Supplementary Table 1. A summary of recent reviews and case-reports used to map relevant neuropsychiatric symptoms and disorders in Table 1b.

Nr.	Reference	Article type	Case-subjects	Nationality	Relevant neuropsychiatric findings
Phenylketonuria (PKU)					
1	(Mainka et al., 2021)	Original	26, 35-57y	German, Slovak	The authors evaluated neuropsychiatric comorbidities in late-treated PKU patients. In 26 patients, the authors found: intellectual disability (100%), speech impairment (50%), obsessive-compulsive behavior (35%), self-injurious behavior (31%), anxiety (27%), aggressive behavior (27%), depression, (19%), tics (19%), and features observed in ASD (19%).
2	(Aitkenhead et al., 2021)	Original	198, 32.38±9.04y (cognitive test group), 33.93±8.90y (questionnaire group)	British	The authors used cognitive tests (n=154) and psychosocial questionnaires (n=149). In 153 patients, the results included: inattention (7.69%) and impaired working memory (6.67%). In 152 patients; learning difficulties (11.61%), impaired executive function (9.39%) and processing speed (23.49%). Finally, in 150 patients, the authors reported impaired language (3.40%) and executive functioning (S) (12.39%). PKU patients' occupational, emotional and social functioning, and quality of life PKU patients did not differ from controls.
3	(Yamada et al., 2021)	Original	85 > 19y	Japanese	The authors compared long-term outcomes in patients diagnosed with newborn screening (NBS) (n=68) to patients diagnosed before its initiation in Japan (n=17). In NBS-diagnosed patients, 14.7% (n=10/68) presented with intellectual disability. This was significantly lower than patients diagnosed pre-NBS, where 70.6% (n=12/17) presented with intellectual disability.
4	(Ashe et al., 2019)	Review	N/A	Multiple	The authors reviewed neuropsychiatric/cognitive function and the effect of treatment. Based on previously published case reports, patients present with ADHD, hyperactivity, inattention, psychotic symptoms or disorders (schizophrenia, bipolar disorder), autistic behavior, aggression, anxiety, depressed mood, impaired social skills, intellectual disability, educational difficulties, sleep disturbances, and impaired executive functioning.
5	(Bilder et al., 2017)	Original	3714, 20-80(+)y	American	The authors studied the prevalence of neuropsychiatric comorbidities in adults with PKU and found a higher prevalence than diabetes mellitus (DM) patients and the general population. They report multiple involved symptoms and disorders in a large cohort (n=3714), such as depression (19.5%), anxiety (15.6%), sleep disturbances (14.4%), intellectual disabilities (4.8%), bipolar disorder (4.1%), psychosis/schizophrenia (3.7%), ADD/ADHD (2.1%), behavior/conduct disorder (2.0%), personality disorder (1.5%), OCD (1.1%),

					ASD (0.7%), and Tourette's syndrome (0.2%). Patients aged 20-39 years (n=2242) scored low compared to the remaining patients, indicating that early, continuous treatment gives better neuropsychiatric outcomes.
6	(Antshel and Waisbren, 2003)	Original	79, 7-16y	N/A (Caucasian)	The authors compared the symptom dimensions of ADHD (and ADHD-like symptoms) in PKU and MPKU offspring and the developmental timing of Phe exposure. 79 children were divided into control (n=18), PKU patients (n=46), and MPKU offspring (n=15). ADHD diagnosis was established in 13% PKU patients and 40% MPKU offspring. Patients also had a higher prevalence of ADHD symptoms (inattention, hyperactivity, impulsivity) than the control group. Notably, MPKU patients scored highest.
Tyrosinemia Type I (TT1), Type II (TT2), and Type III (TT3)					
7	*(Barone et al., 2020)*	Original	8, 6-18y	Norwegian	The authors aimed to measure core ADHD symptoms in HT1 patients and investigate their relation to plasma tyrosine levels. They report significant correlations between (1) inattention symptoms (ADHD RS-IV) and mean tyrosine levels, (2) working memory index on BRIEF and mean tyrosine levels, and (3) t-scores for inattention and working memory.
8	*(van Vliet et al., 2019)*	Original	31, 8-24y	British, Belgian, Dutch	The authors compared children (8-12), adolescents (13-17), and adults (≥18) diagnosed with HT1. Symptoms were deemed within the clinical range if the t-scores deviated from normal (ASEBA). The results showed that children were withdrawn/depressed, anxious/depressed, and had inattention/ADHD. In total, 53% of the children had attention problems of clinical importance. Both children and adolescents had social problems, while all groups had issues with thought and rule-breaking behavior.
9	*(Garcia et al., 2017)*	Original	12, 1-19y	Chilean	The authors investigated long-term neurological outcomes in HT1 patients treated with protein-restricted diets and (or) nitisinone (NTBC). 29-38% of the patients (assessed at different ages) had delayed development (Bayley scale MDI). Also, results indicate that IQ declined over time if the patient was diagnosed before eight months of age.
10	*(van Ginkel et al., 2016)*	Original	19, 7-24y	British, Belgian, Dutch	The authors compared the neuropsychiatric phenotypes in 19 NTBC and dietary treated HT1 children to healthy controls. They found that HT1 patients had lower estimated IQ, executive function (working memory, cognitive flexibility), and social cognition. The authors also report a negative correlation between duration of NTBC treatment and IQ.

