CURRICULUM VITAE

Name: Lina Basel-Salmon, MD, PhD

Faculty/Dept: Sackler Faculty of Medicine, Tel Aviv University, Tel Aviv, Israel/ Pediatrics & Human Genetics

Hospital/Dept: The Raphael Recanati Genetics Institute
Rabin Medical Center – Beilinson Hospital and Schneider Children's Medical Center of Israel, Petach Tikva, Israel

Email: basel@tauex.tau.ac.il

Current position: Director, The Raphael Recanati Institute of Genetics, Rabin Medical Center – Beilinson Hospital, Petach Tikva, Israel

Main achievements:

- Full Professor at the Sackler School of Medicine, Tel Aviv University
- Has led the establishment of the genetic counseling MSc program at Tel Aviv University
- Winner of the Outstanding Lecturer award in 2013
- Recipient of a large number of competitive research grants, including two grants from the Israeli Science Foundation, The Stolz University foundation, The Chief Scientist of the Ministry of Health, the TEMPUS grant by the European Union and others
- Former Head of the Israeli Society of Medical Genetics (2015-2018)
- Board member, European Society of Human Genetics

Main areas of research include identification of new gene-disease corellations and artificial intelligence-based diagnostics of genetic diseases, specifically: 1) automatic recognition of abnormal features from facial photos; 2) the role of artificial intelligence-based platforms in the interpretation of exome sequencing results; 3) the role of clinical geneticists in genomic variant interpretation process of their patients.

EDUCATION

PERIODS OF STUDIES

1984-1990 Vilnius University, Faculty of Medicine, Vilnius, Lithuania
Subject: Medicine

MD degree [with honors]

1991-1995 **PhD degree**
Title of Doctoral Dissertation: Genetic analysis of ataxia-telangiectasia (A-T) locus in the chromosomal region 11q22-23
Supervised by: Prof. Yosef Shiloh

1995-2000 Pediatrics Specialization Board Examinations [with honors]

2000-2002 Genetics Specialization Board Examinations [with honors]

ACADEMIC & PROFESSIONAL ACTIVITIES & ACHIEVEMENTS

Academic Experience:

2002-2004 Instructor, Department of Pediatrics, Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel

2004-2008 Lecturer, Department of Pediatrics, Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel

2008-2013 Senior Lecturer, Department of Pediatrics, affiliated with the Department of Human Genetics and Biochemistry, Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel

2013-2017 Associate Professor, Department of Pediatrics, affiliated with the Department of Human Genetics and Biochemistry, Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel

2017-present Full Professor, Department of Pediatrics, affiliated with the Department of Human Genetics and Biochemistry, Sackler School of Medicine, Tel Aviv University, Tel Aviv, Israel

Professional Experience:

1995-2000 Resident, Pediatrics, Schneider Children's Medical Center of Israel, Petach Tikva, Israel

2000-2002 Resident, Genetics Institute, Rabin Medical Center – Beilinson Hospital, and SCMCI, Petach Tikva, Israel

2002-2008 Senior Geneticist, SCMCI, Petach Tikva and Raphael Recanati Genetic Institute, Rabin Medical Center – Beilinson Hospital, Petach Tikva, Israel

2002-2008 Scientific exchange visits to the Necker Hospital, Paris, France; Boston Children's Hospital, Boston, USA; Chicago University, Chicago, USA; Great Ormond's Children's Hospital, London, UK

2008-2014 Director, Pediatric Genetics Unit, SCMCI, Petach Tikva and The Raphael Recanati Genetic Institute, Rabin Medical Center – Beilinson Hospital, Petach Tikva, Israel

2012-present	Head, Immunology and Genetics Laboratory, Felsenstein Medical Research Center, RMC, Petach Tikva; Tel Aviv University, Tel Aviv, Israel
2015-present	Director, The Raphael Recanati Genetic Institute, Rabin Medical Center – Beilinson Hospital and Schneider Children's Medical Center of Israel, Petach Tikva, Israel
2019-present	Board member, European Society of Human Genetics

AWARDS

2003	Schneider Children's Medical Center of Israel Research Award, "Identification of the gene causing nonsyndromic autosomal recessive mental retardation"
2004	Beilinson Hospital Research Award, "Identification of the gene causing nonsyndromic autosomal recessive mental retardation"
2005	Gertrud Cohen prize for Excellence in Human Genetics for the research project: "Identification of the gene causing nonsyndromic autosomal recessive mental retardation"
2005	Israeli Pediatrics Society research prize for the project "Identification of the gene causing nonsyndromic autosomal recessive mental retardation"
2006	Dani Moran's prize for the scientific paper "Identification of the gene causing nonsyndromic autosomal recessive mental retardation"
2008	Tel Aviv University Sackler School of Medicine prize for Excellence for the poster "Autosomal recessive ichthyosis with hypotrichosis (ARIH) caused by a mutation in ST14, encoding type II transmembrane serine protease matriptase".
2013	Elected as Distinguished Lecturer, the Sackler Faculty of Medicine, Tel Aviv University

SCIENTIFIC PUBLICATIONS

1: Einhorn Y, Einhorn M, Kurolap A, Steinberg D, Mory A, Bazak L, Paperna T, Grinshpun-Cohen J, **Basel-Salmon L**, Weiss K, Singer A, Yaron Y, Baris Feldman H. Community data-driven approach to identify pathogenic founder variants for pan-ethnic carrier screening panels. Hum Genomics. 2023 Mar 28;17(1):30. doi:

10.1186/s40246-023-00472-w. PMID: 36978159; PMCID: PMC10044388.

2: Truty R, Rojahn S, Ouyang K, Kautzer C, Kennemer M, Pineda-Alvarez D, Johnson B, Stafford A, **Basel-Salmon L**, Saitta S, Slavotinek A, Chandrasekharappa SC, Suarez CJ, Burnett L, Nussbaum RL, Aradhya S. Patterns of mosaicism for sequence and copy-number variants discovered through clinical deep sequencing of disease-related genes in one million individuals. Am J Hum Genet. 2023 Apr 6;110(4):551-564. doi: 10.1016/j.ajhg.2023.02.013. Epub 2023 Mar 17. PMID: 36933558.

3: Sukenik-Halevy R, Golbary Kinory E, Laron Kenet T, Brabbing-Goldstein D, Gilboa Y, **Basel-Salmon L**, Perlman S. Prenatal gender-customized head circumference nomograms result in reclassification of microcephaly and macrocephaly. AJOG Glob Rep. 2023 Jan 29;3(1):100171. doi: 10.1016/j.xagr.2023.100171. PMID: 36864987; PMCID: PMC9972400.

