Supplementary material Table S2. Excluded articles and reasons for exclusion(n = 202).

|  |  |  |
| --- | --- | --- |
| Reference | Author,Year | Reason for exclusion |
| ([1](#_ENREF_1)) | Ameis et al 1995 | 1 |
| ([2](#_ENREF_2" \o "Burton, 2015 #37)) | Burton et al 2015 | 1 |
| ([3](#_ENREF_3" \o "Burton, 2017 #38)) | Burton et al 2017 | 1 |
| ([4](#_ENREF_4" \o "Canbay, 2018 #42)) | Canbay et al 2018 | 1 |
| ([5](#_ENREF_5" \o "Castro Narro, 2019 #7046)) | Castro Narro et al 2019 | 1 |
| ([6](#_ENREF_6" \o "Chora, 2017 #50)) | Chora et al 2017 | 1 |
| ([7](#_ENREF_7" \o "Ivashkin, 2017 #115)) | Ivashkin et al 2017 | 1 |
| ([8](#_ENREF_8" \o "Kale, 1995 #121)) | Kale et al 1995 | 1 |
| ([9](#_ENREF_9" \o "Lohse, 2000 #164)) | Lohse et al 2000 | 1 |
| ([10](#_ENREF_10" \o "Muntoni, 1995 #190)) | Muntoni et al 1995 | 1 |
| ([11](#_ENREF_11" \o "Rashu,  #670)) | Rashu et al 2019 | 1 |
| ([12](#_ENREF_12" \o "Sjouke, 2015 #244)) | Sjouke et al 2015 | 1 |
| ([13](#_ENREF_13" \o "Lageron, 1988 #148)) | Lageron et al 1988 | 2 |
| ([14](#_ENREF_14" \o "Young, 1979 #288)) | Young et al 1979 | 2 |
| ([15](#_ENREF_15" \o "Browne, 2003 #35)) | Browne et al 2003 | 2 |
| ([16](#_ENREF_16" \o "Buitrago Mata, 2003 #36)) | Buitrago et al | 2 |
| ([17](#_ENREF_17" \o "Carter, 1974 #44)) | Carter et al 1974 | 2 |
| ([18](#_ENREF_18" \o "Chowdhury, 2009 #51)) | Chowdhury et al 2009 | 2 |
| ([19](#_ENREF_19" \o "Crocker, 1965 #7054)) | Crocker et al 1965 | 2 |
| ([20](#_ENREF_20" \o "Gerner, 2018 #89) | Gerner et al 2018 | 2 |
| ([21](#_ENREF_21" \o "Konno, 1966 #134)) | Konno et al 1966 | 2 |
| ([22](#_ENREF_22" \o "Lake, 1970 #150)) | Lake et al 1970 | 2 |
| ([23](#_ENREF_23" \o "Lowden, 1970 #169)) | Lowden et al 1970 | 2 |
| ([24](#_ENREF_24" \o "Mallikarjuna Swamy, 1997 #173)) | Mallikarjuma Swamy 1997 | 2 |
| ([25](#_ENREF_25" \o "Mello, 1982 #183)) | Mello et al 1982 | 2 |
| ([26](#_ENREF_26" \o "Panchagnula, 2000 #206)) | Panchagnula et al 2000 | 2 |
| ([27](#_ENREF_27" \o "Peña Alonso, 1994 #210)) | Pena-Alonso et al 1994 | 2 |
| ([28](#_ENREF_28" \o "Petropoulou, 2017 #214)) | Petropoulo et al 2017 | 2 |
| ([29](#_ENREF_29" \o "Ramón, 2000 #229)) | Ramon et al 2000 | 2 |
| (30) | Sen et al 2015 | 2 |
| ([31](#_ENREF_31" \o "Shenoy, 2014 #241)) | Shenoy et al 2014 | 2 |
| ([32](#_ENREF_32" \o "Sty, 1978 #251)) | Sty et al 1978 | 2 |
| ([33](#_ENREF_33" \o "Aquino, 2020 #606)) | Aquino et al 2020 | 3 |
| ([34](#_ENREF_34" \o "White, 2015 #280)) | White et al 2015 | 3 |
| ([35](#_ENREF_35" \o "Aslan, 2016 #12)) | Aslan et al 2016 | 3 |
| ([36](#_ENREF_36" \o "Berdon, 1969 #21)) | Berdon et al 1969 | 3 |
| ([37](#_ENREF_37" \o "Bernstein, 2017 #25)) | Bernstein et al 2017 | 3 |
| ([38](#_ENREF_38" \o "Bernstein, 2020 #612)) | Bernstein et al 2020 | 3 |
| ([39](#_ENREF_39" \o "Boldrini, 2000 #28)) | Boldrini et al 2000 | 3 |
| ([40](#_ENREF_40" \o "Borges, 2020 #613)) | Borges et al 2020 | 3 |
| ([41](#_ENREF_41" \o "Breen, 2016 #33)) | Breen et al 2016 | 3 |
| ([42](#_ENREF_42" \o "Burton,  #616)) | Burton et al 2019 | 3 |
| ([43](#_ENREF_43" \o "Burton,  #617)) | Burton et al 2019 | 3 |
| ([44](#_ENREF_44" \o "Kulhas Celik, 2019 #649)) | Kulhas Celik et al 2019 | 3 |
| ([45](#_ENREF_45" \o "Coates, 1978 #52)) | Coates et al 1978 | 3 |
| ([46](#_ENREF_46" \o "Deegan, 2014 #61)) | Deegan et al 2014 | 3 |
| ([47](#_ENREF_47" \o "Drogari, 2013 #64)) | Drogari et al 2013 | 3 |
| ([48](#_ENREF_48" \o "Drogari, 2010 #65)) | Drogari et al 2010 | 3 |
| ([49](#_ENREF_49" \o "Ghosh, 2016 #90)) | Ghosh et al 2016 | 3 |
| ([50](#_ENREF_50" \o "Ghosh, 2017 #91)) | Ghosh et al 2017 | 3 |
| ([51](#_ENREF_51" \o "Glueck, 1992 #92)) | Glueck et al 1992 | 3 |
| ([52](#_ENREF_52" \o "Goncalves, 2016 #94)) | Gonçalves A et al 2016 | 3 |
| ([53](#_ENREF_53" \o "Gramatges, 2009 #97)) | Gramatgets et al 2009 | 3 |
| ([54](#_ENREF_54" \o "Grant, 2020 #638)) | Grant et al 2020 | 3 |
| ([55](#_ENREF_55" \o "Kalkan Ucar, 2010 #122)) | Kalkan et al 2010 | 3 |
| ([56](#_ENREF_56" \o "Lin, 1976 #160)) | Lin et al 1976 | 3 |
| ([57](#_ENREF_57" \o "Long, 2020 #656)) | Long et al 2020 | 3 |
| ([58](#_ENREF_58" \o "Maslen, 1995 #177)) | Maslen et al 1995 | 3 |
| ([59](#_ENREF_59" \o "Pagani, 1996 #203)) | Pagani et al 1996 | 3 |
| ([60](#_ENREF_60" \o "Ramadza, 2015 #228)) | Ramadza et al 2015 | 3 |
| ([61](#_ENREF_61" \o "Tummolo, 2016 #265)) | Tummolo et al 2016 | 3 |
| ([62](#_ENREF_62" \o "Abel, 2014 #1)) | Abel et al 2014 | 4 |
| ([63](#_ENREF_63" \o "Aldeen, 1999 #6)) | Aldeen et al 1999 | 4 |
| ([64](#_ENREF_64" \o "Anderson, 2010 #9)) | Anderson et al 2010 | 4 |
| ([65](#_ENREF_65" \o "Benlian, 2014 #20)) | Benlian et al 2014 | 4 |
| ([66](#_ENREF_66" \o "Besley, 1984 #26)) | Besley et al 1984 | 4 |
| ([67](#_ENREF_67" \o "Byrd Iii, 1979 #39)) | Byrd et al 1979 | 4 |
| ([68](#_ENREF_68" \o "Cebolla, 2016 #48)) | Cebolla et al 2016 | 4 |
| ([69](#_ENREF_69" \o "Erbe, 2014 #74)) | Erbe et al 2014 | 4 |
| ([70](#_ENREF_70" \o "Eto, 1970 #75)) | Eto et al 1970 | 4 |
| ([71](#_ENREF_71" \o "Fang, 2017 #77)) | Fang et al 2017 | 4 |
| ([72](#_ENREF_72" \o "Fulcher, 1998 #85)) | Fulcher et al 1998 | 4 |
| (73) | Fumic et al 2010 | 4 |
| ([74](#_ENREF_74" \o "Gargus, 2012 #87)) | Gargus et al 2012 | 4 |
| ([75](#_ENREF_75" \o "Grabowski, 2013 #96)) | Grabowski 2013 | 4 |
| ([76](#_ENREF_76)) | Harrisson et al 1997 | 4 |
| ([77](#_ENREF_77" \o "Hart, 2016 #104)) | Hart et al 2016 | 4 |
| ([78](#_ENREF_78" \o "Heiner-Fokkema, 2016 #106)) | Heiner-Fokkema et al 2016 | 4 |
| ([79](#_ENREF_79" \o "Hendriksz, 2012 #107)) | Hendrinksz et al 2012 | 4 |
| ([80](#_ENREF_80" \o "Jones, 2014 #116)) | Jones et al 2014 | 4 |
| ([81](#_ENREF_81" \o "Kamalian, 1973 #123)) | Kamalian et al 1973 | 4 |
| ([82](#_ENREF_82" \o "Klaudel-Dreszler, 2016 #130)) | Klaudel-Dreszler et al 2016 | 4 |
| ([83](#_ENREF_83" \o "Kodali, 2016 #132)) | Kodali et al 2016 | 4 |
| ([84](#_ENREF_84" \o "Leone, 1991 #157)) | Leone et al 1991 | 4 |
| ([85](#_ENREF_85" \o "Leone, 1995 #158)) | Leone et al 1995 | 4 |
| ([86](#_ENREF_86" \o "Lourenco, 2013 #167)) | Lourenco et al 2013 | 4 |
| ([87](#_ENREF_87" \o "Lourenco, 2013 #168)) | Lourenco et al 2013 | 4 |
| ([88](#_ENREF_88" \o "Marshall, 1969 #175)) | Marshall et al 1969 | 4 |
| ([89](#_ENREF_89" \o "Matthews, 2004 #178)) | Matthews et al 2004 | 4 |
| ([90](#_ENREF_90" \o "O'Leary, 2013 #196)) | O’Leary et al 2013 | 4 |
| ([91](#_ENREF_91" \o "Ozmen, 1992 #199)) | Ozmen et al 1992 | 4 |
| ([92](#_ENREF_92" \o "Patel, 2018 #209)) | Patel et al 2018 | 4 |
| ([93](#_ENREF_93" \o "Perezpeña Diazconti, 2016 #212)) | Perezpena et al 2016 | 4 |
| ([94](#_ENREF_94" \o "Queloz, 1972 #223)) | Queloz et al 1972 | 4 |
| ([95](#_ENREF_95" \o "Raafat, 1973 #226)) | Raafat et al 1973 | 4 |
| ([96](#_ENREF_96" \o "Rennie, 2018 #231)) | Rennie et al 2018 | 4 |
| ([97](#_ENREF_97" \o "Slae, 2018 #245)) | Slae et al 2018 | 4 |
| ([98](#_ENREF_98" \o "Strokova, 2017 #250)) | Strokova et al 2017 | 4 |
| ([99](#_ENREF_99)) | Uno et al 1973 | 4 |