11	*(Pohorecka et al., 2012)*	Original	8, N/A	Polish	The authors investigated neuropsychiatric comorbidities during NTBC and dietary treatment. The results show a high prevalence of attention deficit, possibly correlated with plasma tyrosine levels. The article also reports a reverse correlation between attention deficit and verbal scale results.
12	*(Barroso et al., 2020)*	Original/ review	2, 0 and 8y	Portuguese	The authors present two siblings born to consanguineous parents and diagnosed with HT3. The younger boy was diagnosed with neonatal screening, while the sister was diagnosed at age 8. Both displayed speech problems, hyperactivity, impulsivity, learning difficulties, and both were diagnosed with ADHD at age 5. Follow-up (age 7 and 15) revealed "borderline" development in the boy and mild intellectual impairment in the older sister. To compare, the authors reviewed 17 published case reports and found large variations in development, from normal to severely intellectually impaired.
13	*(Blundell et al., 2018)*	Original	11, 4.40-19.58y	British	The authors investigated cognitive abilities in mucopolysaccharidosis IV and HT3 patients. In the 11 HT3 patients, the results showed impaired language, sustained attention, and age-related decline compared to healthy controls.
14	*(Gokay et al., 2016)*	Original	2, 1 and 4y	Turkish	The authors present two siblings, consanguineous parents, diagnosed with HT2. In contrast to the boy, the sister was asymptomatic. Both had a novel TAT mutation. Follow-up (two years after) showed that the boys' mental status gradually declined, while the sister remained within normal limits. However, the authors speculate that this could relate to her young age, suggesting that she might display symptoms later like her brother.
15	*(Scott, 2006)*	Review	N/A	N/A	A review on the genetic tyrosinemias HT1, HT2, and HT3. The suggested link between development delay and high tyrosine levels in early life (HT2 patients) is of particular interest. The author also reviewed reports of mental retardation and cognitive decline in HT3 patients; however, a clear association was not yet confirmed at the time of this review.
16	*(Ellaway et al., 2001)*	Original/ Review	13, 0-19y	N/A	The authors presented four HT3 case reports and patients previously found in the literature. While some presented asymptomatic, others had neurological impairments effecting learning and language, developmental delay, mental retardation, hyperactivity, and self-injury. The most common long-term comorbidity was intellectual impairment, present in 75% of the patients (n=9/12).
Alkaptonuria					

17	*(Kisa et al., 2021)*	Original	66, 1-62y	Turkish	The authors investigated AKU phenotype (and genotype) in 66 patients across different centers in Turkey. Most patients present with distinct somatic features. Eight patients were diagnosed with depression following psychiatric evaluations; however, the authors speculate that this could be related to low self-esteem linked to physical appearance.
18	*(Davison et al., 2018)*	Original	63, 16-75y	English, Scottish	The authors compared BDI-II scores pre-NTBC treatment (n=63) to 12 (n=39) and 24 months treatment (n=32), as well as biochemical markers of depression. Although they found a significant increase compared to baseline, most patients were categorized in "minimal" depression. They conclude that NTBC treatment does not substantially alter mood.
Succinic Semialdehyde Dehydrogenase (SSADH) deficiency					
19	*(Pearl et al., 2021)*	Original	28, 18m-40y	American	The authors summarize the SSADH deficiency natural history study at the halfway point (2020). 28 patients were included and presented with intellectual disability (89%), speech delay (89%), ADHD (61%), sleep disturbance (54%), and OCD/anxiety (54%).
20	*(DiBacco et al., 2019)*	Original	133, 8w-63y	N/A	The authors investigated 133 patients, of which 49 participated in a longitudinal study. The patients presented with speech delay (77%), intellectual disability (57%), ADHD (53%), sleep disturbance (44%), and repetitive behavior/OCD (34.6%). In addition, older patients (>12 years) had an increased prevalence of compulsive behavior, sleep disturbances (and seizures).
21	*(Knerr et al., 2008)*	Original	33, 10.1-39.5y	Multiple	The authors studied 33 patients, 6 (18%) of which had parental consanguinity. Overall, 82% had behavior problems, i.e., attention deficit, hyperactivity, anxiety and aggression. Other (relevant) clinical findings included: speech delay (55%), autistic features (12%), sleep disturbances (45%),
22	*(Gibson et al., 2003)*	Original/ review	2, 22- and 36-year-old + 56 reviewed	Turkish	The authors present two case reports (1M, 1F), who presented with language development, hyperactivity, impulsivity, tantrums, aggressiveness, developmental delay, mental retardation, autistic behavior, hallucinations, and sleep disorders. The authors also reviewed 56 cases (51 published, 5 unpublished) and found a 43% incidence of behavior problems.
23	*(Pearl et al., 2003)*	Original/ review	9, 0-21y +51 reviewed	Multiple	The authors compared 51 case reports from the literature to their own 9 cases. The following clinical characteristics were reported for 60 patients: mental retardation (83%), language delay (82%), behavior problems (inattention,