4: Maya I, Salzer Sheelo L, Brabbing-Goldstein D, Matar R, Kahana S, Agmon-Fishman I, Klein C, Gurevitch M, **Basel-Salmon L**, Sagi-Dain L. Clinical utility of expanded non-invasive prenatal screening compared with chromosomal microarray in over 8000 pregnancies without major structural anomalies. Ultrasound Obstet Gynecol. 2023 Feb 13. doi: 10.1002/uog.26177. Epub ahead of print. PMID: 36776119.

5: Brabbing-Goldstein D, **Basel-Salmon L**. Kaufman Oculocerebrofacial Syndrome. 2016 Oct 20 [updated 2022 Jul 28]. In: Adam MP, Mirzaa GM, Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993–2023. PMID: 27763745.

6: Sagi-Dain L, Salzer Sheelo L, Brabbing-Goldstein D, Matar R, Kahana S, Agmon-Fishman I, Klein C, Gurevitch M, **Basel-Salmon L**, Maya I. Chromosomal Microarray Analysis Compared With Noninvasive Prenatal Testing in Pregnancies With Abnormal Maternal Serum Screening. Obstet Gynecol. 2022 May 1;139(5):877-887. doi:

10.1097/AOG.0000000000004758. Epub 2022 Apr 5. PMID: 35576346.

7: Libman V, Macarov M, Friedlander Y, Goldman-Mellor S, Israel S, Hochner-Celnikier D, Sompolinsky Y, Dior UP, Osovsky M, **Basel-Salmon L**, Wiznitzer A, Neumark Y, Meiner V, Frumkin A, Shkedi-Rafid S, Hochner H. Postpartum women's attitudes to disclosure of adult-onset conditions in pregnancy. Prenat Diagn. 2022 Jul;42(8):1038-1048. doi: 10.1002/pd.6162. Epub 2022 May 4. PMID: 35484937; PMCID: PMC9539988.

8: Davidov B, Levon A, Volkov H, Orenstein N, Karo R, Fatal Gazit I, Magal N, **Basel-Salmon L**, Golan Mashiach M. Pathogenic variant-based preconception carrier screening in the Israeli Jewish population. Clin Genet. 2022 May;101(5-6):517-529. doi: 10.1111/cge.14131. Epub 2022 Mar 29. PMID: 35315053.

9: Vegas N, Demir Z, Gordon CT, Breton S, Romanelli Tavares VL, Moisset H, Zechi-Ceide R, Kokitsu-Nakata NM, Kido Y, Marlin S, Gherbi Halem S, Meerschaut I, Callewaert B, Chung B, Revencu N, Lehalle D, Petit F, Propst EJ, Papsin BC, Phillips JH, Jakobsen L, Le Tanno P, Thévenon J, McGaughran J, Gerkes EH, Leoni C, Kroisel P, Tan TY, Henderson A, Terhal P, **Basel-Salmon L**, Alkindy A, White SM, Passos-Bueno MR, Pingault V, De Pontual L, Amiel J. Further delineation of auriculocondylar syndrome based on 14 novel cases and reassessment of 25 published cases. Hum Mutat. 2022 May;43(5):582-594. doi: 10.1002/humu.24349. Epub 2022 Mar 7. PMID: 35170830.

10: Steinberg-Shemer O, Orenstein N, Krasnov T, Noy-Lotan S, Marcoux N, Dgany O, Yacobovich J, Gilad O, Shabad E, **Basel-Salmon L**, Tamary H. Congenital thrombocytopenia associated with a heterozygous variant in the MEIS1 gene encoding a transcription factor essential for megakaryopoiesis. Platelets. 2022 May 19;33(4):645-648. doi: 10.1080/09537104.2021.1961704. Epub 2022 Feb 8. PMID: 35130804.

11: Salzer-Sheelo L, Polak U, Barg A, Kahana S, Yacobson S, Agmon-Fishman I,

Klein C, Matar R, Rurman-Shahar N, Sagi-Dain L, **Basel-Salmon L**, Maya I, Sukenik-Halevy R. Prenatal and postnatal chromosomal microarray analysis in 885 cases of various congenital heart defects. Arch Gynecol Obstet. 2022 Oct;306(4):1007-1013. doi: 10.1007/s00404-021-06366-3. Epub 2022 Jan 27. PMID: 35083553.

12: Sukenik-Halevy R, Perlman S, Ruhrman-Shahar N, Engel O, Orenstein N, Gonzaga-Jauregui C, Shuldiner AR; Regeneron Genetics Center; Magal N, Hagari O, Azulay N, Lidzbarsky GA, Bazak L, **Basel-Salmon L**. The prevalence of prenatal sonographic findings in postnatal diagnostic exome sequencing performed for neurocognitive phenotypes: A cohort study. Prenat Diagn. 2022 May;42(6):717-724. doi: 10.1002/pd.6095. Epub 2022 Jan 24. PMID: 35032046; PMCID: PMC9303252.

13: Fellner A, Goldberg Y, Lev D, **Basel-Salmon L**, Shor O, Benninger F. In-silico phenotype prediction by normal mode variant analysis in TUBB4A-related disease. Sci Rep. 2022 Jan 7;12(1):58. doi: 10.1038/s41598-021-04337-x. PMID: 34997144; PMCID: PMC8741991.

14: Michaeli O, Ladany H, Erez A, Ben Shachar S, Izraeli S, Lidzbarsky G, **Basel-Salmon L**, Biskup S, Maruvka YE, Toledano H, Goldberg Y. Di-genic inheritance of germline POLE and PMS2 pathogenic variants causes a unique condition associated with pediatric cancer predisposition. Clin Genet. 2022 Apr;101(4):442-447. doi: 10.1111/cge.14106. Epub 2022 Jan 7. PMID: 34967012.

15: **Basel-Salmon L**, Sukenik-Halevy R. Challenges in variant interpretation in prenatal exome sequencing. Eur J Med Genet. 2022 Feb;65(2):104410. doi: 10.1016/j.ejmg.2021.104410. Epub 2021 Dec 21. PMID: 34952236.

