Supplementary material 2: List of references for Figure 1 and Table 1.

Neurotransmiter synthesis deficiency	
AD GTP-CH1 deficiency	Wijemanne 2015; Trender-Gerhard 2009; Hyland 2008 [1–3]
Aminoacidopathies	
Cerebral organic acidurias	Gelener 2020; Tabarestani 2020; Mainka 2020; Tsai 2017; Pierson 2015; Herskovitz 2013; Marcel 2012; Karatas 2010; Angle 2008; Periasamy 2008; Kulkens 2005; Bahr 2002; Fujitake 1999 [4–16]
Phenylcetonuria	Chen 2019; Wang 2018; Tufekcioglu 2016; Rosini 2014; Vockley 2014; Bilder 2013; Kasim 2001 [17–23]
Urea cycle disorders	Anderson 2020; Koya 2019; Panza 2019; Bigot 2017; Maillot 2016; Anstey 2015; Atiq 2008; Maillot 2007; Tuchman 2001 [24–32]
Genetic hyperhomocystinemia : MTHFR or Cobalamin metabolism defects and CBS deficiency	Marelli 2021; Vieira 2020; Wei 2020; Weber Hoss 2019; Gales 2018; Morris 2017; Norris 2017; Stabler 2013; Michot 2008; Kelly 2003; Powers 2001 [33-43]
Lysosomal storage disorders	
Niemann Pick-C	Sitarska et Lugowska 2019; Nadjar 2018; Lazzaro 2016; Maubert 2013 [44–47]
Gaucher III	Leurs 2018; El-Beshlawy 2017; Stirnemann 2017; Mistry 2015; Grabowski 2015; Ben Rhouma 2012; Vellodi 2009; Guimaraes 2002; Charrow 1998; Neil 1979 [48–57]
Krabbe	Xia 2020; Cousyn 2019; Zhang 2018; Escolar 2017; Liao 2017; Adachi 2016; Debs 2013 [58–64]
Metachromatic leukodystrophy	Van Rappard 2015; Hahn 1982 [65,66]
Fabry	Ortiz 2018; Curiati 2017; Smid 2015; Ghali 2012; Hegemann 2006 [67–71]
Pompe	Hossain 2018; Chan 2016; Young 2003; Umapathysivam 2001 [72–75]

Peroxysomal disorders	
X-ALD	Rattay 2020; Mannari 2020; Shamim 2017 [76–78]
Refsum	Stepien 2016; Bompaire 2015; Wanders 2011; Britton 1989 [79–82]
AMACR deficiency	Smith 2010; Kapina 2010; Clarke 2004; McLean 2002; Ferdinandusse 2000 [83–87]
Disorders of vitamin metabolism	
Pyridoxine-dependent epilepsy	Osman 2020; Xue 2019 [88,89]
Biotinidase deficiency	Radelfahr 2020; Van Winckel 2020; Van Iseghem 2019; Wolf 2019; Deschamps 2018; Yilmaz 2017; Bottin 2015; Cowan 2010 [90–97]
Abetalipoproteinemia	Lee 2014; Nagappa 2014; Zamel 2008 [98–100]
AVED	El Euch-Fayache 2014; Cavalier 1998; Finckh 1995; Gotoda 1995 [101–104]
Cerebral folate deficiency	Pope 2019; Masingue 2019; Sadighi 2012 [105–107]
TBRBGD	Tabarki 2013; Kono 2009 [108,109]
Riboflavin transporter deficiency	Carreau 2020; Foley 2014 [110,111]
Disorders of energy metabolism	
GLUT 1	Leen 2013; Afawi 2010; Veggiotti 2010 [112–114]
PDC deficiency, Coenzyme Q10 deficiency	Pavlu-Pereira 2020; Rahman 2012; Sedel 2008; Quinzii 2007; Mellick 2004; Ogasahara 1989 [115–120]
LHON	Ciron 2018; Carelli 2017; Martikainen 2016; Pfeffer 2013; Palace 2009; McFarland 2007; Gilhuis 2006; Horvath 2000; Nikoskelainen 1995 [121–129]
Disorders of beta-oxydation: MADD, MCAD and MTP/LCHAD	Nadjar 2020; Chen 2019; De Biase 2017; Wang 2016; Grunert 2014; Rosenbohm 2014; Liewluck 2013; Schatz 2010; Lang 2009; Abdenur 2001; Van Hove 1993 [130–140]
Metal toxicity	

Wilson	Yong 2019; Guillaud 2018; Bandmann 2015; Roberts 2008; Ferenci 2007 [141–145]
Aceruleoplasminemia	Marchi 2019; Kono 2012 [146,147]
SLC30A10 deficiency	Quadri 2012; Tuschl 2012; Gospe 2000 [148–150]
Others	
CTX	Makary 2018; Sasamura 2018; Degos 2016; Nakashima 1994; Leitersdorf 1993 [151–155]
Acute intermitent porphyria	Simon 2018; Kevelam 2016; Bonkovsky 2014; Hervé 2010 [156–159]
Tangier	Hooper 2020; Mercan 2018 [160,161]

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