					hyperactivity, aggression, impulsivity) (50%), hallucinations, OCD, psychotic symptoms, and sleep problems.
24	*(Gibson et al., 1997)*	Original	23, 3m-25y	Multiple	The authors studied 23 patients from 20 families (43% consanguinity). Clinical characteristics included: language delay (78%), mental delay (74%), behavioral problems (30%), psychosis (22% or less). 30% of the patients had normal development.
Maple Syrup Urine Disease (MSUD)					
25	*(Medina et al., 2021)*	Original	45, 1.2-29.1y	Chilean	The authors aimed to determine the phenotypes of patients in the Chilean MSUD cohort. Neurocognitive evaluation (BD-II or WPPSI/WISC-III/WAIS-IV) of 37 patients showed various degrees of developmental delay: borderline-mild (30%, n=11), moderate (3%, n=1), moderate-severe (24%, n=9), and severe (27%, n=10). Six patients (16%) had IQ within normal limits.
26	*(Strauss et al., 2020)*	Original	184, 0.1-52.9y	American	The authors performed long-term investigations of MSUD patients to determine their clinical and biochemical phenotypes. Several of these patients presented with intellectual impairment. In addition, there was a high prevalence of depression, anxiety, and panic disorder among patients that underwent appropriate tests (n=37).
27	*(Abi-Warde et al., 2017)*	Original	35, 2.1-49y	French	The authors studied phenotypes in patients (n=35) diagnosed during their neonatal period. Notably, consanguinity was reported in 23% of the cases. Out of 35 patients: 31% reported delayed academic achievement and 57.1% needed speech therapy or psychomotor/occupational therapy. 56% of patients (n=19/34) was in psychiatric counselling or therapy (occasional: n=14, sustained: n=5). Among these, three had externalizing disorders, four had mood/emotional disorders, and one had anxiety.
28	*(Muelly et al., 2013)*	Original	37, 5-35 years	American (26 Old Order Mennonite)	The authors report an 83% lifetime incidence of depression, anxiety, inattention, and impulsivity. Compared to controls, MSUD patients had lower full-scale IQ scores. In addition, the cumulative lifetime incidence of ADHD differed in dietary treated patients (54%) and liver transplant patients (82%).
X-linked ichthyosis (XLI)					
29	*(Diociaiuti et al., 2019)*	Original	35, 10d-58y	Italian	The authors report clinical and molecular findings in XLI patients. In total, 42.4% (n=14/33) of the patients presented with neuropsychiatric comorbidities. ADHD diagnosis was present in 27%, five of whom had been diagnosed pre-study. In