16: Salzer-Sheelo L, Fellner A, Orenstein N, Bazak L, Lev-El Halabi N, Daue M, Smirin-Yosef P, Van Hout CV, Fellig Y, Ruhrman-Shahar N, Staples J, Magal N, Shuldiner AR, Mitchell BD, Nevo Y, Pollin TI, Gonzaga-Jauregui C, **Basel-Salmon L**. Biallelic truncating variants in the muscular A-type lamin-interacting

protein (MLIP) gene cause myopathy with hyperCKemia. Eur J Neurol. 2022 Apr;29(4):1174-1180. doi: 10.1111/ene.15218. Epub 2022 Jan 7. PMID: 34935254.

17: Sagi-Dain L, **Basel-Salmon L**, Maya I. Physician anxiety or maternal choice? Am J Obstet Gynecol. 2022 Apr;226(4):600-601. doi: 10.1016/j.ajog.2021.12.026. Epub 2021 Dec 16. PMID: 34922924.

18: Orenstein N, Gofin Y, Shomron N, Ruhrman-Shahar N, Magal N, Hagari O, Azulay N, Bazak L, Goldberg Y, **Basel-Salmon L**. DYRK1B haploinsufficiency in a family with metabolic syndrome and abnormal cognition. Clin Genet. 2022 Feb;101(2):265-266. doi: 10.1111/cge.14084. Epub 2021 Nov 16. PMID: 34786696.

19: Maya I, Salzer Sheelo L, Brabbing-Goldstein D, Matar R, Kahana S, Agmon-Fishman I, Klein C, Gurevitch M, **Basel-Salmon L**, Sagi-Dain L. Residual risk for clinically significant copy number variants in low-risk pregnancies, following exclusion of noninvasive prenatal screening-detectable findings. Am J Obstet Gynecol. 2022 Apr;226(4):562.e1-562.e8. doi: 10.1016/j.ajog.2021.11.016. Epub 2021 Nov 8. PMID: 34762861.

20: Ruhrman-Shahar N, Assia Batzir N, Lidzbarsky GA, Bazak L, Magal N, **Basel-Salmon L**. A nonsense variant in the second exon of the canonical transcript of NSD1 does not cause Sotos syndrome. Am J Med Genet A. 2022 Jan;188(1):369-372. doi: 10.1002/ajmg.a.62519. Epub 2021 Sep 24. PMID: 34559457.

21: Kedar I, Walsh L, Levi GR, Lieberman S, Shtaya AA, Nathan SN, Lagovsky I, Tomashov-Matar R, Goldenberg M, **Basel-Salmon L**, Katz L, Aleme O, Peretz TY, Hubert A, Rothstein D, Castellvi-Bel S, Walsh T, King MC, Pritchard CC, Levi Z, Half E, Laish I, Goldberg Y. A novel founder MSH2 deletion in Ethiopian Jews is mainly associated with early-onset colorectal cancer. Fam Cancer. 2022 Apr;21(2):181-188. doi: 10.1007/s10689-021-00249-x. Epub 2021 Apr 10. PMID: 33837488.

22: Wilf-Yarkoni A, Shor O, Fellner A, Hellmann MA, Pras E, Yonath H, Shkedi-Rafid S, **Basel-Salmon L**, Bazak L, Eliahou R, Greenbaum L, Stiebel-Kalish H, Benninger F, Goldberg Y. Mild Phenotype of Wolfram Syndrome Associated With a Common Pathogenic Variant Is Predicted by a Structural Model of Wolframin. Neurol Genet. 2021 Mar 19;7(2):e578. doi: 10.1212/NXG.0000000000000578. PMID: 33763535; PMCID: PMC7983365.

23: Maya I, **Basel-Salmon L**, Sagi-Dain L. Is it time to report carrier state for recessive disorders in every microarray analysis?-A pilot model based on hearing loss genes deletions. Eur J Hum Genet. 2021 Aug;29(8):1292-1300. doi: 10.1038/s41431-021-00856-3. Epub 2021 Mar 22. PMID: 33753912; PMCID: PMC8384849.

24: Sukenik-Halevy R, Ruhrman-Shahar N, Orenstein N, Gonzaga-Jauregui C, Shuldiner AR, Magal N, Hagari O, Azulay N, Lidzbarsky GA, Bazak L, **Basel-Salmon L**. The diagnostic efficacy of exome data analysis using fixed neurodevelopmental gene lists: Implications for prenatal setting. Prenat Diagn. 2021 May;41(6):701-707. doi: 10.1002/pd.5929. Epub 2021 Mar 31. PMID: 33686681.

25: Maya I, Kahana S, Agmon-Fishman I, Klein C, Matar R, Berger R, Josefsberg SBY, Shohat M, Marom D, **Basel-Salmon L**, Sagi-Dain L. The phenotype of 15 cases with rare 8q24.13-q24.3 deletions-A new syndrome or still an enigma? Am J Med Genet A. 2021 May;185(5):1461-1467. doi: 10.1002/ajmg.a.62131. Epub 2021 Feb 22. PMID: 33619900.

26: Shao DD, Straussberg R, Ahmed H, Khan A, Tian S, Hill RS, Smith RS, Majmundar AJ, Ameziane N, Neil JE, Yang E, Al Tenaiji A, Jamuar SS, Schlaeger TM, Al-Saffar M, Hovel I, Al-Shamsi A, **Basel-Salmon L**, Amir AZ, Rento LM, Lim JY, Ganesan I, Shril S, Evrony G, Barkovich AJ, Bauer P, Hildebrandt F, Dong M, Borck G, Beetz C, Al-Gazali L, Eyaid W, Walsh CA. A recurrent, homozygous EMC10 frameshift variant is associated with a syndrome of developmental delay with variable seizures and dysmorphic features. Genet Med. 2021 Jun;23(6):1158-1162. doi: 10.1038/s41436-021-01097-x. Epub 2021 Feb 2. PMID: 33531666; PMCID:

PMC8187145.

27: Fellner A, Lossos A, Kogan E, Argov Z, Gonzaga-Jauregui C, Shuldiner AR, Darawshe M, Bazak L, Lidzbarsky G, Shomron N, **Basel-Salmon L**, Goldberg Y. Two intronic cis-acting variants in both alleles of the POLR3A gene cause progressive spastic ataxia with hypodontia. Clin Genet. 2021 May;99(5):713-718. doi: 10.1111/cge.13929. Epub 2021 Feb 15. PMID: 33491183.

28: Fellner A, Ruhrman-Shahar N, Orenstein N, Lidzbarsky G, Shuldiner AR, Gonzaga-Jauregui C, Brown-Shalev H, Hagari-Bechar O, Bazak L, **Basel-Salmon L**. The role of phenotype-based search approaches using public online databases in diagnostics of Mendelian disorders. Genet Med. 2021 Jun;23(6):1095-1100. doi: 10.1038/s41436-020-01085-7. Epub 2021 Jan 20. PMID: 33473205.