| ([100](#_ENREF_100" \o "Van Spronsen, 2016 #272)) | Van Spronsen et al | 4 |
| ([101](#_ENREF_101" \o "Watanuki, 1967 #277)) | Watanuki et al 1966 | 4 |
| ([102](#_ENREF_102" \o "Wong, 2012 #284)) | Wong et al 2012 | 4 |
| ([103](#_ENREF_103" \o "Yahyaoui, 2017 #286)) | Yahyahoui et al 2017 | 4 |
| ([104](#_ENREF_104" \o "Bernstein, 2018 #24)) | Bernstein et al 2018 | 5 |
| ([105](#_ENREF_105" \o "Eminoglu, 2016 #73)) | Eminoglu et al 2016 | 5 |
| ([106](#_ENREF_106" \o "Eto, 1978 #76)) | Eto et al 1978 | 5 |
| ([107](#_ENREF_107" \o "Guardamagna, 2014 #98)) | Guardamagna et al 2014 | 5 |
| ([108](#_ENREF_108" \o "Huang, 2012 #113)) | Huang et al 2012 | 5 |
| ([109](#_ENREF_109" \o "Konno, 1993 #133)) | Konno 1993 | 5 |
| ([110](#_ENREF_110" \o "Lavrova, 2017 #151)) | Lavrova et al 2017 | 5 |
| ([111](#_ENREF_111" \o "Ma, 2013 #170)) | Ma et al 2013 | 5 |
| ([112](#_ENREF_112)) | Quinn et al 2014 | 5 |
| ([113](#_ENREF_113" \o "Surkov, 2020 #680)) | Surkov et al 2020 | 5 |
| ([114](#_ENREF_114" \o "Tanaka, 1998 #255)) | Tanaka 1998 | 5 |
| ([115](#_ENREF_115" \o "Tanaka, 2006 #256)) | Tanaka 2006 | 5 |
| ([116](#_ENREF_116" \o "Lageron, 1978 #146)) | Lageron et al 1978 | 6 |
| ([117](#_ENREF_117" \o "Amraoui, 2015 #8)) | Amraoui et al 2015 | 6 |
| ([118](#_ENREF_118" \o "Aubert-Tulkens, 1980 #13)) | Aubert-Tulkens 1980 | 6 |
| ([119](#_ENREF_119" \o "Bambirra, 1982 #14)) | Bambirra et all 1982 | 6 |
| ([120](#_ENREF_120" \o "Bernstein, 2013 #22)) | Bernstein et al 2013 | 6 |
| ([121](#_ENREF_121" \o "Bona, 1989 #30)) | Bona et al 1989 | 6 |
| ([122](#_ENREF_122" \o "Bretagne, 1981 #34)) | Bregtane et al 1981 | 6 |
| ([123](#_ENREF_123" \o "Caseiro, 2016 #45)) | Caseiro et al 2016 | 6 |
| ([124](#_ENREF_124" \o "Castro, 1999 #46)) | Castro et al 1999 | 6 |
| ([125](#_ENREF_125" \o "Coelho, 1987 #53)) | Coelho et al 1987 | 6 |
| ([126](#_ENREF_126" \o "Contreras, 1974 #54)) | Contreras et a 1974 | 6 |
| ([127](#_ENREF_127" \o "Elleder, 1999 #69)) | Elleder et al 1999 | 6 |
| ([128](#_ENREF_128" \o "Elsayed, 2014 #72)) | Elsayed et al 2014 | 6 |
| ([129](#_ENREF_129" \o "Feillet, 2009 #79)) | Feillet et al 2009 | 6 |
| ([130](#_ENREF_130" \o "Fernandez-Aragon, 2004 #80)) | Fernandez-Aragon et al 2004 | 6 |
| ([131](#_ENREF_131" \o "Foster, 1987 #82)) | Foster et al 1987 | 6 |
| ([132](#_ENREF_132" \o "Gautier, 1978 #88)) | Gautier et al 1978 | 6 |
| ([133](#_ENREF_133" \o "Guazzi, 1968 #99)) | Guazzi et al 1968 | 6 |
| ([134](#_ENREF_134" \o "Guazzi, 1968 #100)) | Guazzi et al 1968 | 6 |
| ([135](#_ENREF_135" \o "Hanak, 1984 #102)) | Hanak et al 1984 | 6 |
| ([136](#_ENREF_136" \o "Haznedar, 2018 #105)) | Haznedar et al 2018 | 6 |
| ([137](#_ENREF_137" \o "Hicks, 2014 #108)) | Hicks et al 2014 | 6 |
| ([138](#_ENREF_138" \o "Higuchi, 1997 #109)) | Higuchi et al 1997 | 6 |
| ([139](#_ENREF_139" \o "Hill, 1983 #110)) | Hill et al. 1983 | 6 |
| ([140](#_ENREF_140" \o "Ho, 1978 #112)) | Ho et al 1978 | 6 |
| ([141](#_ENREF_141" \o "Justus, 1988 #119)) | Justus et al 1988 | 6 |
| ([142](#_ENREF_142" \o "Kahana, 1968 #120)) | Kahana et al 1988 | 6 |
| ([143](#_ENREF_143" \o "Keller, 1977 #126)) | Keller et al 1977 | 6 |
| ([144](#_ENREF_144" \o "Krebs, 2010 #137)) | Krebs et al 2010 | 6 |
| ([145)](#_ENREF_145" \o "Kunnert, 1977 #142) | Kunnert 1977 | 6 |
| ([146](#_ENREF_146)) | Kunnert et al 1987 | 6 |
| ([147](#_ENREF_147" \o "Kuntz, 1981 #144)) | Kuntz et al 1981 | 6 |
| ([148](#_ENREF_148" \o "Lageron, 1985 #147)) | Lageron et al 1985 | 6 |
| ([149](#_ENREF_149" \o "Lajo, 1974 #149)) | Lajo et al 1974 | 6 |
| ([150](#_ENREF_150" \o "Leclerc, 1971 #154)) | Leclerc et al 1971 | 6 |
| ([151](#_ENREF_151" \o "Loffredo, 1975 #163)) | Loffredo et al 1975 | 6 |
| ([152](#_ENREF_152" \o "Lough, 1970 #166)) | Lough et a 1970 | 6 |
| ([153](#_ENREF_153" \o "Marosvari, 1985 #174)) | Marosvari 1985 | 6 |
| ([154](#_ENREF_154" \o "Martinez De Compañón, 2004 #176)) | Martinez de Companon et al 2004 | 6 |
| ([155](#_ENREF_155" \o "Mazurová, 2014 #180)) | Mazurova et al 2014 | 6 |
| ([156](#_ENREF_156" \o "McKiernan, 2012 #182)) | McKierna et al 2012 | 6 |
| ([157](#_ENREF_157" \o "Miller, 1982 #186)) | Miller et al 1982 | 6 |
| ([158](#_ENREF_158" \o "Muñoz Bartolo, 2007 #189)) | Munoz Bartolo et al 2007 | 6 |
| ([159](#_ENREF_159" \o "Murakami, 2016 #191)) | Muramaki J. et al 2016 | 6 |
| ([160](#_ENREF_160" \o "Nardi, 1971 #192)) | Nardi et al 1971 | 6 |
| ([161](#_ENREF_161" \o "Neustein, 1972 #195)) | Neustein et al 1973 | 6 |
| ([162](#_ENREF_162" \o "Ozoran, 1978 #200)) | Ozoran et al 1978 | 6 |
| ([163](#_ENREF_163" \o "Ozsoylu, 1992 #201)) | Ozsoylu 1992 | 6 |
| ([164](#_ENREF_164" \o "Ozsoylu, 1977 #202)) | Ozsoylu et al 1977 | 6 |
| ([165](#_ENREF_165" \o "Parada Yáñez, 1996 #207)) | Parada Yanez et al 1996 | 6 |
| ([166](#_ENREF_166" \o "Pastor Bevia, 1987 #208)) | Pastor Bevia et al 1987 | 6 |
| ([167](#_ENREF_167" \o "Perez Rodriguez-Cuesta, 1990 #211)) | Perez Rodrigues-Cuesta 1990 | 6 |
| ([168](#_ENREF_168" \o "Permanetter, 1979 #213)) | Permanetter et al 1979 | 6 |
| ([169](#_ENREF_169" \o "Pisciotta, 2017 #217)) | Pisciotta et al 2017 | 6 |
| ([170](#_ENREF_170" \o "Pohlandt, 1988 #220)) | Pohlandt et al 1988 | 6 |
| ([171](#_ENREF_171" \o "Pot, 2008 #222)) | Pot et al 2008 | 6 |
| ([172](#_ENREF_172" \o "Quirici Neto, 1985 #225)) | Quirici et al 1985 | 6 |
| ([173](#_ENREF_173" \o "Rassoul, 2001 #230)) | Rassoul et al 2001 | 6 |
| ([174](#_ENREF_174" \o "Ryzko, 1993 #235)) | Ryzko et al 1993 | 6 |
| ([175](#_ENREF_175" \o "Smolka, 2007 #246)) | Smolka 2007 | 6 |
| ([176](#_ENREF_176" \o "Storm, 1990 #249)) | Storm et al 1990 | 6 |
| ([177](#_ENREF_177" \o "Sundaravalli, 1986 #252)) | Sundaravalli et al 1986 | 6 |
| ([178](#_ENREF_178" \o "Tashiro, 1989 #258)) | Tashiro et al 1989 | 6 |
| ([179](#_ENREF_179" \o "Thavarungkul, 1995 #260)) | Thavarungkul et al 1985 | 6 |
| ([180](#_ENREF_180" \o "Tonissen, 1983 #263)) | Tonissen et al 1983 | 6 |
| ([181](#_ENREF_181" \o "Tylki-Szymanska, 1987 #266)) | Tylki-Szymanska, A et al 1987 | 6 |
| ([182](#_ENREF_182)) | Valerdiz Casasola et al 1997 | 6 |
| ([183](#_ENREF_183)) | Vargas Torcal et al | 6 |
| ([184](#_ENREF_184)) | Vij et al 2018 | 6 |
| ([185](#_ENREF_185)) | Wallis et al 1971 | 6 |
| ([186](#_ENREF_186)) | Weiler et al 2009 | 6 |
| ([187](#_ENREF_187)) | Wolf et al 1974 | 6 |
| ([188](#_ENREF_188)) | Wolman 1992 | 6 |
| ([189](#_ENREF_189)) | Wolman 1993 | 6 |
| ([190](#_ENREF_190)) | Yadav 1995 | 6 |
| ([191](#_ENREF_191)) | Zlatkovic et al 2001 | 6 |
| ([192](#_ENREF_192)) | Zubov et al 1989 | 6 |
| ([193](#_ENREF_193)) | Basu et al 2002 | 7 |
| ([194](#_ENREF_194)) | Leavitt et al 2007 | 7 |
| ([195](#_ENREF_195)) | Philippart et al 1982 | 7 |
| ([196](#_ENREF_196)) | Weiskirchen 2017 | 7 |
| ([197](#_ENREF_197)) | Al-Essa et al 1999 | 8 |
| ([198](#_ENREF_198)) | Arteburn et al 1991 | 8 |
| ([199](#_ENREF_199)) | Jones et al 2016 | 8 |
| ([200](#_ENREF_200)) | Kucukcongar Yava et al 2015 | 8 |
| (201) | Linpinski et al 2018 | 8 |
| ([202](#_ENREF_202)) | Lyons et al 2020 | 8 |

