermal dysplasia with blindness syndrome
734017008	Ectodermal dysplasia, intellectual disability, central nervous system malformation syndrome
733049004	Encephalopathy, intracerebral calcification, retinal degeneration syndrome
733032006	Epilepsy telangiectasia syndrome
733031004	Epilepsy, microcephaly, skeletal dysplasia syndrome
766870005	Epiphyseal dysplasia, hearing loss, dysmorphism syndrome
733417008	Facial dysmorphism, macrocephaly, myopia, Dandy-Walker malformation syndrome
723333000	Faciocardiorenal syndrome
723336008	Fallot complex with intellectual disability and growth delay syndrome
720954000	Filippi syndrome
765089003	Focal epilepsy, intellectual disability, cerebro-cerebellar malformation syndrome
720957007	Fountain syndrome
716709002	FRAXE intellectual disability syndrome
718848000	Fried syndrome
716024001	GMS syndrome
717822006	Goldberg Shprintzen megacolon syndrome
716096005	Goldblatt Wallis syndrome
763186006	Grubben, De Cock, Borghgraef syndrome
721007005	Hair defect with photosensitivity and intellectual disability syndrome
721008000	Hall Riggs syndrome
716089008	Harrod syndrome
721224008	Holmes Gang syndrome
33982008	Hyperphosphatasaemia with intellectual disability
763722004	Hypotonia, speech impairment, severe cognitive delay syndrome
723365002	Hypotrichosis and intellectual disability syndrome Lopes type
763404001	Ichthyosis, alopecia, eclabion, ectropion, intellectual disability syndrome
733097003	Ichthyosis, intellectual disability, dwarfism, renal impairment syndrome
724228005	Infantile choroidocerebral calcification syndrome
1094031000000100	Intellectual development disorder with impairment of behaviour
1094021000000102	Intellectual development disorder with minimal impairment of behaviour
1094011000000108	Intellectual development disorder with significant impairment of behaviour
1094001000000106	Intellectual development disorder without significant impairment of behaviour
110359009	Intellectual disability
764861005	Intellectual disability Birk-Barel type
725906006	Intellectual disability Buenos Aires type
763626009	Intellectual disability due to nutritional deficiency
722003007	Intellectual disability with cataract and kyphosis syndrome
763745005	Intellectual disability Wolff type
763741001	Intellectual disability, alacrima, achalasia syndrome
763744009	Intellectual disability, brachydactyly, Pierre Robin syndrome

726709001	Intellectual disability, cataract, calcified pinna, myopathy syndrome
412787009	Intellectual disability, congenital heart disease, blepharophimosis, blepharoptosis and hypoplastic teeth
722454003	Intellectual disability, craniofacial dysmorphism, hypogonadism, diabetes mellitus syndrome
722456001	Intellectual disability, developmental delay, contracture syndrome
721146009	Intellectual disability, epilepsy, bulbous nose syndrome
722455002	Intellectual disability, hypoplastic corpus callosum, preauricular tag syndrome
764959000	Intellectual disability, myopathy, short stature, endocrine defect syndrome
763350002	Intellectual disability, obesity, brain malformation, facial dysmorphism syndrome
763742008	Intellectual disability, polydactyly, uncombable hair syndrome
763743003	Intellectual disability, spasticity, ectrodactyly syndrome
723332005	Isodicentric chromosome 15 syndrome
721875000	Juberg Marsidi syndrome
722031003	Kapur Toriello syndrome
715989002	Karandikar Maria Kamble syndrome
716112005	Kawashima Tsuji syndrome
724207001	Kleefstra syndrome
109478007	Kohlschutter's syndrome
716996008	L1 syndrome
232059000	Laurence-Moon syndrome
721973006	Lipodystrophy, intellectual disability, deafness syndrome
763773007	Macrocephaly and developmental delay syndrome
722033000	Macrocephaly, short stature, paraplegia syndrome
763795006	Malan overgrowth syndrome
722459008	Male hypergonadotropic hypogonadism, intellectual disability, skeletal anomaly syndrome
722380003	Martsolf syndrome
722035007	MEDNIK syndrome
733522005	Megalocornea with intellectual disability syndrome
722037004	MEHMO syndrome
723403008	Microbrachycephaly, ptosis, cleft lip syndrome
719396000	Microcephalus and intellectual disability with phalangeal and neurological anomaly syndrome
719380003	Microcephalus cardiomyopathy syndrome
719378009	Microcephalus with brachydactyly and kyphoscoliosis syndrome
733090001	Microcephalus, digital anomaly, intellectual disability syndrome
733472005	Microcephalus, glomerulonephritis, marfanoid habitus syndrome
723304001	Microcephaly, seizure, intellectual disability, heart disease syndrome
1093991000000101	Mild intellectual development disorder with impairment of behaviour
1089851000000103	Mild intellectual development disorder with minimal impairment of behaviour
1089841000000101	Mild intellectual development disorder with significant impairment of behaviour
1089831000000105	Mild intellectual development disorder without significant impairment of behaviour
86765009	Mild intellectual disability
1089821000000108	Moderate intellectual development disorder with impairment of behaviour
1089811000000102	Moderate intellectual development disorder with minimal impairment of behaviour
1089791000000103	Moderate intellectual development disorder with significant impairment of behaviour