29: Stern T, Orenstein N, Fellner A, Lev-El Halabi N, Shuldiner AR, Gonzaga-Jauregui C, Lidzbarsky G, **Basel-Salmon L**, Goldberg-Stern H. Epilepsy and electroencephalogram evolution in YWHAG gene mutation: A new phenotype and review of the literature. Am J Med Genet A. 2021 Mar;185(3):901-908. doi: 10.1002/ajmg.a.62026. Epub 2021 Jan 4. PMID: 33393734.

30: **Basel-Salmon L**, Ruhrman-Shahar N, Barel O, Hagari O, Marek-Yagel D, Azulai N, Bazak L, Svirsky R, Reznik-Wolf H, Lidzbarsky GA, Shohat M. Biallelic variants in ETV2 in a family with congenital heart defects, vertebral abnormalities and preaxial polydactyly. Eur J Med Genet. 2021 Feb;64(2):104124. doi: 10.1016/j.ejmg.2020.104124. Epub 2021 Jan 8. PMID: 33359164.

31: Aharoni S, Nevo Y, Orenstein N, **Basel-Salmon L**, Ben-Shachar S, Mussaffi H, Sagi-Dain L, Cohen R, Singer A. Impact of a national population-based carrier-screening program on spinal muscular atrophy births. Neuromuscul Disord. 2020 Dec;30(12):970-974. doi: 10.1016/j.nmd.2020.10.005. Epub 2020 Oct 20. PMID: 33218846.

32: Brownstein Z, Gulsuner S, Walsh T, Martins FTA, Taiber S, Isakov O, Lee MK, Bordeynik-Cohen M, Birkan M, Chang W, Casadei S, Danial-Farran N, Abu-Rayyan A, Carlson R, Kamal L, Arnthórsson AÖ, Sokolov M, Gilony D, Lipschitz N, Frydman M, Davidov B, Macarov M, Sagi M, Vinkler C, Poran H, Sharony R, Samra N, Zvi N, Baris-Feldman H, Singer A, Handzel O, Hertzano R, Ali-Naffaa D, Ruhrman-Shahar N, Madgar O, Sofrin-Drucker E, Peleg A, Khayat M, Shohat M, **Basel-Salmon L**, Pras E, Lev D, Wolf M, Steingrimsson E, Shomron N, Kelley MW, Kanaan MN, Allon-Shalev S, King MC, Avraham KB. Spectrum of genes for inherited hearing loss in the Israeli Jewish population, including the novel human deafness gene ATOH1. Clin Genet. 2020 Oct;98(4):353-364. doi: 10.1111/cge.13817. Epub 2020 Aug 24. PMID: 33111345; PMCID: PMC8045518.

33: Maya I, Sukenik-Halevy R, **Basel-Salmon L**, Sagi-Dain L. Ten points to consider when providing genetic counseling for variants of incomplete penetrance and variable expressivity detected in a prenatal setting. Acta Obstet Gynecol Scand. 2020 Nov;99(11):1427-1429. doi: 10.1111/aogs.13963. PMID: 33084292.

34: Maya I, Smirin-Yosef P, Kahana S, Morag S, Yacobson S, Agmon-Fishman I, Matar R, Bitton E, Shohat M, **Basel-Salmon L**, Salmon-Divon M. A study of normal copy number variations in Israeli population. Hum Genet. 2021 Mar;140(3):553-563. doi: 10.1007/s00439-020-02225-4. Epub 2020 Sep 27. PMID: 32980975.

36: **Basel-Salmon L**, Ruhrman-Shahar N, Orenstein N, Goldberg Y, Gonzaga-Jauregui C, Shuldiner AR, Sukenik-Halevy R, Maya I, Magal N, Hagari O, Azulay N, Lidzbarsky GA, Bazak L. When phenotype does not match genotype: importance of "real-time" refining of phenotypic information for exome data interpretation. Genet Med. 2021 Jan;23(1):215-221. doi: 10.1038/s41436-020-00938-5. Epub 2020 Aug 17. PMID: 32801363.

37: Maya I, Perlman S, Shohat M, Kahana S, Yacobson S, Tenne T, Agmon-Fishman I, Tomashov Matar R, **Basel-Salmon L**, Sukenik-Halevy R. Should We Report 15q11.2

BP1-BP2 Deletions and Duplications in the Prenatal Setting? J Clin Med. 2020 Aug 11;9(8):2602. doi: 10.3390/jcm9082602. PMID: 32796639; PMCID: PMC7463673.

38: Maya I, Kahana S, Agmon-Fishman I, Klein C, Matar R, Berger R, Shohat M, **Basel-Salmon L**, Sharony R, Sagi-Dain L. Based on a cohort of 52,879 microarrays, recurrent intragenic FBN2 deletion encompassing exons 1-8 does not cause Beals syndrome. Eur J Med Genet. 2020 Oct;63(10):104008. doi: 10.1016/j.ejmg.2020.104008. Epub 2020 Jul 21. PMID: 32702406.

39: Michaelson-Cohen R, Salzer-Sheelo L, Sukenik-Halevy R, Koifman A, Fellner A, Reches A, Marom D, Behar DM, Sofrin-Drucker E, Zaks-Hoffer G, Weiss-Hubshmann M, Oresntein N, Kropach-Gilad N, Rhurman-Shahar N, Averbuch NS, Magal N, Bazak L, Josefberg S, Matar R, Goldberg Y, Shohat M, **Basel-Salmon L**, Maya I. Teaching clinicians practical genomic medicine: 7 years' experience in a tertiary care center. Genet Med. 2020 Oct;22(10):1703-1709. doi: 10.1038/s41436-020-0868-4. Epub 2020 Jul 3. PMID: 32616942.

40: Maya I, **Basel-Salmon L**, Singer A, Sagi-Dain L. High-frequency low-penetrance copy-number variant classification: should we revise the existing guidelines? Genet Med. 2020 Jul;22(7):1276-1277. doi: 10.1038/s41436-020-0795-4. Epub 2020 Apr 28. PMID: 32341574.

41: Tan TY, Sedmík J, Fitzgerald MP, Halevy RS, Keegan LP, Helbig I, **Basel-Salmon L**, Cohen L, Straussberg R, Chung WK, Helal M, Maroofian R, Houlden H, Juusola J, Sadedin S, Pais L, Howell KB, White SM, Christodoulou J, O'Connell MA. Bi-allelic ADARB1 Variants Associated with Microcephaly, Intellectual Disability, and Seizures. Am J Hum Genet. 2020 Apr 2;106(4):467-483. doi: 10.1016/j.ajhg.2020.02.015. Epub 2020 Mar 26. PMID: 32220291; PMCID: PMC7118584.

