Hyland K. and Clayton P. T. (1990) Aromatic amino acid decarboxylase deficiency in twins. Journal of Inherited Metabolic Disease 13, 301-304.

Hyland K., Surtees R. A. H., Rodeck C. and Clayton P. T. (1992) Aromatic L-Amino-Acid Decarboxylase Deficiency - Clinical-Features, Diagnosis, and Treatment of A New Inborn Error of Neurotransmitter Amine Synthesis. Neurology 42, 1980-1988.

Hyland K. and Clayton P. T. (1992) Aromatic L-amino acid decarboxylase deficiency: diagnostic methodology. Clinincal Chemistry 38, 2405-2410. Free full text available

Korenke G. C., Christen H. J., Hyland K., Hunneman D. H. and Hanefeld F. (1997) Aromatic L-amino acid decarboxylase deficiency: an extrapyramidal movement disorder with oculogyric crises. European Journal of Paediatric Neurology 1, 67-71.

Maller A., Hyland K., Milstien S., Biaggioni I. and Butler I. J. (1997) Aromatic L-amino acid decarboxylase deficiency: Clinical features, diagnosis, and treatment of a second family. Journal of Child Neurology 12, 349-354.

Abeling N. G. G. M., van Gennip A. H., Barth P. G., van Cruchten A., Westra M. and Wijburg F. A. (1998) Aromatic L-amino acid decarboxylase deficiency: A new case with a mild clinical presentation and unexpected laboratory findings. Journal of Inherited Metabolic Disease 21, 240-242.

Lamers K. J. B. and Wevers R. A. (1998) Abnormalities of biogenic amines affecting the metabolism of serotonin and catecholamines. Multiple Sclerosis 4, 37-38.

Swoboda K. J., Hyland K., Goldstein D. S., Kuban K. C. K., Arnold L. A., Holmes C. S. and Levy H. L. (1999) Clinical and therapeutic observations in aromatic L-amino acid decarboxylase deficiency.Neurology 53, 1205-1211.

Abeling N. G. G. M., Brautigam C., Hoffmann G. F., Barth P. G., Wevers R. A., Jaeken J., Fiumara A., Knust A. and van Gennip A. H. (2000) Pathobiochemical implications of hyperdopaminuria in patients with aromatic L-amino acid decarboxylase deficiency. Journal of Inherited Metabolic Disease 23, 325-328.

Brautigam C., Wevers R. A., Hyland K., Sharma R. K., Knust A. and Hoffmann G. F. (2000) The influence of L-dopa on methylation capacity in aromatic L-amino acid decarboxylase deficiency: Biochemical findings in two patients. Journal of Inherited Metabolic Disease 23, 321-324.

Brautigam C., Hyland K., Wevers R., Sharma R., Wagner L., Stock G. J., Heitmann F. and Hoffmann G. F. (2002) Clinical and laboratory findings in twins with neonatal epileptic encephalopathy mimicking aromatic L-amino acid decarboxylase deficiency. Neuropediatrics 33, 113-117.

Fiumara A., Brautigam C., Hyland K., Sharma R., Lagae L., Stoltenborg B., Hoffmann G. F., Jaeken J. and Wevers R. A. (2002) Aromatic L-amino acid decarboxylase deficiency with hyperdopaminuria. Clinical and laboratory findings in response to different therapies. Neuropediatrics 33, 203-208.

Swoboda K. J., Saul J. P., McKenna C. E., Speller N. B. and Hyland K. (2003) Aromatic L-amino acid decarboxylase deficiency: overview of clinical features and outcomes. Annals of Neurology 54 Suppl 6, S49-S55.

Chang Y. T., Sharma R., Marsh J. L., McPherson J. D., Bedell J. A., Knust A., Brautigam C., Hoffmann G. F. and Hyland K. (2004) Levodopa-responsive aromatic L-amino acid decarboxylase deficiency. Annals of Neurology 55, 435-438.

Pearl P. L., Wallis D. D. and Gibson K. M. (2004) Pediatric neurotransmitter diseases. Current Neurology and Neuroscience Reports 4, 147-152.

Pons R., Ford B., Chiriboga C. A., Clayton P. T., Hinton V., Hyland K., Sharma R. and De Vivo D. C. (2004) Aromatic L-amino acid decarboxylase deficiency: Clinical features, treatment, and prognosis. Neurology 62, 1058-1065.

