SUPPLEMENTARY MATERIAL

Table. Papers and cases included in the study

Author	Cases reported	Cases included	uncontroll	controlled	asymptomatic	No outcome or treatment	w/o genotype or phenotype	duplicated cases	ref	PMID
Zhou Q, et al (2014)	9	5	2	3		4			[1]	24552284
Navon Elkan P, et al (2014)	24	21	5	14	2		3		[2]	24552285
Garg N, et al (2014)	1	1	1						[3]	24737293
van Montfrans J, et al (2014)	2	2	1	1					[4]	25075845
Van Eyck L, et al (2014)	2	2		2					[5]	25075846
Bras J,et al (2014)	3	0				3			[6]	25075847
Van Eyck L, et al (2014)	1	1		1					[7]	25075848
Belot A, et al. (2014)	2	2	2						[8]	25278816
Van Eyck L, Jr., et al. (2015)	2	0						2 cases in 25075846	[9]	25457153
Westendorp WF, et al. (2015)	2	1		1		1			[10]	25888558
Gonzalez Santiago TM, et al (2015)	2	2	1		1				[11]	26131734
Batu ED, et al (2015)	6	3	1	2				3 cases in 24552284 (16-18)	[12]	26233953
Fellmann F, et al. (2016)	2	2	2						[13]	26607704

Van Montfrans JM, et al (2016)	9	5		5				4 cases of 25888558	[14]	26867732
Poswar Fde O, et al (2016)	2	2	1		1				[15]	26914925
Schepp J, et al (2016)	2	2	2						[16]	26922074
Nanthapisal S, et al (2016)	15	15		10	5				[17]	27059682
Keer N, et al (2016)	1	1		1					[18]	27069017
Hsu AP, et al. (2016)	1	1		1					[19]	27130863
Uettwiller F, et al. (2016)	2	2		2					[20]	27252897
Pichard DC, et al. (2016)	1	0						1 case in 24552284	[21]	27444081
Ben-Ami T, et al. (2016)	5	4	4			1			[22]	27514238
Elbracht M, et al. (2017)	1	1		1					[23]	28024309
Hashem H, et al. (2017)	1	0						One case in 28974505	[24]	28230570
Schepp J, et al. (2017)	11	5	3	2		4		2 cases in 26922074	[25]	28493328
Sahin S, et al (2018)	8	5	2	3				One case in 24552285, one in 24737293 and the 3rd case in 30642227	[26]	28516235
Caorsi R, et al. (2017)	17	17		16	1				[27]	28522451
Nihira H, et al (2018)	1	1		1					[28]	28665179
Hashem H, et al. (2017)	1	0						One case in 28974505	[29]	28805790

Skrabl-Baumgartner A, et al (2017)	2	2	2					[30]	28830446
Hashem H, et al. (2017)	14	10		10			One in 27130863, 2nd in 26867732, 3rd in 25075846 and 4th in 25075845	[31]	28974505
Bucciol G, et al. (2017)	1	0					1 case in 25075846	[32]	28993957
Alsultan A, et al (2018)	1	1		1				[33]	29271561
Trotta L, et al. (2018)	9	4	2	1	1	5		[34]	29391253
Gunthner R, et al. (2018)	1	1		1				[35]	29391272
Michniacki TF, et al. (2018)	2	2		1	1			[36]	29411230
Cipe FE, et al. (2018)	1	1	1					[37]	29564582
Lamprecht P, et al (2018)	1	1		1				[38]	29600946
Sundin et al (2019)	1	1		1				[39]	29620681
Rama M et al (2018)	13	0				12	One case in in 28522451 (case 5)	[40]	29681619
Teixeira V, et al (2018)	1	1		1				[41]	29916967
Springer JM, et al. (2018)	2	2		2				[42]	29963054
Van Nieuwenhove E, et al. (2018)	2	1			1		One case in 25075848	[43]	30139808
Maccora I, et al (2018)	1	1		1				[44]	30148442