1089781000000100	Moderate intellectual development disorder without significant impairment of behaviour
61152003	Moderate intellectual disability
724137002	MOMO syndrome
715628009	MORM syndrome
703535000	Mowat-Wilson syndrome
65327002	Mucopolysaccharidosis type I-H
725908007	Neurofaciodigitorenal syndrome
703526007	Neuronal ceroid lipofuscinosis 8
766753005	Nijmegen breakage syndrome-like disorder
723441001	Non-progressive cerebellar ataxia with intellectual disability
205824006	Noonan's syndrome
699297004	Ohdo syndrome, Maat-Kievit-Brunner type
699298009	Ohdo syndrome, Say-Barber-Biesecker-Young-Simpson variant
722065002	Okamoto syndrome
721017000	Oliver syndrome
718681002	Oro-facial digital syndrome type 11
763837007	Oro-facial digital syndrome type 14
718680001	Oro-facial digital syndrome type 9
722110003	Osteogenesis imperfecta, retinopathy, seizures, intellectual disability syndrome
732954002	Osteopenia, intellectual disability, sparse hair syndrome
722111004	Osteopenia, myopia, hearing loss, intellectual disability, facial dysmorphism syndrome
763861000	Pachygyria, intellectual disability, epilepsy syndrome
719020006	Pallister W syndrome
254268004	Partial trisomy 13 in Patau's syndrome
702412005	Partington syndrome
716191002	Perniola Krajewska Carnevale syndrome
719139003	Pettigrew syndrome
702344008	Pitt-Hopkins syndrome
768677000	PPP2R5D-related intellectual disability
89392001	Prader-Willi syndrome
733088002	Preaxial polydactyly, colobomata, intellectual disability syndrome
719140001	Prieto Badia Mulas syndrome
1089731000000104	Profound intellectual development disorder with impairment of behaviour
1089721000000101	Profound intellectual development disorder with minimal impairment of behaviour
1089711000000107	Profound intellectual development disorder with significant impairment of behaviour
1089701000000105	Profound intellectual development disorder without impairment of behaviour
31216003	Profound intellectual disability
733086003	Pseudoprogeria syndrome
719162001	Radioulnar synostosis with microcephaly and scoliosis syndrome
723504000	Ramos Arroyo syndrome
723501008	Renier Gabreels Jasper syndrome
699669001	Renpenning syndrome
724001005	Retinitis pigmentosa, intellectual disability, deafness, hypogenitalism syndrome
68618008	Rett's disorder