42: Mak CCY, Doherty D, Lin AE, Vegas N, Cho MT, Viot G, Dimartino C, Weisfeld-Adams JD, Lessel D, Joss S, Li C, Gonzaga-Jauregui C, Zarate YA, Ehmke N, Horn D, Troyer C, Kant SG, Lee Y, Ishak GE, Leung G, Barone Pritchard A, Yang S, Bend

EG, Filippini F, Roadhouse C, Lebrun N, Mehaffey MG, Martin PM, Apple B, Millan F, Puk O, Hoffer MJV, Henderson LB, McGowan R, Wentzensen IM, Pei S, Zahir FR, Yu M, Gibson WT, Seman A, Steeves M, Murrell JR, Luettgen S, Francisco E, Strom TM, Amlie-Wolf L, Kaindl AM, Wilson WG, Halbach S, **Basel-Salmon L**, Lev-El N, Denecke J, Vissers LELM, Radtke K, Chelly J, Zackai E, Friedman JM, Bamshad MJ, Nickerson DA; University of Washington Center for Mendelian Genomics; Reid RR, Devriendt K, Chae JH, Stolerman E, McDougall C, Powis Z, Bienvenu T, Tan TY, Orenstein N, Dobyns WB, Shieh JT, Choi M, Waggoner D, Gripp KW, Parker MJ, Stoler J, Lyonnet S, Cormier-Daire V, Viskochil D, Hoffman TL, Amiel J, Chung BHY, Gordon CT. MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain. 2020 Jan 1;143(1):55-68. doi: 10.1093/brain/awz379. Erratum in: Brain. 2020 Mar 1;143(3):e24. PMID: 31834374; PMCID: PMC7962909.

43: Li L, Ghorbani M, Weisz-Hubshman M, Rousseau J, Thiffault I, Schnur RE, Breen C, Oegema R, Weiss MM, Waisfisz Q, Welner S, Kingston H, Hills JA, Boon EM, **Basel-Salmon L**, Konen O, Goldberg-Stern H, Bazak L, Tzur S, Jin J, Bi X, Bruccoleri M, McWalter K, Cho MT, Scarano M, Schaefer GB, Brooks SS, Hughes SS, van Gassen KLI, van Hagen JM, Pandita TK, Agrawal PB, Campeau PM, Yang XJ. Lysine acetyltransferase 8 is involved in cerebral development and syndromic intellectual disability. J Clin Invest. 2020 Mar 2;130(3):1431-1445. doi: 10.1172/JCI131145. PMID: 31794431; PMCID: PMC7269600.

44: Horn S, Au M, **Basel-Salmon L**, Bayrak-Toydemir P, Chapin A, Cohen L, Elting MW, Graham JM, Gonzaga-Jauregui C, Konen O, Holzer M, Lemke J, Miller CE, Rey LK, Wolf NI, Weiss MM, Waisfisz Q, Mirzaa GM, Wieczorek D, Sticht H, Abou Jamra R. De novo variants in PAK1 lead to intellectual disability with macrocephaly and seizures. Brain. 2019 Nov 1;142(11):3351-3359. doi: 10.1093/brain/awz264. PMID: 31504246; PMCID: PMC6821231.

45: Toledano H, Orenstein N, Sofrin E, Ruhrman-Shahar N, Amarilyo G, **Basel-Salmon L**, Shuldiner AR, Smirin-Yosef P, Aronson M, Al-Tarrah H, Bazak L,

Gonzaga-Jauregui C, Tabori U, Wimmer K, Goldberg Y. Paediatric systemic lupus erythematosus as a manifestation of constitutional mismatch repair deficiency. J Med Genet. 2020 Jul;57(7):505-508. doi: 10.1136/jmedgenet-2019-106303. Epub 2019 Sep 9. PMID: 31501241.

46: Magini P, Smits DJ, Vandervore L, Schot R, Columbaro M, Kasteleijn E, van der Ent M, Palombo F, Lequin MH, Dremmen M, de Wit MCY, Severino M, Divizia MT, Striano P, Ordonez-Herrera N, Alhashem A, Al Fares A, Al Ghamdi M, Rolfs A, Bauer P, Demmers J, Verheijen FW, Wilke M, van Slegtenhorst M, van der Spek PJ, Seri M, Jansen AC, Stottmann RW, Hufnagel RB, Hopkin RJ, Aljeaid D, Wiszniewski W, Gawlinski P, Laure-Kamionowska M, Alkuraya FS, Akleh H, Stanley V, Musaev D, Gleeson JG, Zaki MS, Brunetti-Pierri N, Cappuccio G, Davidov B, **Basel-Salmon L**, Bazak L, Shahar NR, Bertoli-Avella A, Mirzaa GM, Dobyns WB, Pippucci T, Fornerod M, Mancini GMS. Loss of SMPD4 Causes a Developmental Disorder Characterized by Microcephaly and Congenital Arthrogryposis. Am J Hum Genet. 2019 Oct 3;105(4):689-705. doi: 10.1016/j.ajhg.2019.08.006. Epub 2019 Sep 5. PMID: 31495489; PMCID: PMC6817560.

47: Bend R, Cohen L, Carter MT, Lyons MJ, Niyazov D, Mikati MA, Rojas SK, Person RE, Si Y, Wentzensen IM; Regeneron Genetics Center; Torti E, Lee JA, Boycott KM, **Basel-Salmon L**, Ferreira CR, Gonzaga-Jauregui C. Phenotype and mutation expansion of the PTPN23 associated disorder characterized by neurodevelopmental delay and structural brain abnormalities. Eur J Hum Genet. 2020 Jan;28(1):76-87. doi: 10.1038/s41431-019-0487-1. Epub 2019 Aug 8. PMID: 31395947; PMCID: PMC6906308.

48: Bernstein-Molho R, Barnes-Kedar I, Ludman MD, Reznik G, Feldman HB, Samra NN, Eilat A, Peretz T, Peretz LP, Shapira T, Magal N, Kalis ML, Yerushalmi R, Vinkler C, Liberman S, **Basel-Salmon L**, Shohat M, Levy-Lahad E, Friedman E, Bazak L, Goldberg Y. The yield of full BRCA1/2 genotyping in Israeli Arab high-risk breast/ovarian cancer patients. Breast Cancer Res Treat. 2019 Nov;178(1):231-237. doi: 10.1007/s10549-019-05379-6. Epub 2019 Jul 31. PMID:

31368036.