Sharma A, et al. (2019)	1	1		1						[45]	30165497
Kisla Ekinci RM, et al. (2018)	1	1		1						[46]	30377239
Arts K, et al. (2018)	3	3	2		1					[47]	30386947
Martin H, et al (2018)	2	2	1	1						[48]	30514670
Claassen D, et al. (2018)	1	1	1							[49]	30559313
Liu L, et al (2019)	1	1		1						[50]	30610243
Sahin et al (2019)	2	1		1			1			[51]	30642227
Alabbas F, et al. (2019)	2	2	1	1						[52]	30644014
Insalaco A, et al. (2019)	5	1		1					4 cases in 28522451	[53]	30647181
Barzaghi F, et al (2018)	1	1		1						[54]	30692987
Grossi A, et al. (2019)	1	1		1						[55]	30920658
Ghurye RR, et al (2019)	2	2	1	1						[56]	30924144
Goncalves TDS, et al. (2019)	1	0				1				[57]	30962308
Gibson KM, et al (2019)	11	8	3	5			3			[58]	31008556
Ozen S, et al (2020)	24	18	5	13					6 cases in 26233953	[59]	31043544
Liebowitz J,et al. (2019)	3	3		3						[60]	31092714
Clarke K, et al. (2019)	1	1		1						[61]	31291964

Kisla Ekinci RM, et al. (2020)	5	4		4				one case in 30377239	[62]	31292637
Ekinci RMK et al (2019)	1	1		1					[63]	31522599
Sozeri B, et al. (2019)	5	4		4				Once case in 28516235	[64]	31541281
Akgun-Dogan O, et al. (2019)	1	1	1						[65]	31584751
Alaygut D, et al. (2019)	1	1	1						[66]	31598601
Chong-Neto HJ, et al. (2019)	1	1		1					[67]	31617030
Krutzke S et al. (2019)	2	2		2					[68]	31651641
Goschl L, et al. (2020)	1	1		1					[69]	31686313
Tanatar A, et al. (2020)	5	4		4				One case in 31916720	[70]	31848804
van Well GTJ, et al. (2019)	6	6	1	5					[71]	31856934
Cakan M, et al. (2019)	1	1		1					[72]	31916720
Lee PY, et al. (2020)	15	15	14	1					[73]	31945408
Ganhão S, et al (2020)	2	2	1	1					[74]	32535845
Schnappauf O, et al (2020)	6	6	1	3	2				[75]	32638197
Saettini F, et al (2020)	2	1			1	1			[76]	32659374
Sarrabay G, et al. (2015)	5	0						5 cases in 29681619	[77]	L609058053
Benner MF, et al. (2016)	1	0						case in 26867732	[78]	L619272204

Chang Y, et al. (2018)	1	1		1					[79]	L622389226
Li J, et al. (2018)	1	1	1						[80]	L624305135
Schnider C, et al. (2018)	1	1		1					[81]	L625167642
Naidu GSRSNK, et al. (2018)	3	1		1			1	One case in 30165497	[82]	L626163913
Garbarino F, et al. (2019)	1	1		1					[83]	L627973706
Kilic SS, et al. (2019)	1	0						same case in L629479534	[84]	L628681361
Paola K, et al. (2019)	2	2		2					[85]	L628807871
Ouail DE, et al. (2019)	3	0				1	2		[86]	L628809641
Kilic SS, et al. (2019)	1	1	1						[87]	L629479534
Al Mosawi Z,et al.(2019)	1	1		1					[88]	L629806493
Caorsi R, et al. (2014)	3	0						3 cases in 28522451	[89]	L71686775
Van Montfrans J, et al. (2014)	2	0						2 cases in 25075845	[90]	L71778687
Toz B, et al. (2015)	1	1		1					[91]	L72064769
Sasa GS, et al. (2015)	2	0				1		1 case in 28974505	[92]	L72174305
Loureiro G, et al. (2019)	1	0						1 case in 32535845	[93]	L628841823
Neishabury M (2020)	2	2	1	1					[94]	31097629
Our cases	2	2		2						
Total	**338**	**242**	**68**	**157**	**17**	**34**	**10**	**52**		

Note: A total of 94 papers were enclosed, reporting 336 cases. 52 cases were excluded due to repeated reporting in different papers and 10 cases were excluded due to unconfirmed genotype or lack of detailed phenotype information. A further 34 cases were excluded due to lack of outcome or treatment report. Along with 2 cases from our center, a total of 242 cases were enclosed, with 17 patients were asymptomatic requiring no treatment, 68 patients had uncontrolled disease after treatment and 157 patients reached disease control after treatment.