					addition, two patients had mental retardation, and one presented with autistic features.
30	*(Rodrigo-Nicolas et al., 2018)*	Original	30, 0-80y	Spanish	The authors studied patients with clinical and genetic XLI. All patients had somatic features common for XLI, and 30% were diagnosed with ADHD. In addition, one patient was diagnosed with Tourette's syndrome.
31	*(Chatterjee et al., 2016)*	Original	76, 11±1y (children) 42±2y (adults)	Multiple	The authors collected information via an online survey available worldwide. 58 adult males and 24 boys were included in the study. Together, 41% of the patients were already diagnosed with a neuropsychiatric disorder (mood/development-related). 54% (n=37/68) patients reported ≥4 ADHD-related symptoms. Both young and adults scored higher in inattention, autism-related symptoms, impulsivity, and behavioral difficulties compared to the general populations
32	*(Kent et al., 2008)*	Original	25, >18y	British	The authors performed a targeted study of ADHD and autism in boys diagnosed with XLI. In 25 patients, 40% were diagnosed with ADHD, of which 80% were classified as the inattentive subtype. ASD or related language/communication difficulties were present in five patients.
Mitochondrial Encephalomyopathy with lactic acidosis and stroke-like episodes (MELAS)					
33	*(Moore et al., 2020)*	Review	N/A	N/A	A systematic review of prevalence of cognitive findings in patients with mitochondrial disorders. The authors refer to ten review papers including MELAS patients where clinical findings were: cognitive impairment, learning disabilities, cognitive decline, memory problems/dementia, and developmental delay.
34	*(Kraya et al., 2019)*	Original	10, 24-62y	German	The authors studied clinical severity, cognitive impairment, and MRI changes in MELAS patients. Patients had significantly impaired (visual and divided) attention, executive functioning, visuoperception, and -construction. In addition, the authors found a correlation between lesion load and cognitive impairment.
35	*(El-Hattab et al., 2015)*	Review	N/A	N/A	A minireview detailing the pathophysiology of MELAS patients found in the literature (2015). Multiple neurocognitive findings were reported, for instance, learning disabilities and memory impairment (50-74% prevalence), depression, anxiety, and psychotic disorders (25-49% prevalence).
36	*(Anglin et al., 2012)*	Original/ review	1, 45y +50 reviewed	Canadian, N/A	The authors present a case report of a female MELAS patient. She presented with low mood, sleep impairment, suicidality, auditory hallucinations, psychotic depression, cognitive decline, and self-injury before eventually being diagnosed with MELAS. The authors also reviewed 50 cases of mitochondrial disorders with

					a psychiatric phenotype. 52% were MELAS patients presenting with symptoms/disorders such as: major depressive disorder (n=4), psychotic features (n=1), cognitive impairment (n=11), psychotic disorder (n=15), anxiety disorder (n=6), and personality change (n=2).
37	*(Neargarder et al., 2007)*	Original	2, 13 and 33y	American	The authors present two case reports, a white boy and an African American male. Both patients had impaired language, attention and executive functioning (working memory). The youngest patient also had intellectual impairment and impulsivity.
Mucopolysaccharidosis type III (MPS III, Sanfilippo syndrome)					
38	*(Hoffmann et al., 2020)*	Original/ review	18, 9.8y (mean)	German	The authors analyzed questionnaires from German patients with mucopolysaccharidosis I-III. The response rate was a mere 13.8%, of which 18 patients were diagnosed with MPS III. All patients presented with developmental delay, agitation, sleep disturbance, and hyperactivity.
39	*(Kong et al., 2020)*	Original	34, 1-17y	Chinese	The authors used clinical tests, questionnaires, and scoring systems to analyze neuropsychiatric phenotypes in 34 MPS III patients. Patients presented with speech delay (100%), intellectual disability (100%), and hyperactivity (94.12%).
40	*(Andrade et al., 2015)*	Review	N/A	N/A	The authors reviewed common neuropsychiatric findings from case-reports, usually onset between age 2 and 5. The symptoms included hyperactivity, aggressiveness, anxiety, speech difficulties, developmental delay, and sleep disorders.
41	*(Wijburg et al., 2013)*	Review	N/A	N/A	The authors reviewed misdiagnosis of ADHD, ASD and developmental delay with MPS III. Symptoms usually occur in three phases: (I, 1-3y) developmental delay, often speech-related, (II, 3-4y) behavioral problems, i.e., hyperactivity, impulsivity, autistic-related behavior, and anxiety, and sleep impairment (>90% of reported cases in children), and (III, teens) behavioral problems cease, but patients get progressively worse (somatic and cognitive) usually ending in a vegetative state followed by death.
42	*(Valstar et al., 2011)*	Original	69, 0-70y	Dutch	The authors reported high prevalence of hyperactivity and attention deficit. 39 patients (developmental age > 3 months) underwent psychometric developmental tests. Based on these tests, the authors found high variance in intellectual disability based on MPSIII subtype.
43	*(Heron et al., 2011)*	Original	374	French, British, Greek	A comparative study of French (n=128), British (n=126), and Greek (n=17) patients. During follow-up, most patients presented with neurological

					manifestations, e.g., speech delay, abnormal behavior (hyperactivity, aggression, irritability), ASD, and cognitive delay.
44	*(Bax and Colville, 1995)*	Original	96, 0-14y	British	The study included 96 MPSIII aged 14 year and younger. When assessing neuropsychiatric symptoms, the authors found: sleep problems (86%), destructive behavior (57%), hyperactivity (cannot settle) (69%), and fearful (anxiety) (55%).

ASD: autism spectrum disorder; **OCD:** obsessive-compulsive disorder; **ADD:** attention deficit disorder; **ADHD:** attention deficit hyperactivity disorder; **BDI-II:** Becks Depression Inventory-II (BDI-II); **WPPSI, WISC-III, WAIS-IV**: Wechsler Intelligence Scale

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