Legend: 1) Studies with adults (over 18 years old) ; 2) Studies not performing laboratorial exam to confirm the diagnosis; 3) Studies in which patients features were not evaluated; 4) Secondary studies (articles reviews, letter to the editor, books and book chapters); 5) Studies not published in Latin Roman Alphabet; 6) Full text not available 7) Studies without patient with lysosomal acid lipase deficiency 8) Studies with previously published case.

References

1. Ameis D, Brockmann G, Knoblich R, Merkel M, Ostlund Jr RE, Yang JW, et al. A 5' splice-region mutation and a dinucleotide deletion in the lysosomal acid lipase gene in two patients with cholesteryl ester storage disease. Journal of lipid research. 1995;36(2):241-50.

2. Burton BK, Deegan PB, Enns GM, Guardamagna O, Horslen S, Hovingh GK, et al. Clinical Features of Lysosomal Acid Lipase Deficiency. Journal of pediatric gastroenterology and nutrition. 2015;61(6):619-25.

3. Burton BK, Silliman N, Marulkar S. Progression of liver disease in children and adults with lysosomal acid lipase deficiency. Current medical research and opinion. 2017;33(7):1211-4.

4. Canbay A, Muller MN, Philippou S, Gerken G, Tromm A. Cholesteryl Ester Storage Disease: Fatal Outcome without Causal Therapy in a Female Patient with the Preventable Sequelae of Progressive Liver Disease after Many Years of Mild Symptoms. The American journal of case reports. 2018;19:577-81.

5. Castro Narro GE, Gamboa Domínguez A, Consuelo Sánchez A, Salazar Martínez A, Agramonte Hevia J, Cebolla JJ, et al. Combined Hepatocellular–Cholangiocarcinoma in a Patient With Cirrhosis Due to Cholesteryl Ester Storage Disease. Hepatology (Baltimore, Md). 2019;69(4):1838-41.

6. Chora JR, Alves AC, Medeiros AM, Mariano C, Lobarinhas G, Guerra A, et al. Lysosomal acid lipase deficiency: A hidden disease among cohorts of familial hypercholesterolemia? Journal of clinical lipidology. 2017;11(2):477-84.e2.

7. Ivashkin V, Zharkova M. Cholesteryl Ester Crystals in Lysosomal Acid Lipase Deficiency. The New England journal of medicine. 2017;376(9):e14.

8. Kale AS, Ferry GD, Hawkins EP. End-stage renal disease in a patient with cholesteryl ester storage disease following successful liver transplantation and cyclosporine immunosuppression. Journal of pediatric gastroenterology and nutrition. 1995;20(1):95-7.

9. Lohse P, Maas S, Lohse P, Elleder M, Kirk JM, Besley GT, et al. Compound heterozygosity for a Wolman mutation is frequent among patients with cholesteryl ester storage disease. Journal of lipid research. 2000;41(1):23-31.