References

1. Zhou Q, Yang D, Ombrello AK, Zavialov AV, Toro C, Zavialov AV, et al. Early-onset stroke and vasculopathy associated with mutations in ADA2. N Eng J Med 2014; 370:911-920.

2. Navon Elkan P, Pierce SB, Segel R, Walsh T, Barash J, Padeh S, et al. Mutant adenosine deaminase 2 in a polyarteritis nodosa vasculopathy. N Eng J Med 2014; 370:921-931.

3. Garg N, Kasapcopur O, Foster J, 2nd, Barut K, Tekin A, Kizilkilic O, et al. Novel adenosine deaminase 2 mutations in a child with a fatal vasculopathy. Eur J Pediatr 2014; 173:827-830.

4. van Montfrans J, Zavialov A, Zhou Q: Mutant ADA2 in vasculopathies. N Eng J Med 2014; 371:478.

5. Van Eyck L, Liston A, Meyts I: Mutant ADA2 in vasculopathies. N Eng J Med 2014; 37:478-479.

6. Bras J, Guerreiro R, Santo GC: Mutant ADA2 in vasculopathies. N Eng J Med 2014; 371:478-480.

7. Van Eyck L, Liston A, Wouters C: Mutant ADA2 in vasculopathies. N Eng J Med 2014; 371:480.

8. Belot A, Wassmer E, Twilt M, Lega JC, Zeef LA, Oojageer A, et al. Mutations in CECR1 associated with a neutrophil signature in peripheral blood. Pediatr Rheumatol Online J 2014; 12:44.

9. Van Eyck L, Hershfield MS, Pombal D, Kelly SJ, Ganson NJ, Moens L, et al. Hematopoietic stem cell transplantation rescues the immunologic phenotype and prevents vasculopathy in patients with adenosine deaminase 2 deficiency. J Allergy Clin Immunol 2015; 135:283-287.

10. Westendorp WF, Nederkoorn PJ, Aksentijevich I, Hak AE, Lichtenbelt KD, Braun KP. Unexplained early-onset lacunar stroke and inflammatory skin lesions: Consider ADA2 deficiency. Neurology 2015; 84:2092-2093.

11. Gonzalez Santiago TM, Zavialov A, Saarela J, Seppanen M, Reed AM, Abraham RS, et al. Dermatologic Features of ADA2 Deficiency in Cutaneous Polyarteritis Nodosa. JAMA Dermatol 2015; 151:1230-1234.

12. Batu ED, Karadag O, Taskiran EZ, Kalyoncu U, Aksentijevich I, Alikasifoglu M, et al. A Case Series of Adenosine Deaminase 2-deficient Patients Emphasizing Treatment and Genotype-phenotype Correlations. J Rheumatol 2015; 42:1532-1534.

13. Fellmann F, Angelini F, Wassenberg J, Perreau M, Arenas Ramirez N, Simon G, et al. IL-17 receptor A and adenosine deaminase 2 deficiency in siblings with recurrent infections and chronic inflammation. J Allergy Clin Immunol 2016; 137:1189-1196 e1182.

14. Van Montfrans JM, Hartman EA, Braun KP, Hennekam EA, Hak EA, Nederkoorn PJ, et al. Phenotypic variability in patients with ADA2 deficiency due to identical homozygous R169Q mutations. Rheumatology 2016; 55:902-910.

15. Poswar Fde O, da Fonseca RM, de Albuquerque LC, Zhou Q, Jardim LB, Monte TL, et al. Adenosine deaminase 2 deficiency presenting as spastic paraplegia and systemic vasculitis. J Neurol 2016; 263:818-820.

16. Schepp J, Bulashevska A, Mannhardt-Laakmann W, Cao H, Yang F, Seidl M, et al. Deficiency of Adenosine Deaminase 2 Causes Antibody Deficiency. J Clin Immunol 2016; 36:179-186.

17. Nanthapisal S, Murphy C, Omoyinmi E, Hong Y, Standing A, Berg S, et al. Deficiency of Adenosine Deaminase Type 2: A Description of Phenotype and Genotype in Fifteen Cases. Arthritis Rheumatol 2016; 68:2314-2322.