722002002	Scholte syndrome
721207002	Seizure, sensorineural deafness, ataxia, intellectual disability, electrolyte imbalance syndrome
723994004	Seizures and intellectual disability due to hydroxylysinuria
1089771000000102	Severe intellectual development disorder with impairment of behaviour
1089761000000109	Severe intellectual development disorder with minimal impairment of behaviour
1089751000000106	Severe intellectual development disorder with significant impairment of behaviour
1089741000000108	Severe intellectual development disorder without significant impairment of behaviour
40700009	Severe intellectual disability
723676007	Severe intellectual disability, epilepsy, anal anomaly, distal phalangeal hypoplasia syndrome
722213009	Severe X-linked intellectual disability Gustavson type
721073008	Short stature with webbed neck and congenital heart disease syndrome
726672000	Short stature, unique facies, enamel hypoplasia, progressive joint stiffness, high-pitched voice syndrome
719069008	Shprintzen Goldberg craniosynostosis syndrome
715428003	Skeletal dysplasia with epilepsy and short stature syndrome
722478008	Skeletal dysplasia with intellectual disability syndrome
719212004	Smith Fineman Myers syndrome
401315004	Smith-Magenis syndrome
702416008	Snyder-Robinson syndrome
733455003	Spastic paraplegia, glaucoma, intellectual disability syndrome
722209002	Spastic paraplegia, intellectual disability, palmoplantar hyperkeratosis syndrome
723621000	Spastic tetraplegia, retinitis pigmentosa, intellectual disability syndrome
719202006	Spondyloepiphyseal dysplasia tarda Kohn type
718766002	Spondyloepiphyseal dysplasia, craniosynostosis, cleft palate, cataract and intellectual disability syndrome
733072002	Stimmler syndrome
718900002	Syndromic X-linked intellectual disability type 11
719160009	Syndromic X-linked intellectual disability type 7
725140007	Temple Baraitser syndrome
719947004	Temtamy syndrome
733117001	Thumb stiffness, brachydactyly, intellectual disability syndrome
717157006	Trisomy 10p
716334004	Urban Rogers Meyer syndrome
733110004	Van den Bosch syndrome
726670008	Weaver Williams syndrome
763618001	Wiedemann Steiner syndrome
719834005	Wilson Turner syndrome
718226002	Wolf Hirschhorn syndrome
717223008	X-linked epilepsy with learning disability and behaviour disorder syndrome
719018008	X-linked intellectual disability Abidi type
719155005	X-linked intellectual disability and epilepsy with progressive joint contracture and facial dysmorphism syndrome
719157002	X-linked intellectual disability and hypotonia with facial dysmorphism and aggressive behaviour syndrome
719017003	X-linked intellectual disability Armfield type
725912001	X-linked intellectual disability Brooks type

719811001	X-linked intellectual disability Cabezas type
719016007	X-linked intellectual disability Cantagrel type
719013004	X-linked intellectual disability Cilliers type
726727003	X-linked intellectual disability Hedera type
719012009	X-linked intellectual disability Miles Carpenter type
726732002	X-linked intellectual disability Nascimento type
719011002	X-linked intellectual disability Pai type
719010001	X-linked intellectual disability Schimke type
718897009	X-linked intellectual disability Seemanova type
718905007	X-linked intellectual disability Shrimpton type
718908009	X-linked intellectual disability Siderius type
718909001	X-linked intellectual disability Stevenson type
718910006	X-linked intellectual disability Stocco Dos Santos type
718911005	X-linked intellectual disability Stoll type
718912003	X-linked intellectual disability Turner type
718914002	X-linked intellectual disability Van Esch type
719009006	X-linked intellectual disability Wilson type
719136005	X-linked intellectual disability with cerebellar hypoplasia syndrome
719137001	X-linked intellectual disability with corpus callosum agenesis and spastic quadriparesis syndrome
422437002	X-linked intellectual disability with marfanoid habitus
719812008	X-linked intellectual disability with plagiocephaly syndrome
719810000	X-linked intellectual disability with seizure and psoriasis syndrome
718846001	X-linked intellectual disability Zorick type
765471005	X-linked intellectual disability, hypogonadism, ichthyosis, obesity, short stature syndrome
732246009	X-linked intellectual disability, limb spasticity, retinal dystrophy, diabetes insipidus syndrome
725163002	X-linked spasticity, intellectual disability, epilepsy syndrome

Appendix 2: List of codes that may indicate a learning disability

1. A record of one or more of the following codes will not automatically result in a patient being added to the register of patients with a learning disability. Patients would need to be individually assessed to determine if they have a learning disability and added to the register if appropriate. It is likely that these patients will already be familiar to clinicians in the practice. We advise that when adding a patient to the learning disability register that practices use the following code: "On learning disability register (finding)" (SNOMED CT code 416075005).

