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The proposed new list of hereditary choreas

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| **New designation** | **Less common movement phenotype** | **Clinical clues** | **Inheritance** | **Locus symbol** |
| CHOR-*HTT[1]* |  | Huntington’s disease (HD): Chorea and dementia, young onset may have predominant parkinsonism (Westphal variant)  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1305/  OMIM 143100 | AD | None |
| CHOR-*PRNP[2]* |  | HD-like phenotype, seizures, dementia (variable)  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1305/  OMIM 603218 | AD | HDL1 |
| CHOR-*JPH3[3]* |  | HD-like phenotype  To date only found in patients of African descent  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1529/  OMIM 606438 | AD | HDL2 |
| CHOR-*NKX2*-1[4] |  | Phenotypes  1. Brain–lung–  thyroid syndrome (50%): infantile onset global developmental delay, childhood onset chorea-athetosis, hypothyroidism and pulmonary dysfunction  2. Brain and thyroid disease (30%): infantile onset global developmental delay childhood onset chorea-athetosis, hypothyroidism  3. Isolated benign hereditary chorea (13%)[5]  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK185066/  OMIM [600635](http://omim.org/entry/600635) | AD | None |
| CHOR-*VPS13A[6], [7]* |  | Neuroacanthocytosis [8]:  Chorea, occasionally parkinsonism, dystonia (feeding dystonia)  Additional clinical features: Orofacial dyskinesias, seizures, dementia, myopathy, psychiatric symptoms, acanthocytosis (variable), elevated CK, reduced chorein[9]GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1387/  OMIM 605978 | AR | none |
| CHOR-*XK[10]* |  | McLeod syndrome [11]: Chorea  Additional clinical features: Behavioral and psychiatric symptoms, seizures, myopathy, cardiomyopathy, cardiac arrhythmias, neuropathy, acanthocytosis, elevated CK and liver enzymes, reduced or absent Kx and Kell blood group antigens  GeneReviews  <http://www.ncbi.nlm.nih.gov/books/NBK1354/>  OMIM: 314850 | X-linked | none |
| NBIA/CHOREA-*FTL[12]* | Dystonia, parkinsonism | Neuroferritinopathy [13]:  Dystonia, chorea, parkinsonism  Iron accumulation: GP, caudate, putamen, SN, red nucleus; cystic BG changes – pallidal necrosis  Additional clinical features: Oromandibular dyskinesia, dysphagia, cognitive impairment, behavioral symptoms, low serum ferritin  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1141/  OMIM 606159 | AD | NBIA3 |
| CHOR-PDE10A[14], [15] |  | AR: Childhood onset axial hypotonia, chorea, ballism, variable orofacial dyskinesia. Variable cognition and normal MRI brain.  OMIM 616921  AD: heterozygous cases: slowly progressive chorea with normal cognition. MRI brain bilateral T2 striatal hyperintensity  OMIM 616922 | AR/AD de novo mutation | IOLOD  ADSD2 |
| **Combined phenotypes: where chorea coexists with (an)other movement disorder(s) as a prominent and consistent feature** | | | | |
| CHOR/DYT-*ADCY5[16]* |  | Facial dyskinesias, occasional myoclonus. May have paroxysmal worsening  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK263441/  OMIM [600293](http://omim.org/entry/600293) | AD | None |
| DYT/CHOR-*HPRT[17]* |  | Lesch-Nyhan syndrome [18]: Dystonia, chorea, occasionally ballism  Additional clinical features: Hyperuricemia, crystalluria, developmental delay/intellectual disability, eye movement abnormalities, spasticity, compulsive self-injurious behavior, gouty arthritis, nephrolithiasis, renal failure behavior  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1149/  OMIM 300322 | X-linked | none |