49: Sagi-Dain L, Goldberg Y, Peleg A, Sukenik-Halevy R, Sofrin-Drucker E, Appelman Z, Josefsberg BYS, Ben-Shachar S, Vinkler C, **Basel-Salmon L**, Maya I. The rare 13q33-q34 microdeletions: eight new patients and review of the literature. Hum Genet. 2019 Oct;138(10):1145-1153. doi: 10.1007/s00439-019-02048-y. Epub 2019 Jul 18. PMID: 31321490.

50: Lieberman S, Beeri R, Walsh T, Schechter M, Keret D, Half E, Gulsuner S, Tomer A, Jacob H, Cohen S, **Basel-Salmon L**, Mansur M, Berger R, Katz LH, Golomb E, Peretz T, Levy Z, Kedar I, King MC, Levy-Lahad E, Goldberg Y. Variable Features of Juvenile Polyposis Syndrome With Gastric Involvement Among Patients With a Large Genomic Deletion of BMPR1A. Clin Transl Gastroenterol. 2019 Jul;10(7):e00054. doi: 10.14309/ctg.0000000000000054. PMID: 31259752; PMCID: PMC6708668.

51: Shen J, Oza AM, Del Castillo I, Duzkale H, Matsunaga T, Pandya A, Kang HP, Mar-Heyming R, Guha S, Moyer K, Lo C, Kenna M, Alexander JJ, Zhang Y, Hirsch Y, Luo M, Cao Y, Wai Choy K, Cheng YF, Avraham KB, Hu X, Garrido G, Moreno-Pelayo MA, Greinwald J, Zhang K, Zeng Y, Brownstein Z, **Basel-Salmon L**, Davidov B, Frydman M, Weiden T, Nagan N, Willis A, Hemphill SE, Grant AR, Siegert RK, DiStefano MT, Amr SS, Rehm HL, Abou Tayoun AN; ClinGen Hearing Loss Working Group. Consensus interpretation of the p.Met34Thr and p.Val37Ile variants in GJB2 by the ClinGen Hearing Loss Expert Panel. Genet Med. 2019 Nov;21(11):2442-2452. doi: 10.1038/s41436-019-0535-9. Epub 2019 Jun 4. PMID: 31160754; PMCID: PMC7235630.

52: Sagi-Dain L, Cohen Vig L, Kahana S, Yacobson S, Tenne T, Agmon-Fishman I, Klein C, Matar R, **Basel-Salmon L**, Maya I. Chromosomal microarray vs. NIPS: analysis of 5541 low-risk pregnancies. Genet Med. 2019 Nov;21(11):2462-2467. doi: 10.1038/s41436-019-0550-x. Epub 2019 May 24. PMID: 31123319.

53: Paperna T, Sharon-Shwartzman N, Kurolap A, Goldberg Y, Moustafa N, Carasso Y, Feinstien M, Mory A, Reznick-Levi G, Gonzaga-Jauregui C, Shuldiner AR, **Basel-Salmon L**, Ofran Y, Half EE, Baris Feldman H. Homozygosity for CHEK2 p.Gly167Arg leads to a unique cancer syndrome with multiple complex chromosomal translocations in peripheral blood karyotype. J Med Genet. 2020 Jul;57(7):500-504. doi: 10.1136/jmedgenet-2018-105824. Epub 2019 Mar 11. PMID: 30858171.

54: Weisz-Hubshman M, Meirson H, Michaelson-Cohen R, Beeri R, Tzur S, Bormans C, Modai S, Shomron N, Shilon Y, Banne E, Orenstein N, Konen O, Marek-Yagel D, Veber A, Shalva N, Imagawa E, Matsumoto N, Lev D, Lerman Sagie T, Raas-Rothschild A, Ben-Zeev B, **Basel-Salmon L**, Behar DM, Heimer G. Novel WWOX deleterious variants cause early infantile epileptic encephalopathy, severe developmental delay and dysmorphism among Yemenite Jews. Eur J Paediatr Neurol. 2019 May;23(3):418-426. doi: 10.1016/j.ejpn.2019.02.003. Epub 2019 Feb 19. PMID: 30853297.

55: Rabinowitz T, Polsky A, Golan D, Danilevsky A, Shapira G, Raff C, **Basel-Salmon L**, Matar RT, Shomron N. Bayesian-based noninvasive prenatal diagnosis of single-gene disorders. Genome Res. 2019 Mar;29(3):428-438. doi: 10.1101/gr.235796.118. Epub 2019 Feb 20. PMID: 30787035; PMCID: PMC6396420.

56: Gurovich Y, Hanani Y, Bar O, Nadav G, Fleischer N, Gelbman D, **Basel-Salmon L**, Krawitz PM, Kamphausen SB, Zenker M, Bird LM, Gripp KW. Identifying facial phenotypes of genetic disorders using deep learning. Nat Med. 2019 Jan;25(1):60-64. doi: 10.1038/s41591-018-0279-0. Epub 2019 Jan 7. PMID: 30617323.

57: **Basel-Salmon L**, Orenstein N, Markus-Bustani K, Ruhrman-Shahar N, Kilim Y, Magal N, Hubshman MW, Bazak L. Improved diagnostics by exome sequencing following raw data reevaluation by clinical geneticists involved in the medical care of the individuals tested. Genet Med. 2019 Jun;21(6):1443-1451. doi:

10.1038/s41436-018-0343-7. Epub 2018 Oct 31. PMID: 30377382.

58: Shohet A, Cohen L, Haguel D, Mozer Y, Shomron N, Tzur S, Bazak L, **Basel Salmon L**, Krause I. Variant in SCYL1 gene causes aberrant splicing in a family with cerebellar ataxia, recurrent episodes of liver failure, and growth retardation. Eur J Hum Genet. 2019 Feb;27(2):263-268. doi: 10.1038/s41431-018-0268-2. Epub 2018 Sep 26. PMID: 30258122; PMCID: PMC6336772.