2. The shaded codes at the top of the list have the highest prevalence and should be used as a starting point to identify patients to be assessed for inclusion on the register. Not all codes are currently available in all general practice clinical systems, but they are included in this list for completeness.

SNOMED CT code	SNOMED CT preferred term
722287002	Autism and facial port-wine stain syndrome (disorder)
733623005	Autism spectrum disorder, epilepsy, arthrogryposis syndrome (disorder)
408856003	Autistic disorder
43614003	Autistic disorder of childhood onset
373618009	Autistic spectrum disorder with isolated skills
128188000	Cerebral palsy
307756005	Cerebral palsy, not congenital or infantile, acute
76880004	Angelman syndrome
23560001	Asperger's disorder
21111006	Complete trisomy 13 syndrome
51500006	Complete trisomy 18 syndrome
41040004	Complete trisomy 21 syndrome
205788004	Fetal alcohol syndrome
613003	Fragile X syndrome
190745006	Galactosaemia
254268004	Partial trisomy 13 in Patau's syndrome
254266000	Partial trisomy 18 in Edward's syndrome
254264002	Partial trisomy 21 in Down's syndrome
254281006	Turner's phenotype - ring chromosome karyotype
719046005	12q14 microdeletion syndrome
719583002	17q11.2 microduplication syndrome

719600006	1p21.3 microdeletion syndrome (disorder)
767263007	22q11.2 deletion syndrome
17122004	4p partial monosomy syndrome
70173007	5p partial monosomy syndrome
205258009	Acrocephalosyndactyly type I
442314000	Active but odd autism
191692007	Active disintegrative psychoses
191689008	Active infantile autism (disorder)
766824003	Activity dependent neuroprotector homeobox related multiple congenital anomalies, intellectual disability, autism spectrum disorder (disorder)
230312006	Aicardi Goutieres syndrome
80651009	Aicardi's syndrome
720981000	Alopecia and intellectual disability with hypergonadotropic hypogonadism syndrome
733116005	Aniridia, renal agenesis, psychomotor retardation syndrome
22155002	Anterior chamber cleavage syndrome
278512001	Ataxic cerebral palsy
75019001	Athetoid cerebral palsy
231536004	Atypical autism
718393002	Atypical Rett syndrome
234138005	Bannayan syndrome
205828009	Biemond's syndrome
702319000	Bilateral cerebral palsy (disorder)
904531000000100	Bilateral spastic cerebral palsy (disorder)
928241000000105	Birth to 25 education, health and care plan-based care stopped (situation)
928221000000103	Birth to 25 education, health and care plan-based care started (situation)
717913006	Blepharonasofacial malformation syndrome
77287004	Borderline intellectual disability
765761009	Brachydactyly, mesomelia, intellectual disability, heart defect syndrome
720573009	Brachymorphism with onychodysplasia and dysphalangism syndrome
703389002	CASK related intellectual disability
726621009	Caudal appendage deafness syndrome
726669007	Central nervous system calcification, deafness, tubular acidosis, anaemia syndrome
720855003	Cerebrooculonasal syndrome
763136000	Charcot-Marie-Tooth disease, deafness, intellectual disability syndrome
71961003	Childhood disintegrative disorder
702316007	Choreic cerebral palsy (disorder)
885831000000109	Choreoathetoid cerebral palsy (disorder)
7573000	Classical phenylketonuria
21086008	Cockayne syndrome
10007009	Coffin-Siris syndrome
764455002	Cognitive impairment, coarse facies, heart defects, obesity, pulmonary involvement, short stature, skeletal dysplasia syndrome
720639008	Coloboma, congenital heart disease, ichthyosiform dermatosis, intellectual disability ear anomaly syndrome
88425004	Congenital anomaly of nervous system
719102004	Congenital cataract with ataxia and deafness syndrome