| DYT/CHOR-*ACAT1[19]* |  | Mitochondrial acetoacetyl-CoA thiolase deficiency metabolic decompensation and basal ganglia injury during acute stress resulting in dystonia and chorea[20]  GeneReviews  n/a  OMIM 607809 | AR | none |
| DYT/CHOR-*GCDH[21]* |  | Glutaric aciduria type I [22], [23]: Dystonia, chorea (usually following acute metabolic crises), parkinsonism (later)  Additional clinical features: Acute metabolic crises with basal ganglia injury (predominantly putamen and caudate nucleus), severe truncal hypotonia, macrocephaly, orofacial dyskinesias, spasticity, cognitive impairment (variable), enlarged subdural space, subdural hygroma/hemorrhages, headaches, seizures[24]  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1134/  OMIM 231670 | AR | None |
| DYT/CHOR-*MUT[25]* |  | Methylmalonic aciduria [26]: Dystonia, chorea, occasionally ataxia  Additional clinical features: Neonatal-onset vomiting, seizures, lethargy and hypotonia, ketoacidosis, hyperammonemia, developmental delay, spasticity, pancreatitis, nephritis, growth failure, acute metabolic crises with confusion / encephalopathy, basal ganglia injury (predominantly globus pallidus)  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1134/  OMIM 251000 | AR | None |
| DYT/CHOR-*PCCA/PCCB[27]* |  | Propionic aciduria [26]: Dystonia, occasionally chorea  Additional clinical features: Neonatal-onset vomiting, seizures, lethargy and hypotonia, ketoacidosis, hyperammonemia, developmental delay, spasticity, cardi omyopathy, acute metabolic crises with confusion / encephalopathy, basal ganglia injury (predominantly putamen and caudate nucleus)  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1134/  OMIM 606054 | AR | None |
| **Disorders that usually present with other phenotypes but can have predominant chorea** | | | | |
| *C9orf72* [28] | Chorea[29] | Chorea, dystonia myoclonus, parkinsonism, cognitive decline in small percentage.  More common phenotype: frontotemporal dementia, amyotrophic lateral sclerosis.  Gene Reviews  http://www.ncbi.nlm.nih.gov/books/NBK268647/ | AD | None |
| SCA-*TBP[30]* | Chorea[31] | Marked non-ataxia features, can present with predominant chorea. May be HD-like  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1438/  OMIM 607136 | AD | SCA17, HDL4 |
| SCA-*ATN1[32]* | Chorea[33] | Dentatorubropallidoluysian atrophy (DRPLA): Myoclonus, chorea, parkinsonism, dementia, supranuclear gaze palsy, seizures (particularly in young patients)  GeneReviews  http://www.ncbi.nlm.nih.gov/books/NBK1491/  OMIM: 607462 | AD | None |
| NBIA/DYT/PARK-*CP[34]* | Chorea | Aceruloplasminemia [35]: Dystonia, ataxia, chorea, parkinsonism, tremors  Iron accumulation: More homogeneous involvement of primarily, caudate, putamen, thalamus, dentate  Additional clinical features: Cognitive impairment, psychiatric symptoms, diabetes mellitus, retinal degeneration, anemia, liver iron storage  GeneReviews http://www.ncbi.nlm.nih.gov/books/NBK1493/  OMIM 604290 | AR |  |
| NBIA/DYT- *DCAF17[36]* | chorea[37] | Woodhouse-Sakati syndrome [38]:  Iron accumulation: GP, SN, other BG (variable)  Additional clinical features: Dysarthria, deafness, seizures, cognitive impairment, hypogonadism, alopecia, diabetes mellitus, thyroid dysfunction, acanthosis nigrans, keratoconus, camptodactyly  GeneReviews: n/a  OMIM 241080 | AR | None |
| NBIA/DYT-*PANK2[39]* | parkinsonism,[40] chorea | Pantothenate kinase-associated neurodegeneration (PKAN):  Iron accumulation: GP – eye of the tiger sign  Additional clinical features: Spasticity, dysarthria, cognitive decline, gaze palsy, psychiatric symptoms, pigmentary retinopathy  GeneReviews http://www.ncbi.nlm.nih.gov/books/NBK121988/  OMIM 234200 | AR | NBIA1 |

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