10. Muntoni S, Wiebusch H, Funke H, Ros E, Seedorf U, Assmann G. Homozygosity for a splice junction mutation in exon 8 of the gene encoding lysosomal acid lipase in a Spanish kindred with cholesterol ester storage disease (CESD). Human Genetics. 1995;95(5):491-4.

11. Rashu EB, Junker AE, Danielsen KV, Dahl E, Hamberg O, Borgwardt L, et al. Cholesteryl ester storage disease of clinical and genetic characterisation: A case report and review of literature. World J Clin Cases.8(9):1642-50.

12. Sjouke B, van der Stappen JW, Groener JE, Pepping A, Wevers RA, Gouw A, et al. Hypercholesterolaemia and hepatosplenomegaly: two manifestations of cholesteryl ester storage disease. The Netherlands journal of medicine. 2015;73(3):129-32.

13. Lageron A, Polonovski J. Histochemical abnormalities in liver and jejunal biopsies from a case of cholesterol ester storage disease. Journal of inherited metabolic disease. 1988;11:139-42.

14. Young LW, Sty JR, Babbitt DP. Wolman's disease. American journal of diseases of children (1960). 1979;133(9):959-60.

15. Browne M, Somers G, Savoia H, Kukuruzovic R. Wolman's disease in an infant. British journal of haematology. 2003;122(4):522.

16. Buitrago Mata P, Giacomin Carmiol L, Vallejo Serrano A, Santamaría S. Enfermedad de Wolman: revisión y reporte de un caso. Acta pediátr costarric. 2003;17(2):46-51.

17. Carter AR, France NE, Lewis BW, Shaw DG. Cholesterol ester storage disease. Radiological features. Pediatric radiology. 1974;2(2):135-6.

18. Chowdhury FI, Kabir A, Banik J, Paul P, Kamal M, Parveen HH, et al. A fourteen years old boy with cholesterol ester storage disease. Journal of Medicine. 2009;10(2):146-8.

19. Crocker AC, Vawter GF, Neuhauser EB, Rosowsky A. WOLMAN'S DISEASE: THREE NEW PATIENTS WITH A RECENTLY DESCRIBED LIPIDOSIS. Pediatrics. 1965;35:627-40.

20. Gerner P, Weinhold N, Scheurlen W, Steinbach D, Leichsenring M. Rapid diagnosis of Wolman disease is possible by consideration of characteristic primary symptoms. Journal of pediatric gastroenterology and nutrition. 2018;66:684-5.

21. Konno T, Fujii M, Watanuki T, Koizumi K. Wolman's disease: the first case in Japan. The Tohoku journal of experimental medicine. 1966;90(4):375-89.

22. Lake BD, Patrick AD. Wolman's disease: deficiency of E600-resistant acid esterase activity with storage of lipids in lysosomes. The Journal of pediatrics. 1970;76(2):262-6.

23. Lowden JA, Barson AJ, Wentworth P. Wolman's disease: a microscopic and biochemical study showing accumulation of ceroid and esterified cholesterol. Canadian Medical Association journal. 1970;102(4):402-5.

24. Mallikarjuna Swamy P, Mallikarjuna HB, Shantala CC, Prashanth S, Maiya PP, Dandekar C. Wolman's Disease. Indian journal of pediatrics. 1997;64(4):561-3.

25. Mello AM, Ellinger F, Lourenco LA, Castro LM, Zimmerman MA. Doenca de Wolman (xantomatose familiar).Relato de um caso. Pediatria (Säo Paulo). 1982;4(2):159-62.

26. Panchagnula R, Britto C, Vinod J, Sitalakshmi, Anuradha, Damodar P. Wolman's Disease - A Case Report. Indian Journal of Pathology and Microbiology. 2000;43(1):91-2.

27. Peña Alonso YR, Ramón García G. Enfermedad de Wolman en una niña mexicana. Bol méd Hosp Infant Méx. 1994;51(10):660-4.

28. Petropoulou E, Jones R, Daly A, Ashmore C, Hoban R, Kitchen S, et al. Dietary management of early onset lysosomal acid lipase deficiency (LAL-D/wolman disease): A case report. Journal of Inborn Errors of Metabolism and Screening. 2017;5:75.

29. Ramón GG, Díaz ponce H, Díaz Pérez C, Delgado González E. Niña de tres meses con fiebre, distensión abdominal, vómito e ictericia. Gac méd Méx. 2000;136(4):361-7.

30. Sen D, Satija L, Saxena S, Rastogi V, Singh M. A rare constellation of imaging findings in Wolman disease. Medical journal, Armed Forces India. 2015;71:S448-51.

31. Shenoy P, Karegowda L, Sripathi S, Mohammed N. Wolman disease in an infant. BMJ case reports. 2014;2014.

32. Sty JR, Starshak RJ. Scintigraphy in Wolman's disease. Clinical Nuclear Medicine. 1978;3(10):397.

33. Aquino MB, Borges MS, Lopes IF, Vagnini L, Fonseca JHR, Carneiro ZA, et al. Early enzyme replacement therapy in Wolman disease: Challenges and outcomes. Molecular genetics and metabolism. 2020;129(2):S22.

34. White FJ, Gallagher J, Boyle F, Breen C, Broomfield A, Cawtherley S, et al. Nutritional management of infants with Lysosomal Acid Lipase (LAL) deficiency (Wolman Disease). Journal of inherited metabolic disease. 2015;38(1):S40.

35. Aslan M, Ucar AK, Habibi HA, Namdar Y, Kurugoglu S, Adaletli I. Imaging findings of wolman disease. Pediatric radiology. 2016;46:S205.

36. Berdon WE, Baker DH. Radiographic findings in adrenal disease in infants and children. Adrenal hemorrhage, Wolman's familial xanthomatosis with adrenal calcifications, benign and malignant adrenal tumors. New York state journal of medicine. 1969;69(21):2773-8.

37. Bernstein DL, Schiano T, Desnick RJ. Rapidly progressive hepatosteatosis and liver failure five years after liver transplantation in a patient with lysosomal acid lipase deficiency. Molecular genetics and metabolism. 2017;120(1):S27.

38. Bernstein DL, Desnick RJ. Lysosomal acid lipase deficiency and hematologic cancer predisposition. Molecular genetics and metabolism. 2020;129(2):S31.

39. Boldrini R, Castro M, Marcellini M, Piemonte F, Bosman C. [Wolman's disease: a case with malabsorption and 2 cases with virus- negative fatty liver cirrhosis]. Pathologica. 2000;92(2):130-1.

40. Borges MS, Aquino MB, Vagnini L, Carneiro ZA, Fonseca JHR, Lourenco CM. Lysosomal acid lipase deficiency across ages: Unraveling the clinical spectrum of an under-recognized genetic disorder. Molecular genetics and metabolism. 2020;129(2):S32-S3.

41. Breen C, Jones SA, Douzgou S. A facial gestalt in early onset lysosmal acid lipase deficiency, an evolving phenotype. Molecular genetics and metabolism. 2016;117(2):S30.

42. Burton B, Feillet F, Furuya K, Marulkar S, Balwani M. BENEFITS OF SEBELIPASE ALFA IN CHILDREN AND ADULTS WITH LYSOSOMAL ACID LIPASE DEFICIENCY ARE SUSTAINED FOR UP TO 5 YEARS (ARISE STUDY). Hepatology (Baltimore, Md).70:103A-4A.

43. Burton B, Sanchez AC, Kostyleva M, Martins AM, Marulkar S, Abel F, et al. Efficacy and safety of sebelipase alfa over 144 weeks in a diverse population of children and adults with lysosomal acid lipase deficiency. Journal of hepatology.70(1):E121-E2.

44. Kulhas Celik I, Kucukcongar Yavas A, Unal Uzun O, Siyah Bilgin B, Dibek Misirlioglu E, Gunduz M. Successful sebelipase alfa desensitization in a pediatric patient. Journal of Allergy and Clinical Immunology: In Practice. 2019;7(2):732-3.

45. Coates PM, Cortner JA, Mennuti MT, Wheeler JE. Prenatal diagnosis of Wolman disease. American journal of medical genetics. 1978;2(4):397-407.

46. Deegan P, Burton B, Di Rocco M, Enns G, Guardmagna O, Horslen S, et al. Dyslipidemia and sustained elevations in transaminases from early childhood are common in lysosomal acid lipase deficiency. Atherosclerosis. 2014;235(2):e55-e6.

47. Drogari E, Mollaki V, Skouma A, Zellou A, Kyrkou E, Manolaki N, et al. Cholesteryl ester storage disease in Greece: Longterm follow-up of 24 patients. Journal of inherited metabolic disease. 2013;36(2):S291.