18. Keer N, Hershfield M, Caskey T, Unizony S. Novel compound heterozygous variants in CECR1 gene associated with childhood onset polyarteritis nodosa and deficiency of ADA2. Rheumatology 2016; 55:1145-1147.

19. Hsu AP, West RR, Calvo KR, Cuellar-Rodriguez J, Parta M, Kelly SJ, et al. Adenosine deaminase type 2 deficiency masquerading as GATA2 deficiency: Successful hematopoietic stem cell transplantation. J Allergy Clin Immunol 2016; 138:628-630 e622.

20. Uettwiller F, Sarrabay G, Rodero MP, Rice GI, Lagrue E, Marot Y, et al. ADA2 deficiency: case report of a new phenotype and novel mutation in two sisters. RMD Open 2016; 2:e000236.

21. Pichard DC, Ombrello AK, Hoffmann P, Stone DL, Cowen EW. Early-onset stroke, polyarteritis nodosa (PAN), and livedo racemosa. J Am Acad Dermatol 2016; 75:449-453.

22. Ben-Ami T, Revel-Vilk S, Brooks R, Shaag A, Hershfield MS, Kelly SJ, et al. Extending the Clinical Phenotype of Adenosine Deaminase 2 Deficiency. J Pediatr 2016; 177:316-320.

23. Elbracht M, Mull M, Wagner N, Kuhl C, Abicht A, Kurth I, et al. Stroke as Initial Manifestation of Adenosine Deaminase 2 Deficiency. Neuropediatrics 2017; 48:111-114.

24. Hashem H, Egler R, Dalal J. Refractory Pure Red Cell Aplasia Manifesting as Deficiency of Adenosine Deaminase 2. J Pediatr Hematol Oncol 2017; 39:e293-e296.

25. Schepp J, Proietti M, Frede N, Buchta M, Hubscher K, Rojas Restrepo J, et al. Screening of 181 Patients With Antibody Deficiency for Deficiency of Adenosine Deaminase 2 Sheds New Light on the Disease in Adulthood. Arthritis Rheumatol 2017; 69:1689-1700.

26. Sahin S, Adrovic A, Barut K, Ugurlu S, Turanli ET, Ozdogan H, et al. Clinical, imaging and genotypical features of three deceased and five surviving cases with ADA2 deficiency. Rheumatol Int 2018; 38:129-136.

27. Caorsi R, Penco F, Grossi A, Insalaco A, Omenetti A, Alessio M, et al. ADA2 deficiency (DADA2) as an unrecognised cause of early onset polyarteritis nodosa and stroke: a multicentre national study. Ann Rheum Dis 2017; 76:1648-1656.

28. Nihira H, Nakagawa K, Izawa K, Kawai T, Yasumi T, Nishikomori R, et al. Fever of unknown origin with rashes in early infancy is indicative of adenosine deaminase type 2 deficiency. Scand J Rheumatol 2018; 47:170-172.

29. Hashem H, Vatsayan A, Gupta A, Nagle K, Hershfield M, Dalal J. Successful reduced intensity hematopoietic cell transplant in a patient with deficiency of adenosine deaminase 2. Bone Marrow Transplant 2017; 52:1575-1576.

30. Skrabl-Baumgartner A, Plecko B, Schmidt WM, Konig N, Hershfield M, Gruber-Sedlmayr U, et al. Autoimmune phenotype with type I interferon signature in two brothers with ADA2 deficiency carrying a novel CECR1 mutation. Pediatr Rheumatol Online J 2017; 15:67.

31. Hashem H, Kumar AR, Muller I, Babor F, Bredius R, Dalal J, et al. Hematopoietic stem cell transplantation rescues the hematological, immunological, and vascular phenotype in DADA2. Blood 2017; 130:2682-2688.

32. Bucciol G, Delafontaine S, Segers H, Bossuyt X, Hershfield MS, Moens L, et al. Hematopoietic Stem Cell Transplantation in ADA2 Deficiency: Early Restoration of ADA2 Enzyme Activity and Disease Relapse upon Drop of Donor Chimerism. J Clin Immunol 2017; 37:746-750.