Hsieh H. J., Lin S. H. and Liu H. M. (2005) Visualisation of impaired dopamine biosynthesis in a case of aromatic L-amino acid decarboxylase deficiency by co-registered F-18-FDOPA PET and magnetic resonance imaging. European Journal of Nuclear Medicine and Molecular Imaging 32, 517.

Abdenur J. E., Abeling N. A., Specola N., Jorge L., Schenone A. B., van Cruchten A. C. and Chamoles N. A. (2006) Aromatic L-aminoacid decarboxylase deficiency: Unusual neonatal presentation and additional findings in organic acid analysis. Molecular Genetics and Metabolism 87, 48-53.

Anselm I. A. and Darras B. T. (2006) Catecholamine toxicity in aromatic L-amino acid decarboxylase deficiency. Pediatric Neurology 35, 142-144.

Berkowitz D. H. and Ganesh A. (2006) Combined general and regional anesthetic in a child with aromatic L-amino acid decarboxylase deficiency. Anesthesia and Analgesia 103,1630-1631. Free full text available

Vutskits L., Menache C., Manzano S., Haenggeli C. A. and Habre W. (2006) Anesthesia management in a young child with aromatic l-amino acid decarboxylase deficiency. Paediatric Anaesthesia 16, 82-84.

Hyland K. (2007) Inherited disorders affecting dopamine and serotonin: Critical neurotransmitters derived from aromatic amino acids. Journal of Nutrition 137, 1568S-1572S. Free full text available

Pearl P. L., Taylor J. L., Trzcinski S. and Sokohl A. (2007) The pediatric neurotransmitter disorders. Journal of Child Neurology 22, 606-616.

Tay S. K. H., Poh K. S., Hyland K., Pang Y. W., Ong H. T., Low P. S. and Goh D. L. M. (2007) Unusually mild phenotype of AADC deficiency in 2 siblings. Molecular Genetics and Metabolism 91, 374-378.

Verbeek M. M., Geurtz P. B. H., Willemsen M. A. A. P. and Wevers R. A. (2007) Aromatic L-amino acid decarboxylase enzyme activity in deficient patients and heterozygotes. Molecular Genetics and Metabolism 90, 363-369.

Haavik J., Blau N. and Thony B. (2008) Mutations in human monoamine-related neurotransmitter pathway genes. Human Mutation 29, 891-902.

Ito S., Nakayama T., Ide S., Ito Y., Oguni H., Goto Y. and Osawa M. (2008) Aromatic L-amino acid decarboxylase deficiency associated with epilepsy mimicking non-epileptic involuntary movements. Developmental Medicine and Child Neurology 50, 876-878.

Allen G. F. G., Land J. M. and Heales S. J. R. (2009) A new perspective on the treatment of aromatic L-amino acid decarboxylase deficiency. Molecular Genetics and Metabolism 97, 6-14.

Lee H. F., Tsai C. R., Chi C. S., Chang T. M. and Lee H. J. (2009) Aromatic L-amino acid decarboxylase deficiency in Taiwan. European Journal of Paediatric Neurology 13, 135-140.

Lee W. T., Weng W. C. Peng S. F. and Tzen K. Y. (2009) Neuroimaging findings in children with paediatric neurotransmitter diseases. Journal of Inherited Metabolic Disease 32, 361-370.

Manegold C., Hoffmann G. F., Degen I., Ikonomidou H., Knust A., Laass M. W., Pritsch M., Wilichowski E. and Horster F. (2009) Aromatic L-amino acid decarboxylase deficiency: clinical features, drug therapy and follow-up. Journal of Inherited Metabolic Disease 32,371-380.

Allen G. F. G., Neergheen V., Oppenheim M., Fitzgerald J. C., Footitt E., Hyland K., Clayton P. T., Land J. M. and Heales S. J. R. (2010) Pyridoxal 5'-phosphate deficiency causes a loss of aromatic l-amino acid decarboxylase in patients and human neuroblastoma cells, implications for aromatic l-amino acid decarboxylase and vitamin B-6 deficiency states. Journal of Neurochemistry 114, 87-96.

Brun L., Ngu L. H., Keng W. T., Ch'Ng G. S., Choy Y. S., Hwu W. L., Lee W. T., Willemsen M. A. A. P., Verbeek M. M., Wassenberg T., Regal L., Orcesi S., Tonduti D., Accorsi P., Testard H., Abdenur J. E., Tay S., Allen G. F., Heales S., Kern I., Kato M., Burlina A., Manegold C., Hoffmann G. F. and Blau N. (2010) Clinical and biochemical features of aromatic L-amino acid decarboxylase deficiency. Neurology 75, 64-71.