48. Drogari E, Progias P, Koniari E, Manolaki N, Christomanou H. Cholesteryl ester storage disease (acid lipase deficiency): Another factor for early atherosclerosis disease. Experience from the Greek population. Atherosclerosis Supplements. 2010;11(2):114-5.

49. Ghosh A, Ashcroft P, Jones SA, Döffinger R, Schneider M, Wynn RF, et al. Haemophagocytic lymphohistiocytosis and other immunological anomalies in infantile onset lysosomal acid lipase deficiency. Molecular genetics and metabolism. 2016;117(2):S49.

50. Ghosh A, Lum SH, Jones S, Wynn RF. Haematopoietic stem cell transplantation alongside enzyme replacement therapy in infantile onset lysosomal acid lipase deficiency (LALD, Wolman disease). Molecular genetics and metabolism. 2017;120(1):S53.

51. Glueck CJ, Lichtenstein P, Tracy T, Speirs J. Safety and efficacy of treatment of pediatric cholesteryl ester storage disease with lovastatin. Pediatric research. 1992;32(5):559-65.

52. Goncalves AB, Marques CL, Cavalcanti TTSL, Marcolin AC, Goncalves-Ferri WA, Pereira-Celini FM, et al. Case report: LAL Deficiency (LA-D) lysosomal storage disease. European journal of pediatrics. 2016;175(11):1737-8.

53. Gramatges MM, Dvorak CC, Regula DP, Enns GM, Weinberg K, Agarwal R. Pathological evidence of Wolman's disease following hematopoietic stem cell transplantation despite correction of lysosomal acid lipase activity. Bone marrow transplantation. 2009;44(7):449-50.

54. Grant CL, Berger S, Lal TR, Crosby K, Hicks R, Julca D, et al. Persistent EBV viremia in siblings with lysosomal acid lipase deficiency (LAL-D). Molecular genetics and metabolism. 2020;129(2):S67.

55. Kalkan Ucar S, Church H, Savage W, Habif S, Coker M. Fatty liver and dyslipidemia as important clues for diagnosis of cholesterol ester storage disease. Journal of inherited metabolic disease. 2010;33:S123.

56. Lin HJ, Lie Ken Jie MS, Ho FC. Accumulation of glyceryl ether lipids in Wolman's disease. Journal of lipid research. 1976;17(1):53-6.

57. Long V, Li H. A case report of successful treatment of teenage patient with lysosomal acid lipase deficiency. Molecular genetics and metabolism. 2020;129(2):S100.

58. Maslen CL, Babcock D, Illingworth DR. Occurrence of a mutation associated with Wolman disease in a family with cholesteryl ester storage disease. Journal of inherited metabolic disease. 1995;18(5):620-3.

59. Pagani F, Garcia R, Pariyarath R, Stuani C, Gridelli B, Paone G, et al. Expression of lysosomal acid lipase mutants detected in three patients with cholesteryl ester storage disease. Human molecular genetics. 1996;5(10):1611-7.

60. Ramadza DP, Cuk M, Zibar K, Baric M, Sarnavka V, Bilic K, et al. [Lysosomal acid lipase deficiency in children: our experience and a novel possibility of enzyme replacement therapy]. Lijecnicki vjesnik. 2015;137(3):81-7.

61. Tummolo A, Ortolani F, Masciopinto M, Melpignano L, DiMauro AM, Piccinno E, et al. Early treatment with Sebelipase-alfa of two young LAL-D siblings: First outcome data. Journal of inherited metabolic disease. 2016;39:S216.

62. Abel F, Burton B, Deegan P, Di Rocco M, Enns G, Guardamagna O, et al. Dyslipidemia and sustained transaminase elevations from early childhood are common in lysosomal acid lipase deficiency. Journal of inherited metabolic disease. 2014;37(1):S160-S1.

63. Aldeen HAZ, Al-Hilli F, Mohammad AM, Jamsheer NS, Romani KHR. Wolman's disease. The first two cases from Bahrain. Bahrain Medical Bulletin. 1999;21(1):24-6.

64. Anderson G, Burke D, Krywawych S, Leaky J. Dicarboxylic aciduria in Wolman's disease. Journal of inherited metabolic disease. 2010;33:S142.

65. Benlian P, Lacaille F, Mary S, Feillet F, Noel E, Eyer D, et al. Cholesteryl ester storage disease: a predominant lipa genotype underlies disease manifestations and progression - a review of 12 french cases. Atherosclerosis. 2014;235(2):e178-e9.

66. Besley GTN, Broadhead DM, Lawlor E. Acid esterase deficiency: comparison of biochemical findings in infantile and adult forms. Journal of inherited metabolic disease. 1984;7:155-6.

67. Byrd Iii JC, Powers JM. Wolman's disease: Ultrastructural evidence of lipid accumulation in central and peripheral nervous systems. Acta neuropathologica. 1979;45(1):37-42.

68. Cebolla JJ, Irun P, Gonzalez-Dieguez L, Del Valle Loarte P, Barba-Romero MA, Garcia-Jimenez I, et al. Dried blood spot screening of lysosomal acid lipase deficiency and confirmatory studies in Spanish suspected patients. Molecular genetics and metabolism. 2016;117(2):S33.

69. Erbe RW, Sims KB, Goss KA, Martin DR. An unremarkable clinical presentation of lysosomal acid lipase deficiency (LALD) in a 5 year old with advanced liver pathology. Molecular genetics and metabolism. 2014;111(2):S42.

70. Eto Y, Kitagawa T. Wolman's disease with hypolipoproteinemia and acanthocytosis: clinical and biochemical observations. The Journal of pediatrics. 1970;77(5):862-7.

71. Fang W, Lu Y. Clinical features of a novel LIPA gene mutation: A case report of Wolman disease. Hepatology International. 2017;11(1):S362.

72. Fulcher AS, Das Narla L, Hingsbergen EA. Pediatric case of the day. Wolman disease (primary familial xanthomatosis with involvement and clacification of the adrenal glands). Radiographics : a review publication of the Radiological Society of North America, Inc. 1998;18(2):533-5.

73. Fumić KF, Calandra SC, Verheijen FWV, Huljev Frković SHF, Vuković JV, Rajić LJR, et al. High plasma chitotriosidase activity in an infant with rapidly progressive wolman's disease and novel mutation in lipa gene. Journal of inherited metabolic disease. 2010;33:S130.

74. Gargus JJ, Verrell K, Wencel M, Bocian M, Kimonis V. Highly discordant phenotype in siblings with wolman syndrome and absent lal enzyme activity. Molecular genetics and metabolism. 2012;105(2):S29-S30.

75. Grabowski G. Therapy for lysosomal acid lipase deficiency: Replacing a missing link. Hepatology (Baltimore, Md). 2013;58(3):850-2.

76. Harrison RB, Francke Jr P. Radiographic findings in Wolman's disease. Radiology. 1977;124(1):188.

77. Hart CE, Wu HY, Sharrard M, Tylee K, Church H, Jones SA. Persistent finding of suberic acid, azelaic acid and pimelic acid in organic acid profiles from a patient subsequently diagnosed with Wolman's disease. Journal of inherited metabolic disease. 2016;39:S135.

78. Heiner-Fokkema MR, Niezen-Koning KE, Van Der Sluijs FJ, Bontekoe AB, Janssens-Puister J, De Koning TJ, et al. Urine dicarboxylic acids and other biomarkers for diagnosis and follow-up of Wolman disease. Journal of inherited metabolic disease. 2016;39:S207.

79. Hendriksz C, Wraith E, Dhawan A, Whitley C, Banikazemi M, Bialer M, et al. Natural history study of early onset lysosomal acid lipase (LAL) deficiency (wolman disease) confirms a severe and rapid disease course. Journal of inherited metabolic disease. 2012;35(1):S21.

80. Jones SA, Bernstein D, Bialer M, Dhawan A, Hendriksz C, Whitley CB, et al. Severe and rapid disease course in the natural history of infants with lysosomal acid lipase deficiency. Molecular genetics and metabolism. 2014;111(2):S57-S8.

81. Kamalian N, Dudley AW, Jr., Beroukhim F. Wolman disease with jaundice and subarachnoid hemorrhage. American journal of diseases of children (1960). 1973;126(5):671-5.

82. Klaudel-Dreszler M, Bakuła A, Orłowska E, Tylki-Szymańska A, Ługowska A, Zakharova EY, et al. Early onset LAL deficiency mimicking haemophagocytic lymphohistiocytosis. Journal of pediatric gastroenterology and nutrition. 2016;62:513.

83. Kodali S, Shoreibah MG, Young S, Singal AK. Lysosomal acid lipase deficiency (LAL-D). Gastroenterology. 2016;150(4):S342.