33. Alsultan A, Basher E, Alqanatish J, Mohammed R, Alfadhel M. Deficiency of ADA2 mimicking autoimmune lymphoproliferative syndrome in the absence of livedo reticularis and vasculitis. Pediatr Blood Cancer 2018; 65 .

34. Trotta L, Martelius T, Siitonen T, Hautala T, Hamalainen S, Juntti H, et al. ADA2 deficiency: Clonal lymphoproliferation in a subset of patients. J Allergy Clin Immunol 2018; 141:1534-1537 e1538.

35. Gunthner R, Wagner M, Thurm T, Ponsel S, Hofele J, Lange-Sperandio B. Identification of co-occurrence in a patient with Dent's disease and ADA2-deficiency by exome sequencing. Gene 2018; 649:23-26.

36. Michniacki TF, Hannibal M, Ross CW, Frame DG, DuVall AS, Khoriaty R, et al. Hematologic Manifestations of Deficiency of Adenosine Deaminase 2 (DADA2) and Response to Tumor Necrosis Factor Inhibition in DADA2-Associated Bone Marrow Failure. J Clin Immunol 2018; 38:166-173.

37. Cipe FE, Aydogmus C, Serwas NK, Keskindemirci G, Boztug K. Novel Mutation in CECR1 Leads to Deficiency of ADA2 with Associated Neutropenia. J Clin Immunol 2018; 38:273-277.

38. Lamprecht P, Humrich JY, Diebold I, Riemekasten G. Diagnosis of deficiency of adenosine deaminase 2 with early onset polyarteritis nodosa in an adult patient with a novel compound heterozygous CECR1 mutation. Clin Exp Rheumatol 2018; 36 Suppl 111:177.

39. Sundin M, Marits P, Nierkens S, Kolios AGA, Nilsson J. "Immune" Thrombocytopenia as Key Feature of a Novel ADA2 Deficiency Variant: Implication on Differential Diagnostics of ITP in Children. J Pediatr Hematol Oncol 2019; 41:155-157.

40. Rama M, Duflos C, Melki I, Bessis D, Bonhomme A, Martin H, et al. A decision tree for the genetic diagnosis of deficiency of adenosine deaminase 2 (DADA2): a French reference centres experience. Eur J Hum Genet 2018; 26:960-971.

41. Teixeira V, Oliveira-Ramos F, Costa M. Severe and Refractory Polyarteritis Nodosa Associated With CECR1 Mutation and Dramatic Response to Infliximab in Adulthood. J Clin Rheumatol 2020;26:e66-e69.

42. Springer JM, Gierer SA, Jiang H, Kleiner D, Deuitch N, Ombrello AK, et al. Deficiency of Adenosine Deaminase 2 in Adult Siblings: Many Years of a Misdiagnosed Disease With Severe Consequences. Front Immunol 2018; 9:1361.

43. Van Nieuwenhove E, Humblet-Baron S, Van Eyck L, De Somer L, Dooley J, Tousseyn T, et al. ADA2 Deficiency Mimicking Idiopathic Multicentric Castleman Disease. Pediatrics 2018; 142:e20172266.

44. Maccora I, Frongia I, Azzari C, Ricci S, Cimaz R, Simonini G. A misleading case of deficiency of adenosine deaminase 2 (DADA2): the magnifying glass of the scientific knowledge drives the tailored medicine in real life. Clin Exp Rheumatol 2018; 36(6 Suppl 115):146.

45. Sharma A, Naidu G, Chattopadhyay A, Acharya N, Jha S, Jain S. Novel CECR1 gene mutations causing deficiency of adenosine deaminase 2, mimicking antiphospholipid syndrome. Rheumatology 2019; 58:181-182.

46. Kisla Ekinci RM, Balci S, Bisgin A, Hershfield M, Atmis B, Dogruel D, et al. Renal Amyloidosis in Deficiency of Adenosine Deaminase 2: Successful Experience With Canakinumab. Pediatrics 2018; 142:e20180948.

47. Arts K, Bergerson JRE, Ombrello AK, Similuk M, Oler AJ, Agharahimi A, et al. Warts and DADA2: a Mere Coincidence? J Clin Immunol 2018; 38:836-843.