59: Hemati P, Revah-Politi A, Bassan H, Petrovski S, Bilancia CG, Ramsey K, Griffin NG, Bier L, Cho MT, Rosello M, Lynch SA, Colombo S, Weber A, Haug M, Heinzen EL, Sands TT, Narayanan V, Primiano M, Aggarwal VS, Millan F, Sattler-Holtrop SG, Caro-Llopis A, Pillar N, Baker J, Freedman R, Kroes HY, Sacharow S, Stong N, Lapunzina P, Schneider MC, Mendelsohn NJ, Singleton A, Loik Ramey V, Wou K, Kuzminsky A, Monfort S, Weiss M, Doyle S, Iglesias A, Martinez F, Mckenzie F, Orellana C, van Gassen KLI, Palomares M, Bazak L, Lee A, Bircher A, **Basel-Vanagaite L**, Hafström M, Houge G; C4RCD Research Group; DDD study; Goldstein DB, Anyane-Yeboa K. Refining the phenotype associated with GNB1 mutations: Clinical data on 18 newly identified patients and review of the literature. Am J Med Genet A. 2018 Nov;176(11):2259-2275. doi: 10.1002/ajmg.a.40472. Epub 2018 Sep 8. PMID: 30194818.

60: Barnes-Kedar I, Bernstein-Molho R, Ginzach N, Hartmajer S, Shapira T, Magal N, Kalis ML, Peretz T, Shohat M, **Basel-Salmon L**, Friedman E, Bazak L, Goldberg Y. The yield of full BRCA1/2 genotyping in Israeli high-risk breast/ovarian cancer patients who do not carry the predominant mutations. Breast Cancer Res Treat. 2018 Nov;172(1):151-157. doi: 10.1007/s10549-018-4887-7. Epub 2018 Jul 16. PMID: 30014164.

61: Orenstein N, Goldberg-Stern H, Straussberg R, Bazak L, Weisz Hubshman M, Kropach N, Gilad O, Scheuerman O, Dory Y, Kraus D, Tzur S, Magal N, Kilim Y, Shkalim Zemer V, **Basel-Salmon L**. A de novo GABRA2 missense mutation in severe early-onset epileptic encephalopathy with a choreiform movement disorder. Eur J

Paediatr Neurol. 2018 May;22(3):516-524. doi: 10.1016/j.ejpn.2017.12.017. Epub 2017 Dec 30. PMID: 29422393.

62: Weisz Hubshman M, Broekman S, van Wijk E, Cremers F, Abu-Diab A, Khateb S, Tzur S, Lagovsky I, Smirin-Yosef P, Sharon D, Haer-Wigman L, Banin E, **Basel-Vanagaite L**, de Vrieze E. Whole-exome sequencing reveals POC5 as a novel gene associated with autosomal recessive retinitis pigmentosa. Hum Mol Genet. 2018 Feb 15;27(4):614-624. doi: 10.1093/hmg/ddx428. PMID: 29272404.

63: Abbott JA, Meyer-Schuman R, Lupo V, Feely S, Mademan I, Oprescu SN, Griffin LB, Alberti MA, Casasnovas C, Aharoni S, **Basel-Vanagaite L**, Züchner S, De Jonghe P, Baets J, Shy ME, Espinós C, Demeler B, Antonellis A, Francklyn C. Substrate interaction defects in histidyl-tRNA synthetase linked to dominant axonal peripheral neuropathy. Hum Mutat. 2018 Mar;39(3):415-432. doi: 10.1002/humu.23380. Epub 2017 Dec 26. PMID: 29235198; PMCID: PMC5983030.

64: **Basel-Vanagaite L**, Pillar N, Isakov O, Smirin-Yosef P, Lagovsky I, Orenstein N, Salmon-Divon M, Tamary H, Zaft T, Bazak L, Meyerovitch J, Pelli T, Botchan S, Farberov L, Weissglas-Volkov D, Shomron N. Corrigendum to "X-linked elliptocytosis with impaired growth is related to mutated AMMECR1" [Gene 606C (2017) 47-52]. Gene. 2018 Feb 20;644:155. doi: 10.1016/j.gene.2017.11.051. Epub 2017 Nov 27. Erratum for: Gene. 2017 Mar 30;606:47-52. PMID: 29174631.

65: Yilmaz R, Szakszon K, Altmann A, Altunoglu U, Senturk L, McGuire M, Calabrese O, Madan-Khetarpal S, **Basel-Vanagaite L**, Borck G. Kaufman oculocerebrofacial syndrome: Novel UBE3B mutations and clinical features in four unrelated patients. Am J Med Genet A. 2018 Jan;176(1):187-193. doi: 10.1002/ajmg.a.38538. Epub 2017 Nov 21. PMID: 29160006.

66: Cuckle H. Re: Cut-off value of nuchal translucency as indication for chromosomal microarray analysis. I. Maya, S. Yacobson, S. Kahana, J. Yeshaya, T. Tenne, I. Agmon-Fishman, L. Cohen-Vig, M. Shohat, **L. Basel-Vanagaite** and R.

Sharony. Ultrasound Obstet Gynecol 2017; 50: 332-335. Ultrasound Obstet Gynecol. 2017 Sep;50(3):293-294. doi: 10.1002/uog.18814. PMID: 28938059.

67: Bardin R, Hadar E, Haizler-Cohen L, Gabbay-Benziv R, Meizner I, Kahana S, Yeshaya J, Yacobson S, Cohen-Vig L, Agmon-Fishman I, **Basel-Vanagaite L**, Maya I. Cytogenetic analysis in fetuses with late onset abnormal sonographic findings. J Perinat Med. 2018 Nov 27;46(9):975-982. doi: 10.1515/jpm-2017-0071. PMID: 28915119.

68: Maya I, Sharony R, Yacobson S, Kahana S, Yeshaya J, Tenne T, Agmon-Fishman I, Cohen-Vig L, Goldberg Y, Berger R, **Basel-Salmon L**, Shohat M. When genotype is not predictive of phenotype: implications for genetic counseling based on 21,594 chromosomal microarray analysis examinations. Genet Med. 2018 Jan;20(1):128-131. doi: 10.1038/gim.2017.89. Epub 2017 Jul 20. PMID: 28726807.

69: Solomon-Zemler R, **Basel-Vanagaite L**, Steier D, Yakar S, Mel E, Phillip M, Bazak L, Bercovich D, Werner H, de Vries L. A novel heterozygous IGF-1 receptor mutation associated with hypoglycemia. Endocr Connect. 2017 Aug;6(6):395-403. doi: 10.1530/EC-17-0038. Epub 2017 Jun 25. PMID: 28649085; PMCID: PMC5551424.