Ide S., Sasaki M., Kato M., Shiihara T., Kinoshita S., Takahashi J. and Goto Y. (2010) Abnormal glucose metabolism in aromatic L-amino acid decarboxylase deficiency. Brain & Development 32, 506-510.

Lee H. H. C., Lai C. K., Siu T. S., Chan K. Y., Yau E. K. C., Mak C. M., Yuen Y. P., Chan A. Y. W., Tam S. and Lam C. W. (2010) Modified clinical protocol for the investigations of dystonia: non-invasive biochemical diagnosis of aromatic L-amino acid decarboxylase deficiency in Chinese. Clinica Chimica Acta 411, 908-909.

Wassenberg T., Willemsen M. A. A. P., Geurtz P. B. H., Lammens M., Verrijp K., Wilmer M., Lee W. T., Wevers R. A. and Verbeek M. M. (2010) Urinary dopamine in aromatic L-amino acid decarboxylase deficiency: The unsolved paradox. Molecular Genetics and Metabolism 101, 349-356.

Wojtowicz P., Zrostlikova J., Kovalczuk T., Schurek J. and Adam T. (2010) Evaluation of comprehensive two-dimensional gas chromatography coupled to time-of-flight mass spectrometry for the diagnosis of inherited metabolic disorders using an automated data processing strategy. Journal of Chromatography A 1217, 8054-8061.

Kuo, S.-J., Ma, G.-C., Chang, S.-P., Wu, H.-H., Chen, C.-P., Chang, T.-M. and Chen, M. (2011) Preimplantation and prenatal genetic diagnosis of aromatic L-amino acid decarboxylase deficiency with an amplification refractory mutation system-quantitative polymerase chain reaction. Taiwanese Journal of Obstetrics & Gynecology 50, 468–73. Free full text available

Montioli, R., Cellini, B. and Borri Voltattorni, C. (2011) Molecular insights into the pathogenicity of variants associated with the aromatic amino acid decarboxylase deficiency. Journal of Inherited Metabolic Disease 34, 1213–24.

Hwu, W.-L., Muramatsu, S., Tseng, S.-H., Tzen, K.-Y., Lee, N.-C., Chien, Y.-H. and Wu, R.-M. (2012) Gene therapy for aromatic L-amino acid decarboxylase deficiency. Science Translational Medicine 4, 134ra61.

Lee, H.-C. H., Lai, C.-K., Yau, K.-C. E., Siu, T.-S., Mak, C. M., Yuen, Y.-P. and Chan, A. Y.-W. (2012) Non-invasive urinary screening for aromatic L-amino acid decarboxylase deficiency in high-prevalence areas: a pilot study. Clinica Chimica Acta 413, 126–30.

Wassenberg, T., Monnens, L. A. H., Geurtz, B. P. B. H., Wevers, R. A., Verbeek, M. M. and Willemsen, M. A. A. P. (2012) The paradox of hyperdopaminuria in aromatic L-amino acid deficiency explained. JIMD Reports 4, 39–45. Free full text available

Zwagerman, N. T. and Richardson, R. M. (2012) Gene therapy for aromatic L-amino acid decarboxylase deficiency. Neurosurgery 71(4), N10–2.

Allen G. F. G., Ullah Y., Hargreaves I. P., Land J. M. and Heales S. J. R. (2013) Dopamine but not L-dopa stimulates neural glutathione metabolism. Potential implications for Parkinson's and other dopamine deficiency states. Neurochemistry International 62, 684-694.

Arnoux, J.-B., Damaj, L., Napuri, S., Serre, V., Hubert, L., Cadoudal, M. and de Lonlay, P. (2013) Aromatic L-amino acid decarboxylase deficiency is a cause of long-fasting hypoglycemia. The Journal of Clinical Endocrinology and Metabolism 98, 4279–84. Free full text available

Lee, N.-C., Shieh, Y.-D., Chien, Y.-H., Tzen, K.-Y., Yu, I.-S., Chen, P.-W. and Hwu, W.-L. (2013) Regulation of the dopaminergic system in a murine model of aromatic L-amino acid decarboxylase deficiency. Neurobiology of Disease 52, 177–90.