48. Martin H, Bursztejn AC, Cuny JF, Sarrabay G, Schmutz JL, Touitou I, et al. Chronic leg ulcer revealing adenosine deaminase 2 deficiency: an atypical presentation. Eur J Dermatol 2018; 28:847-848.

49. Claassen D, Boals M, Bowling KM, Cooper GM, Cox J, Hershfield M, et al. Complexities of genetic diagnosis illustrated by an atypical case of congenital hypoplastic anemia. Cold Spring Harb Mol Case Stud 2018; 4:a003384.

50. Liu L, Wang W, Wang Y, Hou J, Ying W, Hui X, et al. A Chinese DADA2 patient: report of two novel mutations and successful HSCT. Immunogenetics 2019; 71:299-305.

51. Sahin S, Adrovic A, Barut K, Baran S, Tahir Turanli E, Canpolat N, et al. A 9.5-year-old boy with recurrent neurological manifestations and severe hypertension, treated initially for polyarteritis nodosa, was subsequently diagnosed with adenosine deaminase type 2 deficiency (DADA2) which responded to anti-TNF-alpha. Paediatr Int Child Health 2019;1-4.

52. Alabbas F, Elyamany G, Alsharif O, Hershfield M, Meyts I. Childhood Hodgkin Lymphoma: Think DADA2. J Clin Immunol 2019; 39:26-29.

53. Insalaco A, Moneta GM, Pardeo M, Caiello I, Messia V, Bracaglia C, et al. Variable Clinical Phenotypes and Relation of Interferon Signature with Disease Activity in ADA2 Deficiency. J Rheumatol 2019; 46:523-526.

54. Barzaghi F, Minniti F, Mauro M, Bortoli M, Balter R, Bonetti E, et al. ALPS-Like Phenotype Caused by ADA2 Deficiency Rescued by Allogeneic Hematopoietic Stem Cell Transplantation. Front Immunol 2018; 9:2767.

55. Grossi A, Cusano R, Rusmini M, Penco F, Schena F, Podda RA, et al. ADA2 deficiency due to a novel structural variation in 22q11.1. Clin Genet 2019; 95:732-733.

56. Ghurye RR, Sundaram K, Smith F, Clark B, Simpson MA, Fairbanks L, et al. Novel ADA2 mutation presenting with neutropenia, lymphopenia and bone marrow failure in patients with deficiency in adenosine deaminase 2 (DADA2). Br J Haematol 2019; 186:e60-e64.

57. Goncalves TDS, Alves C, da Paz JA, Lucato LT. Teaching NeuroImages: Lacunar stroke and polyarteritis nodosa: Consider ADA2 deficiency (DADA2). Neurology 2019; 92:e1801-e1802.

58. Gibson KM, Morishita KA, Dancey P, Moorehead P, Drogemoller B, Han X, et al. Identification of Novel Adenosine Deaminase 2 Gene Variants and Varied Clinical Phenotype in Pediatric Vasculitis. Arthritis Rheumatol 2019; 71:1747-1755.

59. Özen S, Batu ED, Taşkıran EZ, Özkara HA, Ünal Ş, Güleray N, et al. A Monogenic Disease with a Variety of Phenotypes: Deficiency of Adenosine Deaminase 2. J Rheumatol 2020; 47:117-125.

60. Liebowitz J, Hellmann DB, Schnappauf O. Thirty Years of Followup in 3 Patients with Familial Polyarteritis Nodosa due to Adenosine Deaminase 2 Deficiency. J Rheumatol 2019; 46:1059-1060.

61. Clarke K, Campbell C, Omoyinmi E, Hong Y, Al Obaidi M, Sebire N, et al. Testicular ischemia in deficiency of adenosine deaminase 2 (DADA2). Pediatr Rheumatol Online J 2019; 17:39.

62. Kisla Ekinci RM, Balci S, Hershfield M, Bisgin A, Dogruel D, Altintas DU, et al. Deficiency of adenosine deaminase 2: a case series revealing clinical manifestations, genotypes and treatment outcomes from Turkey. Rheumatology 2020; 59:254-256.