722378009	Congenital cataract with deafness and hypogonadism syndrome
278503003	Congenital hypothyroidism with diffuse goiter (disorder)
237565000	Congenital iodine deficiency syndrome - mixed type (disorder)
237566004	Congenital iodine deficiency syndrome - neurological type
217710005	Congenital iodine deficiency syndrome (disorder)
192949002	Congenital paraplegia
275468009	Congenital quadriplegia
32219008	Craniorachischisis
720401009	Cystic fibrosis with gastritis and megaloblastic anaemia syndrome
720825005	Cystic leukoencephalopathy without megalencephaly
190948002	Defect in post-translational modification of lysosomal enzymes
205630009	Deletion of long arm of chromosome 13
270889005	Deletion of long arm of chromosome 18
254274004	Deletion of part of autosome
270890001	Deletion of short arm of chromosome 18
721089006	Dentinogenesis imperfecta, short stature, hearing loss, intellectual disability syndrome
1855002	Developmental academic disorder
58193001	Diplegic cerebral palsy
238045003	Disorder of glycoprotein metabolism
238043005	Disorder of glycosaminoglycan metabolism
238006008	Disorder of purine and pyrimidine metabolism
715438008	Distal partial deletion of long arm of chromosome 11
16171003	Double athetosis
230780007	Dyskinetic cerebral palsy
733050004	Dysmorphism, short stature, deafness, disorder of sex development syndrome
702315006	Dystonic cerebral palsy (disorder)
230781006	Dystonic/rigid cerebral palsy (disorder)
716107009	Early onset parkinsonism and intellectual disability syndrome
276854003	Educationally subnormal
440092001	Endemic congenital iodine deficiency syndrome of myxedematous type (disorder)
75065003	Endemic cretinism
75065003	Endemic cretinism (disorder)
716706009	Female restricted epilepsy with intellectual disability syndrome
720955004	Fine Lubinsky syndrome
702450004	FOXG1 syndrome
205720009	Fragile X chromosome
254150007	Francois syndrome (disorder)
720958002	Frank-Ter Haar syndrome
50967008	Gangliosidosis
224958001	Global developmental delay
29633007	Glycogen storage disease
1035691000000100	Has birth to 25 education, health and care plan (finding)
43486001	Hemiplegic cerebral palsy
765434008	HIVEP2-related intellectual disability
30915001	Holoprosencephaly sequence

5667009	Hunter's syndrome, mild form (disorder)
73146005	Hunter's syndrome, severe form (disorder)
721841001	Hypogonadism with mitral valve prolapse and intellectual disability syndrome
192958009	Hypotonic cerebral palsy
408857007	Infantile autism
408858002	Infantile psychosis
2438005	Iniencephaly
75979009	Johanson blizzard syndrome
724178000	Laryngeal abductor paralysis with intellectual disability syndrome
10406007	Lesch-Nyhan syndrome
10741005	Lipid storage disease
14210003	Lipofuscinosis
721974000	Lowry MacLean syndrome
205661000	Major partial trisomy
733062000	Marfanoid habitus with autosomal recessive intellectual disability syndrome
69463008	Maroteaux-Lamy syndrome
26015003	Maroteaux-Lamy syndrome, intermediate form (disorder)
67854007	Maroteaux-Lamy syndrome, mild form (disorder)
58263000	Maroteaux-Lamy syndrome, severe form (disorder)
715441004	McDonough syndrome
59178007	Menkes kinky-hair syndrome
47437004	Mental handicap (finding)
170695009	Mental handicap problem
733419006	Metaphyseal dysostosis, intellectual disability, conductive deafness syndrome
702816000	Methyl-cytosine phosphate guanine binding protein-2 duplication syndrome
765758008	Microcephalic primordial dwarfism Montreal type
1829003	Microcephalus (disorder)
205662007	Minor partial trisomy
702318008	Mixed cerebral palsy (disorder)
56409008	Monoplegic cerebral palsy (disorder)
378007	Morquio syndrome
11380006	Mucopolysaccharidosis
41572006	Mucopolysaccharidosis III-A (disorder)
59990008	Mucopolysaccharidosis III-B (disorder)
75238000	Mucopolysaccharidosis III-C (disorder)
15892005	Mucopolysaccharidosis III-D (disorder)
7259005	Mucopolysaccharidosis IV-A (disorder)
26745009	Mucopolysaccharidosis type I-H/S (disorder)
73123008	Mucopolysaccharidosis type I-S (disorder)
238044004	Mucopolysaccharidosis type IVB (disorder)
43916004	Mucopolysaccharidosis type VII (disorder)
75610003	Mucopolysaccharidosis, MPS-I
70737009	Mucopolysaccharidosis, MPS-II
64491003	Myxedematous form of cretinism (disorder)
111308000	Neurologic form of cretinism (disorder)
702314005	Non-spastic cerebral palsy (disorder)