84. Leone L, Ippoliti PF, Antonicelli R. Use of simvastatin plus cholestyramine in the treatment of lysosomal acid lipase deficiency. The Journal of pediatrics. 1991;119(6):1008-9.

85. Leone L, Ippoliti PF, Antonicelli R, Balli F, Gridelli B. Treatment and liver transplantation for cholesterol ester storage disease. The Journal of pediatrics. 1995;127(3):509-10.

86. Lourenco C, Timm F, Burin M, Giugliani R, Hamilton J, Burg J, et al. “You can't always get what you want”: Filipin staining unmasking lysosomal acid lipase (LAL) deficiency in a young child. Molecular genetics and metabolism. 2013;108(2):S62.

87. Lourenco CM, Timm F, Burin M, Burg J, Quinn A, Hamilton J, et al. Abnormal filipin staining unmasking lysosomal acid lipase (LAL) deficiency: Not all that shines is Niemann-Pick type C. Journal of inherited metabolic disease. 2013;36(2):S299.

88. Marshall WC, Ockenden BG, Fosbrooke AS, Cumings JN. Wolman's disease. A rare lipidosis with adrenal calcification. Archives of disease in childhood. 1969;44(235):331-41.

89. Matthews RP, Haber BA, Mamula P, Piccoli DA. Cases in pediatric gastroenterology from The Children's Hospital of Philadelphia. A 5-year-old girl with massive hepatosplenomegaly, fever, and rash. MedGenMed : Medscape general medicine. 2004;6(2):14.

90. O'Leary OA, Hawkes CP, Cody D, Hughes JAF. Atypical wolman disease with evidence of immune dysfunction. Journal of inherited metabolic disease. 2013;36(2):S284.

91. Ozmen MN, Aygun N, Kilic I, Kuran L, Yalcin B, Besim A. Wolman's disease: ultrasonographic and computed tomographic findings. Pediatric radiology. 1992;22(7):541-2.

92. Patel K, Hodock K. Wolman disease and HLH, a case of overlapping clinical features. Pediatric Blood and Cancer. 2018;65:S19-S20.

93. Perezpeña Diazconti M, Serrano Bello C, Herrera Segura M, Consuelo Sanchez A, Valencia Mayoral P. Distinctive histopathological findings in liver biopsies from 10 children with lysosomal acid lipase deficiency. Virchows Archiv. 2016;469:S18.

94. Queloz JM, Capitanio MA, Kirkpatrick JA. Wolman's disease. Roentgen observations in 3 siblings. Radiology. 1972;104(2):357-9.

95. Raafat F, Hashemian MP, Abrishami MA. Wolman's disease: report of two new cases, with a review of the literature. American journal of clinical pathology. 1973;59(4):490-7.

96. Rennie C, Johnstone N, Cochrane B, McMahon S, Cozens A. Successful dietary management of infantile onset lysosomal acid li-pase deficiency (LAL-D). Journal of inherited metabolic disease. 2018;41:S87-S8.

97. Slae M, Ghosh A, Arvonen M, Fecarotta S, Gargus JJ, Gokcay G, et al. Experience of the nutritional management of infantile onset lysosomal acid lipase deficiency (LAL-D). Journal of pediatric gastroenterology and nutrition. 2018;66:928.

98. Strokova T, Bagaeva M, Tumanova E, Grishina A, Kamenets E, Zakharova E. Lysosomal acid lipase deficiency (LAL D): Clinical and morphological progression in children and young patients. Journal of Inborn Errors of Metabolism and Screening. 2017;5:338-9.

99. Uno Y, Taniguchi A, Tanaka E. Histochemical studies in Wolman's disease--report of an autopsy case accompanied with a large amount of milky ascites. Acta pathologica japonica. 1973;23(4):779-90.

100. Van Spronsen FJ, Derks TGJ, De Koning TJ, Van Rijn M, Niezen-Koning KE, Heiner-Fokkema MR, et al. Lysosomal acid lipase deficiency (LAL-D;Wolman disease): Diagnosed at 2 months needing 3 months to really improve on Sebelipase alpha. Journal of inherited metabolic disease. 2016;39:S219.

101. Watanuki T, Nakamura K, Koizumi K, Konno T, Fujii M. [Wolman's disease]. Saishin igaku Modern medicine. 1967;22(2):302-14.

102. Wong D, Cederbaum S, Enns G, Wraith E, Whitley C, C.J H, et al. The natural history of wolman disease, the early onset form of lysosomal acid lipase (LAL) deficiency, confirms a severe and rapid disease course. Pediatric Critical Care Medicine. 2012;13(6):716.

103. Yahyaoui R, Rodríguez-García E, Gil-Serret M, García-Jiménez MC, González-Diéguez ML, Del Valle Loarte P, et al. Characterization of plasma lipoprotein particles in Spanish patients with lysosomal acid lipase deficiency. Journal of Inborn Errors of Metabolism and Screening. 2017;5:340-1.

104. Bernstein DL, Lobritto S, Iuga A, Remotti H, Schiano T, Fiel MI, et al. Lysosomal acid lipase deficiency allograft recurrence and liver failure- clinical outcomes of 18 liver transplantation patients. Molecular genetics and metabolism. 2018;124(1):11-9.

105. Eminoglu FT, Bolkent MG, Kuloglu Z, Kansu A. Case presentation: A girl with cholesterol ester storage disease. Molecular genetics and metabolism. 2016;117(2):S45.

106. Eto Y, Sekiguchi S. [Wolman's disease]. Nihon rinsho Japanese journal of clinical medicine. 1978:1386-7.

107. Guardamagna O, Cagliero P, Zimetti F, Brunatti P, Favari E. Lysosomal acid lipase deficiency in children: Clinical and biochemical presentation. Atherosclerosis. 2014;235(2):e71.

108. Huang YL, Sheng HY, Zhao XY, Yu JK, Li L, Liu HS, et al. [Wolman disease with novel mutation of LIPA gene in a Chinese infant]. Zhonghua er ke za zhi = Chinese journal of pediatrics. 2012;50(8):601-5.

109. Konno T. [Wolman disease]. Ryoikibetsu shokogun shirizu. 1993(1):397-9.

110. Lavrova AE, Konovalova EY, Davydova DA, Shelyakhin VE, Lobanova EV. Deficiency of lysosomal acid lipase in a 5 years old child. Pediatriya - Zhurnal im GN Speranskogo. 2017;96(6):183-6.

111. Ma YY, Chen L. [Wolman disease: report of a case]. Zhonghua bing li xue za zhi = Chinese journal of pathology. 2013;42(4):276-7.

112. Quinn AG, Burton B, Deegan P, Di Rocco M, Enns G, Guardamagna O, et al. Sustained elevations in LDL cholesterol and serum transaminases from early childhood are common in lysosomal acid lipase deficiency. Molecular genetics and metabolism. 2014;111(2):S89.

113. Surkov AN, Anushenko AO, Movsisyan GB, Potapov AS, Kulebina EA, Bessonov EE, et al. The first Russia experience with the use of enzyme replacement therapy with sebelipase alpha in a child with an infantile form of lysosomal acid lipase deficiency. Pediatriya - Zhurnal im GN Speranskogo. 2020;99(1):226-32.

114. Tanaka A. Acid lipase deficiency (Wolman disease and cholesteryl ester storage disease: CESD). Ryoikibetsu shokogun shirizu. 1998(19):427-30.

115. Tanaka A. Wolman disease. Nippon rinsho Japanese journal of clinical medicine. 2006:750-2.

116. Lageron A. Histoenzymology of cholesterol ester storage disease. A propos of 5 cases. Medecine et Chirurgie Digestives. 1978;7(2):155-9.

117. Amraoui Y, Mengel E, Gerner P, Baba HA, Beck M. Cholesteryl ester storage disease. Rare disease or frequently underdiagnosed? Tagliche Praxis. 2015;56(1):55-61.

118. Aubert-Tulkens G, Van Hoof F. Acid lipase deficiency: Clinical and biochemical heterogeneity. Acta paediatrica Belgica. 1980;32(4):239-45.

119. Bambirra EA, Tafuri WL, Borges HH, Carvalho C, Naves IL, Bogliolo L, et al. Wolman's disease: a clinicopathologic, electron microscopic, and histochemical study. Southern medical journal. 1982;75(5):595-6.

120. Bernstein D, Desnick R. Cholesteryl ester storage disease (CESD): An under-recognized and treatable LSD with liver dysfunction/failure and dyslipidemia. Molecular genetics and metabolism. 2013;108(2):S24.

121. Bona G, Bracco G, Gallina MR, Iavarone A, Artesani L, Perona A, et al. A case of acid lipase deficiency: Wolman's disease. Panminerva medica. 1989;31(1):49-53.