63. Ekinci RMK, Balci S, Bisgin A, Sasmaz I, Leblebisatan G, Incecik F, et al. A homozygote novel L451W mutation in CECR1 gene causes deficiency of adenosine deaminase 2 in a pediatric patient representing with chronic lymphoproliferation and cytopenia. Pediatr Hematol Oncol 2019; 36:376-381.

64. Sozeri B, Ercan G, Dogan OA, Yildiz J, Demir F, Doganay L. The same mutation in a family with adenosine deaminase 2 deficiency. Rheumatol Int 2021; 41:227-233.

65. Akgun-Dogan O, Simsek-Kiper PO, Taskiran E, Lissewski C, Brinkmann J, Schanze D, et al. ADA2 deficiency in a patient with Noonan syndrome-like disorder with loose anagen hair: The co-occurrence of two rare syndromes. Am J Med Genet A 2019; 17912:1-7.

66. Alaygut D, Alparslan C, Oncel EP, Mutlubas F, Ozdemir T, Yavascan O, et al. A Child Diagnosed With Treatment-Resistant Polyarteritis Nodosa: Can the Clinical Diagnosis Be Different? Arch Rheumatol 2019; 34:338-342.

67. Chong-Neto HJ, Segundo GRS, Bandeira M, Chong-Silva DC, Rosario CS, Riedi CA, et al. Homozygous Splice ADA2 Gene Mutation Causing ADA-2 Deficiency. J Clin Immunol 2019; 39:842-845.

68. Krutzke S, Horneff G. Treatment of Two Male Children Suffering From Deficiency of Adenosine Deaminase Type 2 (DADA2) With TNF-Inhibitor Etanercept. J Clin Rheumatol 2019. doi: 10.1097/RHU.0000000000001145

69. Goschl L, Winkler S, Dmytrus J, Heredia RJ, Lagler H, Ramharter M, et al. Unreported Missense Mutation in the Dimerization Domain of ADA2 Leads to ADA2 Deficiency Associated with Severe Oral Ulcers and Neutropenia in a Female Somalian Patient-Addendum to the Genotype-Phenotype Puzzle. J Clin Immunol 2020; 40:223-226.

70. Tanatar A, Karadag SG, Sozeri B, Sonmez HE, Cakan M, Kendir Demirkol Y, et al: ADA2 Deficiency. Case Series of Five Patients with Varying Phenotypes. J Clin Immunol 2020; 402:253-258.

71. van Well GTJ, Kant B, van Nistelrooij A, Sirma Ekmekci S, Henriet SV, Hoppenreijs E, et al. Phenotypic variability including Behcet's disease-like manifestations in DADA2 patients due to a homozygous c.973-2A>G splice site mutation. Clin Exp Rheumatol 2019; 37 Suppl 121:142-146.

72. Cakan M, Aktay-Ayaz N, Karadag SG, Tahir-Turanli E, Stafstrom K, Bainter W, et al. Atypical phenotype of an old disease or typical phenotype of a

new disease: deficiency of adenosine deaminase 2. Turk J Pediatr 2019; 61:413-417.

73. Lee PY, Kellner ES, Huang Y, Furutani E, Huang Z, Bainter W, et al. Genotype and functional correlates of disease phenotype in deficiency of adenosine deaminase 2 (DADA2). J Allergy Clin Immunol 2020; 1456:1664-1672.

74. Ganhão S, Loureiro GB, Oliveira DR, Dos-Reis-Maia R, Aguiar F, Quental R, et aI. Two cases of ADA2 deficiency presenting as childhood polyarteritis nodosa: novel ADA2 variant, atypical CNS manifestations, and literature review. Clin Rheumatol 2020; 39:3853-3860.

75. Schnappauf O, Zhou Q, Moura NS, Ombrello AK, Michael DG, Deuitch N, et al. Deficiency of Adenosine Deaminase 2 (DADA2): Hidden Variants, Reduced Penetrance, and Unusual Inheritance. J Clin Immunol 2020; 40:917-926.

76. Saettini F, Fazio G, Corti P, Quadri M, Bugarin C, Gaipa G, et al. Two siblings presenting with novel ADA2 variants, lymphoproliferation, persistence of large granular lymphocytes, and T-cell perturbations. Clin Immunol 2020; 218:108525.