Mastrangelo, M., Caputi, C., Galosi, S., Giannini, M. T. and Leuzzi, V. (2013) Transdermal rotigotine in the treatment of aromatic L-amino acid decarboxylase deficiency. Movement Disorders 28, 556–7.

Montioli, R., Oppici, E., Cellini, B., Roncador, A., Dindo, M. and Voltattorni, C. B. (2013) S250F variant associated with aromatic amino acid decarboxylase deficiency: molecular defects and intracellular rescue by pyridoxine. Human Molecular Genetics 22, 1615–24. Free full text available

Pearl, P. L. (2013) Monoamine neurotransmitter deficiencies. Handbook of Clinical Neurology 113, 1819–25.

Shih, D.-F., Hsiao, C.-D., Min, M.-Y., Lai, W.-S., Yang, C.-W., Lee, W.-T. and Lee, S.-J. (2013) Aromatic L-amino acid decarboxylase (AADC) is crucial for brain development and motor functions. PloS One 8(8), e71741.

Alfadhel, M., and Kattan, R. (2014) Aromatic amino acid decarboxylase deficiency not responding to pyridoxine and bromocriptine therapy: case report and review of response to treatment. Journal of Central Nervous System Disease 6, 1–5. Free full text available

Bertoldi, M. (2014) Mammalian dopa decarboxylase: structure, catalytic activity and inhibition. Archives of Biochemistry and Biophysics 546, 1–7.

Chen, H.-F., Chang, S.-P., Wu, S.-H., Lin, W.-H., Lee, Y.-C., Ni, Y.-H. and Chen, M. (2014) Validating a rapid, real-time, PCR-based direct mutation detection assay for preimplantation genetic diagnosis. Gene 548, 299–305.

Chen, P.-W., Lee, N.-C., Chien, Y.-H., Wu, J.-Y., Wang, P.-C. and Hwu, W.-L. (2014) Diagnosis of aromatic L-amino acid decarboxylase deficiency by measuring 3-O-methyldopa concentrations in dried blood spots. Clinica Chimica Acta 431, 19–22.

Gücüyener, K., Kasapkara, C. S., Tümer, L. and Verbeek, M. M. (2014) Aromatic L-amino acid decarboxylase deficiency: A new case from Turkey with a novel mutation. Annals of Indian Academy of Neurology 17, 234–6. Free full text available

Helman, G., Pappa, M. B. and Pearl, P. L. (2014) Widening Phenotypic Spectrum of AADC Deficiency, a Disorder of Dopamine and Serotonin Synthesis. JIMD Reports 17, 23–7. Free full text available

Lee, L. K., Cheung, K. M., Cheng, W. W., Ko, C. H., Lee, H. H. C., Ching, C. K. and Mak, C. M. (2014) A rare cause of severe diarrhoea diagnosed by urine metabolic screening: aromatic L-amino acid decarboxylase deficiency. Hong Kong Medical Journal 20, 161–4. Free full text available

Marecos, C., Ng, J. and Kurian, M. A. (2014) What is new for monoamine neurotransmitter disorders? Journal of Inherited Metabolic Disease 37, 619–26.

Montioli, R., Dindo, M., Giorgetti, A., Piccoli, S., Cellini, B. and Voltattorni, C. B. (2014) A comprehensive picture of the mutations associated with aromatic amino acid decarboxylase deficiency: from molecular mechanisms to therapy implications. Human Molecular Genetics 23, 5429–40.

San Sebastian, W., Kells, A. P., Bringas, J., Samaranch, L., Hadaczek, P., Ciesielska, A. and Bankiewicz, K. S. (2014) Safety and tolerability of MRI-guided infusion of AAV2-hAADC into the mid-brain of non-human primate. Molecular Therapy. Methods & Clinical Development 3, 14049. Free full text available

Atwal, P. S., Donti, T. R., Cardon, A. L., Bacino, C. A., Sun, Q., Emrick, L. and Elsea, S. H. (2015) Aromatic l-amino acid decarboxylase deficiency diagnosed by clinical metabolomic profiling of plasma. Molecular Genetics and Metabolism 115, 91-94.

Leuzzi, V., Mastrangelo, M., Polizzi, A., Artiola, C., van Kuilenburg, A. B. P., Carducci, C. and Carducci, C. (2015) Report of two never treated adult sisters with aromatic L-amino acid decarboxylase deficiency: a portrait of the natural history of the disease or an expanding phenotype? JIMD Reports 15, 39–45. Free full text available

Chien, Y. H., Chen, P. W., Lee, N. C., Hsieh, W. S., Chiu, P. C., Hwu, W. L., Tsai, F. J., Lin, S. P., Chu, S. Y., Jong, Y. J. & Chao, M. C. (2016) 3-O-methyldopa levels in newborns: Result of newborn screening for aromatic l-amino-acid decarboxylase deficiency. Mol Genet Metab, 118, 259-63.