722056009	Oculocerebrofacial syndrome Kaufman type
722105002	Oro-facial digital syndrome type 5
722106001	Oro-facial digital syndrome type 8
371079004	Paraplegic cerebral palsy (disorder)
205660004	Partial trisomy syndromes
702321005	Pentaplegic cerebral palsy (disorder)
35919005	Pervasive developmental disorder
39951000119105	Pervasive developmental disorder of residual state
449817000	Peters plus syndrome
723454008	Phosphoribosylpyrophosphate synthetase superactivity
72991005	Polyploidy syndrome
702356009	PPM-X syndrome
254261005	Pseudotrisomy 18
724039002	Psychomotor retardation due to S-adenosylhomocysteine hydrolase deficiency
768473009	PURA syndrome
48721008	Quadriplegic cerebral palsy (disorder)
721883006	Radioulnar synostosis with developmental delay and hypotonia syndrome
204032005	Reduction deformities of brain
708037001	Residual Asperger's disorder
191693002	Residual disintegrative psychoses
191690004	Residual infantile autism (disorder)
88393000	Sanfilippo syndrome
432091002	Savant syndrome
734173003	SCARF syndrome
765170001	SCN8A-related epilepsy with encephalopathy
230773005	Spastic cerebral palsy
813921000000104	Spastic hemiplegic cerebral palsy (disorder)
732958004	Spastic paraplegia with precocious puberty syndrome
84781002	Sporadic cretinism (disorder)
719161008	Syndromic X-linked intellectual disability due to JARID1C mutation
722477003	Toriello Carey syndrome
702320006	Triplegic cerebral palsy (disorder)
254272000	Triploidy and polyploidy
66651005	Triploidy syndrome
205651007	Trisomy 10
205652000	Trisomy 11
205653005	Trisomy 12
205620000	Trisomy 13 - mitotic nondisjunction mosaicism
205619006	Trisomy 13, meiotic nondisjunction
205623003	Trisomy 18 - meiotic nondisjunction
205624009	Trisomy 18 - mitotic nondisjunction mosaicism
205655003	Trisomy 22
205647005	Trisomy 6
205648000	Trisomy 7
205649008	Trisomy 8
205650008	Trisomy 9

270521004	Trisomy and partial trisomy of autosome
719909009	Trisomy Xq28 syndrome
7199000	Tuberous sclerosis syndrome
719042007	Uveal coloboma with cleft lip and palate and intellectual disability syndrome
63119004	Weaver syndrome
205636003	Whole chromosome monosomy - meiotic nondisjunction
270520003	Whole chromosome monosomy - mitotic nondisjunction mosaicism
254269007	Whole chromosome trisomy meiotic nondisjunction
205646001	Whole chromosome trisomy syndrome
205657006	Whole chromosome trisomy, mosaicism
63247009	Williams syndrome
719825000	X-linked intellectual disability, macrocephaly, macroorchidism syndrome
88469006	Zellweger syndrome