77. Sarrabay G, Insalaco A, Uettwiller F, Tieulié N, Quartier-dit-maire P, Melki J, et aI. Identification of three ADA2 deficiency families with novel CECR1 mutations. Pediatr Rheumatol 2015; 13:229DUMMY.

78. Benner MF, Jansen PM, Kollen WJW, Vermeer MH. Adenosine deaminase 2 (ADA2) deficiency. Nederlands Tijdschrift voor Dermatologie en Venereologie 2016; 26:686-688.

79. Chang Y, Derfalvi B, Issekutz A, Shi J, Alonzo P, Pascual CJ, et al. ADA2 deficiency: Case report of a rare phenotype with alps and CVID-like presentation. J Clin Immunol 2018; 38:341-342.

80. Li J, Song H. A case report of adenosine deaminase 2 deficiency in children and review of literature. Int J Rheum Dis 2018; 21:127-128.

81. Schnider C, Theodoropoulou K, Candotti F, Angelini F, Perreau M, Riccio O, et al. A family case of ADA2 deficiency with cecr1 mutation. Pediatric Rheumatology 2018, 16.

82. Naidu GSRSNK, Acharya N, Jha S, Chattopadhyay A, Dhir V, Goyal M, et al. Deficiency of adenosine deaminase 2: Report of three cases from single center in North India. Indian J Rheumatol 2018; 13:S211-S212.

83. Garbarino F, Caorsi R, Volpi S, Grossi A, Ceccherini I, Gattorno M. A case of adenosine deaminase 2 deficiency (DADA2) with an uncommon clinical presentation and response to IV IG. Pediatr Rheumatol 2019; 17(Suppl 1):P1027.

84. Kilic SS, Cekic S, Karali Y. Severe neutropenia in ADA2 deficiency. Arch Dis Child 2019; 104(Suppl 3):A304.

85. Paola K, Gomes FHR, Benevides LC, Leite MF, Medeiros P, Santos AC, et al. CECR1/ADA2 mutation in a Brazilian family. Ann Rheum Dis 2019;

78:2009.

86. Ouail DE, Tebbani M, Si Ahmed D, Bouali F. Youth hypertension associated with ADA2 deficiency. About three cases. J Hypertens 2019; 37:e215.

87. Kilic SS, Cekic S, Karali Y. Cerebral ischemic attacks in ADA2 deficiency treated with adalimumab. Allergy 2019; 74(Suppl 106):828.

88. Al Mosawi Z, Abduljawad H, Busehail M, Al Moosawi B. Adenosine deaminase 2 deficiency with a novel variant of CECR1 gene mutation: Responding to tumor necrosis factor antagonist therapy. Indian J Rheumatol 2019; 14:236-240.

89. Caorsi R, Omenetti A, Picco P, Buoncompagni A, Minoia F, Federici S, et al. Long-term efficacy of etanercept in ADA2 deficiency. Pediatr Rheumatol 2014; 12(Suppl 1).

90. Van Montfrans J, Van Royen-Kerkhof A, Bierings M, Aksentijevich I, Zavialov A, Zhou Q. Hematological stem cell transplantation in ADA2 deficiency. J Clin Immunol 2014; 34:S230-S231.

91. Toz B, Erer B, Kamali S, Ocal L, Gul A. Differential response to anakinra and adalimumab in a patient with DADA2 syndrome. Pediatr Rheumatol 2015; 13:276DUMMY.

92. Sasa GS, Elghetany MT, Bergstrom K, Nicholas S, Himes R, Krance RA, et al. Adenosine deaminase 2 deficiency as a cause of pure red cell aplasia mimicking diamond blackfan anemia. Blood 2015; 126:3615.

93. Loureiro G, Oliveira D, Ganhão S, Aguiar F, Rodrigues M, Brito I. ADA2 deficiency presenting as infantile polyarteritis nodosa. Ann Rheum Dis 2019; 78(Suppl 2):1985.

94. Neishabury M, Mehri M, Fattahi Z, Najmabadi H, Azarkeivan A. Novel variants in Iranian individuals suspected to have inherited red blood cell disorders, including bone marrow failure syndromes. Haematologica. 2020;105:e1-e4