Kojima, K., Anzai, R., Ohba, C., Goto, T., Miyauchi, A., Thöny, B., Saitsu, H., Matsumoto, N., Osaka, H. & Yamagata, T. 2016. A female case of aromatic l-amino acid decarboxylase deficiency responsive to MAO-B inhibition. Brain Dev, 38, 959-963.

Sanchez-Jiménez, F., Pino-Ángeles, A., Rodríguez-López, R., Morales, M. & Urdiales, J. L. (2016) Structural and functional analogies and differences between histidine decarboxylase and aromatic l-amino acid decarboxylase molecular networks: Biomedical implications. Pharmacol Res, 114, 90-102.

Caine, C., Shohat, M., Kim, J. K., Nakanishi, K., Homma, S., Mosharov, E. V. & Monani, U. R. (2017) A pathogenic S250F missense mutation results in a mouse model of mild aromatic l-amino acid decarboxylase (AADC) deficiency. Hum Mol Genet, 26, 4406-4415.

Chien, Y. H., Lee, N. C., Tseng, S. H., Tai, C. H., Muramatsu, S. I., Byrne, B. J. & Hwu, W. L. (2017) Efficacy and safety of AAV2 gene therapy in children with aromatic L-amino acid decarboxylase deficiency: an open-label, phase 1/2 trial. Lancet Child Adolesc Health, 1, 265-273.

Lee, W. T., Lin, J. H., Weng, W. C. & Peng, S. S. (2017) Microstructural changes of brain in patients with aromatic L-amino acid decarboxylase deficiency. Hum Brain Mapp, 38, 1532-1540.

Spitz, M. A., Nguyen, M. A., Roche, S., Heron, B., Milh, M., De Lonlay, P., Lion-François, L., Testard, H., Napuri, S., Barth, M., Fournier-Favre, S., Christa, L., Vianey-Saban, C., Corne, C. & Roubertie, A. (2017) Chronic Diarrhea in L-Amino Acid Decarboxylase (AADC) Deficiency: A Prominent Clinical Finding Among a Series of Ten French Patients. JIMD Rep, 31, 85-93.

Wassenberg, T., Molero-Luis, M., Jeltsch, K., Hoffmann, G. F., Assmann, B., Blau, N., Garcia-Cazorla, A., Artuch, R., Pons, R., Pearson, T. S., Leuzzi, V., Mastrangelo, M., Pearl, P. L., Lee, W. T., Kurian, M. A., Heales, S., Flint, L., Verbeek, M., Willemsen, M. & Opladen, T. (2017) Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. Orphanet J Rare Dis, 12, 12.

Wiznitzer, M. (2017) Gene therapy for children with AADC deficiency. Lancet Child Adolesc Health, 1, 250-251.

Dai, L., Ding, C. & Fang, F. (2018) A novel DDC gene deletion mutation in two Chinese mainland siblings with aromatic l-amino acid decarboxylase deficiency. Brain Dev.

Hwu, W. L., Chien, Y. H., Lee, N. C. & Li, M. H. (2018) Natural History of Aromatic L-Amino Acid Decarboxylase Deficiency in Taiwan. JIMD Rep, 40, 1-6.

Montioli, R., Janson, G., Paiardini, A., Bertoldi, M. & Borri Voltattorni, C. (2018) Heterozygosis in aromatic amino acid decarboxylase deficiency: Evidence for a positive interallelic complementation between R347Q and R358H mutations. IUBMB Life, 70, 215-223.

Portaro, S., Gugliandolo, A., Scionti, D., Cammaroto, S., Morabito, R., Leonardi, S., Fraggetta, F., Bramanti, P. & Mazzon, E. (2018) When dysphoria is not a primary mental state: A case report of the role of the aromatic L-aminoacid decarboxylase. Medicine (Baltimore), 97, e10953.

Tsai, C. R., Lee, H. F., Chi, C. S., Yang, M. T. & Hsu, C. C. (2018) Antisense oligonucleotides modulate dopa decarboxylase function in aromatic l-amino acid decarboxylase deficiency. Hum Mutat, 39, 2072-2082.