122. Bretagne MC, Rivoal-Jeanjean E, Beley G, Vidailhet M, Treheux A. [Wolman's disease. A rare cause of adrenal calcification in infants: a case report (author's transl)]. Journal de radiologie. 1981;62(3):197-9.

123. Caseiro C, Silva E, Ribeiro I, Laranjeira F, Pinto E, Oliva T, et al. Wolman disease-a misdiagnosis case with hemophagocytic lymphohistiocytosis. Journal of inherited metabolic disease. 2016;39:S204-S5.

124. Castro M, Rosati P, Boldrini R, Lucidi V, Gambarara M, Bosman C. Wolman's disease diagnosed by intestinal biopsy. Italian journal of gastroenterology and hepatology. 1999;31(7):610-2.

125. Coelho CAR, Balarin MAS, Coelho KIR. Cholesterylester storage disease: report of a case. Arq gastroenterol. 1987;24(3):184-7.

126. Contreras F, Alvarez I, Nistal M, Claver M. Wolman's disease. PATOLOGIA. 1974;7(2):189-200.

127. Elleder M, Poupetova H, Ledvinova J, Hyanek J, Zeman J, Sykora J, et al. [Lysosomal acid lipase deficiency. Overview of Czech patients]. Casopis lekaru ceskych. 1999;138(23):719-24.

128. Elsayed S, Elsobky E, Tantawy A, Ragab I, Grandin V, Gil M, et al. Wolman disease presenting as familial hemophagocytic lymophohistocytosis. Haematologica. 2014;99:627.

129. Feillet F, Salmon A, Clement L, Levade T, Digeon B, Bednarek N, et al. Successful cord blood transplant in a child with wolman's disease (WD). Molecular genetics and metabolism. 2009;98(1):72.

130. Fernandez-Aragon M, Cervantes-Bustamante R, De Leon-Bojorge B, Zarate-Mondragon F, Mata-Rivera N, Barrios EM, et al. [Cholesterol ester storage disease]. Revista de gastroenterologia de Mexico. 2004;69(3):171-5.

131. Foster D, Goutet JM, Sirinelli D. [Iconographic rubric. Wolman's disease]. Archives francaises de pediatrie. 1987;44(7):521-2.

132. Gautier M, Lapous D, Raulin J. [Cholesterol ester storage disease in children. Comparative biochemistry of hepatocyte and fibroblast cultures]. Archives francaises de pediatrie. 1978;35(10):38-49.

133. Guazzi GC, Martin JJ, Philippart M, Roels H, Hooft C, Van der Eecken H, et al. Wolman's disease. Distribution and significance of the central nervous system lesions. Pathologia Europaea. 1968;3(2):266-77.

134. Guazzi GC, Martin JJ, Philippart M, Roels H, van der Eecken H, Vrints L, et al. Wolman's disease. European neurology. 1968;1(6):334-62.

135. Hanak J, Elleder M. [Cholesterol ester storage disease (CESD)]. Ceskoslovenska pediatrie. 1984;39(12):721-5.

136. Haznedar P, Kuloglu Z, Kansu A, Eminoglu FT. A rare cause of hepatomegaly in the childhood: Lysosomal acid lipase deficiency. The Turkish journal of gastroenterology : the official journal of Turkish Society of Gastroenterology. 2018;29(4):518-9.

137. Hicks J, Wartchow E, Mierau G. Cholesterol ester storage disease: Role of electron microscopy in diagnosis and guiding molecular confirmation. Laboratory Investigation. 2014;94:534A.

138. Higuchi Y, Mori R, Kamitamari A, Kioshita E, Baba T, Matsumoto T, et al. A case of mild type wolman disease. Japanese Journal of Human Genetics. 1997;42(1):105.

139. Hill SC, Hoeg JM, Dwyer AJ, Vucich JJ, Doppman JL. CT findings in acid lipase deficiency: wolman disease and cholesteryl ester storage disease. Journal of computer assisted tomography. 1983;7(5):815-8.

140. Ho FC, Lin HJ, Chan WC. Wolman's disease: the first reported Chinese patient. Modern medicine of Asia. 1978;14(11):23-6.

141. Justus J, Schulze J, Kemmer C, Riedel H. [Liver morphology and clinical aspects of a case of cholesterol ester storage disease]. Deutsche Zeitschrift fur Verdauungs- und Stoffwechselkrankheiten. 1988;48(3):198-207.

142. Kahana D, Berant M, Wolman M. Primary familial xanthomatosis with adrenal involvement (Wolman's disease). Report of a further case with nervous system involvement and pathogenetic considerations. Pediatrics. 1968;42(1):70-6.

143. Keller E, Kunnert B, Braun W. [Cholesterol-ester storage disease of the liver in childhood]. Deutsche Zeitschrift fur Verdauungs- und Stoffwechselkrankheiten. 1977;37(5):231-6.

144. Krebs A, Aslanidis C, März W, Krebs K, Doerfer J, Greiner P, et al. Hepato(spleno)megaly and hypercholesterolemia. Cardinal symptoms of cholesterol ester storage disease. Padiatrische Praxis. 2010;75(4):601-11.

145. Kunnert B. Liver biopsy for diagnosis of cholesterol ester storage disease with histochemical and histochromatographic methods. Acta Histochemica. 1977;59:277-80.

146. Kunnert B, Pohlandt K, Ruschke I, Keller E. [Cholesterol ester storage disease and sea-blue histiocytes]. Zentralblatt fur allgemeine Pathologie u pathologische Anatomie. 1987;133(6):517-25.

147. Kuntz HD, May B, Schejbal V, Assmann G. [Cholesteryl ester storage disease in the liver (author's transl)]. Leber, Magen, Darm. 1981;11(6):258-63.

148. Lageron A, Gautier M, Scotto J. [Clinical and histoenzymological peculiarities of cholesterol storage in 2 children of the same family]. Archives francaises de pediatrie. 1985;42:605-11.

149. Lajo A, Gracia R, Navarro M. Wolman's disease. Acute infantile form. Anales espanoles de pediatria. 1974;7(5):438-46.

150. Leclerc JL, Hould F, Lelievre M, Gagne F. [Wolman's disease: anatomo-clinical study of a recent case with absence of radiologic and macroscopic adrenal calcifications]. Laval medical. 1971;42(5):461-7.

151. Loffredo L, De Filippo P, Magri B. [A case of Wolman's disease]. La Pediatria. 1975;83(1):155-64.

152. Lough J, Fawcett J, Wiegensberg B. Wolman's disease. An electron microscopic, histochemical, and biochemical study. Archives of pathology. 1970;89(2):103-10.

153. Marosvari I. Wolman disease in twins. Acta paediatrica Hungarica. 1985;26(1):61-4.

154. Martinez De Compañón Z, Trenchs V, Alsina L, Mainou C, Caritg J. Evaluation of diagnostic criteria for cholestatic jaundice during the first year of life. Pediatria Catalana. 2004;64(2):56-60.

155. Mazurová S, Poupětová H, Hůlková H, ͖oukálková L, Urbanová Z, Zeman J, et al. Cholesteryl Ester Storage Disease (CESD): Clinical, laboratory and histological characteristics in 6 patients. Cesko-Slovenska Pediatrie. 2014;69(3):148-60.

156. McKiernan PJ, Hodges S, Sharif K, Santra S. Liver transplantation for cholesterol ester storage disease: Outcome at five years. Journal of inherited metabolic disease. 2012;35(1):S21.

157. Miller R, Bialer MG, Rogers JF, Jonsson HT, Jr., Allen RV, Hennigar GR. Wolman's disease: report of a case, with multiple studies. Archives of pathology & laboratory medicine. 1982;106(1):41-4.

158. Muñoz Bartolo G, González De Zarate A, Frauca E, Martín L, Hierro L, Camarena C, et al. Cholesterol ester storage disease, an entity to consider in the differential diagnosis of fatty liver. Pediatrika. 2007;27(4):22.

159. Murakami J, Kuranobu N, Fukushima K, Nishimura R, Amano K, Eto Y, et al. Enzyme replacement therapy for lysosomal acid lipase deficiency: A report of two Japanese patients. Journal of inherited metabolic disease. 2016;39:S212.

160. Nardi F, Borri P. [Morphological and chemical characterization of a case of Wolman's disease. Preliminary note]. Acta neurologica. 1971;26(3):270-8.

161. Neustein HB, O'Brien JS, Rosser RJ, Fillerup DL. Chronic nephritis and renal foam cells. Cholesterol ester storage. Archives of pathology. 1972;93(6):503-9.

162. Ozoran Y, Ozoran A, Kerse I, Gurgey A, Ozsoylu S, Kocak N, et al. An ultrastructural study in a case of Wolman's disease (clinical, biochemical, light and electron microscopic study). The Turkish journal of pediatrics. 1978;20(3):100-7.

163. Ozsoylu S. Therapeutic trial in Wolman disease. The Turkish journal of pediatrics. 1992;34(4):267-8.

164. Ozsoylu S, Gurgey A, Kocak N, Ozoran Y, Ozoran A, Kerse I, et al. Wolman's disease. A case report with lipid, chromosome and electronmicroscopic studies. The Turkish journal of pediatrics. 1977;19(1):57-66.

165. Parada Yáñez J, Blanco Docanto M, Reparaz Vidal R, Gallego Pastoriza M, Fontaiña Martínez M, Gómez Pérez I, et al. Wolman's sickness: A case report. Anales espanoles de pediatria. 1996;45(4):428-30.

166. Pastor Bevia E, Pedraz Garcia C, Heras de Pedro MI, Garcia Parron A, Escudero Bueno G, Benito Zaballos MF, et al. [Wolman's disease: report of 3 cases]. Anales espanoles de pediatria. 1987;26(4):301-4.

167. Perez Rodriguez-Cuesta JM, Suarez Tomas JI, Suarez Mene ndez ME, Dominguez Gonzalez J. Cholesterol ester storage disease in two brothers. Anales espanoles de pediatria. 1990;32(3):249-52.

168. Permanetter W, Muller-Hocker J, Hubner G, Schaub J. [Wolman's disease (Wolman's syndrome)]. Die Medizinische Welt. 1979;30(47):1783-9.

169. Pisciotta L, Fresa R, Michelakakis H, Lycopoulou L, Calandra S, Bertolini S. Severe lysosomal ACID lipase deficiency in a greek child homozygous for a novel splicing mutation of the lipa gene. European Journal of Clinical Investigation. 2017;47:95.

170. Pohlandt K, Ruschke I, Kunnert B. Cholesterol Ester Storage Disease in Childhood - a Case-Report. Zeitschrift Fur Klinische Medizin-Zkm. 1988;43(15):1325-9.

171. Pot DJ, Kardux JJ. [Diagnostic image (358). A neonate with splenomegaly and calcified adrenal glands]. Nederlands tijdschrift voor geneeskunde. 2008;152(4):202.

172. Quirici Neto F, Silva LH, Silva FC, Uemura L, Lederman HM. Caso radiológico do mês: doença de Wolman. Rev paul pediatr. 1985;3(9):40-1.

173. Rassoul F, Richter V, Lohse P, Naumann A, Purschwitz K, Keller E. Long-term administration of the HMG-CoA reductase inhibitor lovastatin in two patients with cholesteryl ester storage disease. International journal of clinical pharmacology and therapeutics. 2001;39(5):199-204.

174. Ryzko J, Celinska-Cedro D, Karpinska D, Tylki-Szymanska A. [A case of Wolman's disease in a two-month old boy]. Wiadomosci lekarskie (Warsaw, Poland : 1960). 1993;46(3):147-50.

175. Smolka V. Cholesteryl ester storage disease - The effect of long-term treatment with lovastatin. Ceska a Slovenska Gastroenterologie a Hepatologie. 2007;61(1):30-3.

176. Storm W, Wendel U, Sprenkamp M, Seidler A. [Wolman's disease in an infant]. Monatsschrift Kinderheilkunde : Organ der Deutschen Gesellschaft fur Kinderheilkunde. 1990;138(2):88-90.

177. Sundaravalli N, Bhaskar Raju B, Prema A, Pushpa V, Nedunchelian N, Shetty MV, et al. Wolman's disease. Indian pediatrics. 1986;23(11):950-3.

178. Tashiro Y, Watanabe Y. An electron microscopic observation on acid lipase deficiency: Cholecteryl ester storage disease and Wolman's disease. Journal of Clinical Electron Microscopy. 1989;22(5):588.

179. Thavarungkul P, Hemsrichart V, Supradish P. Cholesterol ester storage disease: a reported case. Journal of the Medical Association of Thailand = Chotmaihet thangphaet. 1995;78(3):164-8.

180. Tonissen R, Kuntz HD, May B. [Morphology and differential diagnosis of cholesterol ester storage disease]. Die Medizinische Welt. 1983;34(24):704-6.

181. Tylki-Szymanska A, Maciejko D, Wozniewicz B, Muszynska B. Two cases of cholesteryl ester storage disease (CESD) acid lipase deficiency. Hepato-gastroenterology. 1987;34(3):98-9.

182. Valerdiz-Casasola S, Velasco-Garcia R, Rodriguez-Rodriguez R. [Wolman's disease. Pulmonary storage of lipids]. Anales espanoles de pediatria. 1997;47(4):427-8.

183. Vargas Torcal F, Gomez Garcia A, Cuevas J, Young EP. [Wolman's disease]. Anales espanoles de pediatria. 1987;27(3):195-8.

184. Vij M, Bachina P. Liver histology in cholesteryl ester storage disease. Indian journal of pathology & microbiology. 2018;61(2):302-4.

185. Wallis K, Gross M, Kohn R, Zaidman J. A case of Wolman's disease. Helvetica paediatrica acta. 1971;26(1):98-111.

186. Weiler C, Freudenberg F, Müller-Höcker J. Cholesterol ester storage disease : AA rare disease or a rare diagnosis? Der Pathologe. 2009;30(1):65-9.

187. Wolf H, Hug G, Michaelis R, Nolte K. [Unusual congenital cholesterol ester storage in the liver]. Helvetica paediatrica acta. 1974;29(2):105-18.

188. Wolman EM. Wolman disease: suggestions for effective treatment. Indian pediatrics. 1992;29(7):938.

189. Wolman M. [To pediatricians who care for infants with Wolman's disease. Advice for an efficacious treatment]. Minerva pediatrica. 1993;45(3):131.

190. Yadav TP. Wolman's disease. Indian pediatrics. 1995;32(12):1324.

191. Zlatkovic M, Stankovic I, Prokic D, Plamenac P. [Pathohistologic diagnosis of cholesterol ester storage disease]. Srpski arhiv za celokupno lekarstvo. 2001;129(7):207-10.

192. Zubov NA, Zakharova S, Vibe AB. [Wolman disease in children in the same family]. Pediatriia. 1989(5):90-1.

193. Basu D, Ferns S, Prasad MA, Nalini P. Failure to thrive in a 3 month old boy. Postgraduate medical journal. 2002;78(923):567, 9.

194. Leavitt JA, Kotagal S. The "Cherry Red" Spot. Pediatric Neurology. 2007;37(1):74-5.

195. Philippart M, Durand P, Borrone C. Neutral lipid storage with acid lipase deficiency: a new variant of Wolman's disease with features of the Senior syndrome. Pediatric research. 1982;16(11):954-9.

196. Weiskirchen R. Fast progression of liver damage in lysosomal acid lipase deficiency. Current medical research and opinion. 2017;33(11):2081-3.

197. al-Essa MA, Bakheet SM, Patay ZJ, Powe JE, Ozand PT. Normal fluorine-18-labelled 2-fluoro-2-deoxyglucose positron emission tomography and magnetic resonance imaging of the brain in Wolman disease. Journal of inherited metabolic disease. 1999;22(7):846-8.

198. Arterburn JN, Lee WM, Wood RP, Shaw BW, Markin RS. Orthotopic liver transplantation for cholesteryl ester storage disease. Journal of clinical gastroenterology. 1991;13(4):482-5.

199. Jones SA, Valayannopoulos V, Schneider E, Eckert S, Banikazemi M, Bialer M, et al. Rapid progression and mortality of lysosomal acid lipase deficiency presenting in infants. Genetics in medicine : official journal of the American College of Medical Genetics. 2016;18(5):452-8.

200. Kucukcongar Yavas A, Genc¸ P, Kilic¸ N, Erdoʇan H, Özdemir Ö, Ekici A, et al. Secondary hemophagocytosis in a patient with wolman disease. Journal of inherited metabolic disease. 2015;38(1):S277.

201. Lipiński P, Ługowska A, Zakharova EY, Socha P, Tylki-Szymańska A. Clinical picture and biochemical findings in the course of late onset lysosomal acid lipase deficiency in Polish patients. Journal of pediatric gastroenterology and nutrition. 2018;66:762.

202. Lyons H, Vouyoukas E, Higgins M, Maciejko JJ. Clinical and Histologic Liver Improvement in Siblings With Lysosomal Acid Lipase Deficiency After Enzyme Replacement. Journal of pediatric gastroenterology and nutrition. 2020;70(5):635-9.