70: Cohen L, Orenstein N, Weisz-Hubshman M, Bazak L, Davidov B, Reinstein E, Tzur S, Behar D, Smirin-Yosef P, Salmon-Divon M, Gross A, Shohat M, **Basel-Vanagaite L**. [UTILIZATION OF WHOLE EXOME SEQUENCING IN DIAGNOSTICS OF GENETIC DISEASE: RABIN MEDICAL CENTER'S EXPERIENCE]. Harefuah. 2017 Apr;156(4):212-216. Hebrew. PMID: 28551919.

71: Salpietro V, Lin W, Delle Vedove A, Storbeck M, Liu Y, Efthymiou S, Manole A, Wiethoff S, Ye Q, Saggar A, McElreavey K, Krishnakumar SS; SYNAPS Study Group; Pitt M, Bello OD, Rothman JE, **Basel-Vanagaite L**, Hubshman MW, Aharoni S, Manzur AY, Wirth B, Houlden H. Homozygous mutations in VAMP1 cause a presynaptic congenital myasthenic syndrome. Ann Neurol. 2017 Apr;81(4):597-603. doi: 10.1002/ana.24905. Epub 2017 Mar 29. PMID: 28253535; PMCID: PMC5413866.

72: Lee JYW, Hsu CK, Michael M, Nanda A, Liu L, McMillan JR, Pourreyron C, Takeichi T, Tolar J, Reid E, Hayday T, Blumen SC, Abu-Mouch S, Straussberg R, **Basel-Vanagaite L**, Barhum Y, Zouabi Y, Al-Ajmi H, Huang HY, Lin TC, Akiyama M, Lee JYY, McLean WHI, Simpson MA, Parsons M, McGrath JA. Large Intragenic Deletion in DSTYK Underlies Autosomal-Recessive Complicated Spastic Paraparesis, SPG23. Am J Hum Genet. 2017 Feb 2;100(2):364-370. doi: 10.1016/j.ajhg.2017.01.014. PMID: 28157540; PMCID: PMC5294675.

73: Maya I, Yacobson S, Kahana S, Yeshaya J, Tenne T, Agmon-Fishman I, Cohen-Vig L, Shohat M, **Basel-Vanagaite L**, Sharony R. Cut-off value of nuchal translucency as indication for chromosomal microarray analysis. Ultrasound Obstet Gynecol. 2017 Sep;50(3):332-335. doi: 10.1002/uog.17421. Epub 2017 Jul 26. PMID: 28133835.

74: **Basel-Vanagaite L**, Pillar N, Isakov O, Smirin-Yosef P, Lagovsky I, Orenstein N, Salmon-Divon M, Tamary H, Zaft T, Bazak L, Meyerovitch J, Pelli T, Botchan S, Farberov L, Weissglas-Volkov D, Shomron N. X-linked elliptocytosis with impaired growth is related to mutated AMMECR1. Gene. 2017 Mar 30;606:47-52. doi: 10.1016/j.gene.2017.01.001. Epub 2017 Jan 9. Erratum in: Gene. 2017 Nov 23;: PMID: 28089922.

75: Smirin-Yosef P, Zuckerman-Levin N, Tzur S, Granot Y, Cohen L, Sachsenweger J, Borck G, Lagovsky I, Salmon-Divon M, Wiesmüller L, **Basel-Vanagaite L**. A Biallelic Mutation in the Homologous Recombination Repair Gene SPIDR Is Associated With Human Gonadal Dysgenesis. J Clin Endocrinol Metab. 2017 Feb 1;102(2):681-688. doi: 10.1210/jc.2016-2714. Erratum in: J Clin Endocrinol Metab. 2018 Jan 1;103(1):364. PMID: 27967308.

76: Weisz Hubshman M, **Basel-Vanagaite L**, Krauss A, Konen O, Levy Y, Garty BZ, Smirin-Yosef P, Maya I, Lagovsky I, Taub E, Marom D, Gaash D, Shichrur K, Avigad S, Hayman-Manzur L, Villa A, Sobacchi C, Shohat M, Yaniv I, Stein J. Homozygous

deletion of RAG1, RAG2 and 5' region TRAF6 causes severe immune suppression and atypical osteopetrosis. Clin Genet. 2017 Jun;91(6):902-907. doi: 10.1111/cge.12916. Epub 2017 Mar 19. PMID: 27808398.

77: Cohen R, Halevy A, Aharoni S, Kraus D, Konen O, **Basel-Vanagaite L**, Goldberg-Stern H, Straussberg R. Polymicrogyria and myoclonic epilepsy in autosomal recessive cutis laxa type 2A. Neurogenetics. 2016 Oct;17(4):251-257. doi: 10.1007/s10048-016-0491-3. Epub 2016 Sep 8. PMID: 27631729.

78: Weiss K, Terhal PA, Cohen L, Bruccoleri M, Irving M, Martinez AF, Rosenfeld JA, Machol K, Yang Y, Liu P, Walkiewicz M, Beuten J, Gomez-Ospina N, Haude K, Fong CT, Enns GM, Bernstein JA, Fan J, Gotway G, Ghorbani M; DDD Study; van Gassen K, Monroe GR, van Haaften G, **Basel-Vanagaite L**, Yang XJ, Campeau PM, Muenke M. De Novo Mutations in CHD4, an ATP-Dependent Chromatin Remodeler Gene, Cause an Intellectual Disability Syndrome with Distinctive Dysmorphisms. Am J Hum Genet. 2016 Oct 6;99(4):934-941. doi: 10.1016/j.ajhg.2016.08.001. Epub 2016 Sep 8. PMID: 27616479; PMCID: PMC5065651.

79: Maya I, Kahana S, Yeshaya J, Tenne T, Yacobson S, Agmon-Fishman I, Cohen-Vig L, Levi A, Reinstein E, **Basel-Vanagaite L**, Sharony R. Chromosomal microarray analysis in fetuses with aberrant right subclavian artery. Ultrasound Obstet Gynecol. 2017 Mar;49(3):337-341. doi: 10.1002/uog.15935. PMID: 27063194.

80: Eskin-Schwartz M, **Basel-Vanagaite L**, David M, Lagovsky I, Ben-Amitai D, Smirin-Yosef P, Atzmony L, Hodak E. Intra-familial Variation in Clinical Phenotype of CARD14-related Psoriasis. Acta Derm Venereol. 2016 Nov 2;96(7):885-887. doi: 10.2340/00015555-2405. PMID: 26984337.

81: **Basel-Vanagaite L**, Wolf L, Orin M, Larizza L, Gervasini C, Krantz ID, Deardoff MA. Recognition of the Cornelia de Lange syndrome phenotype with facial dysmorphology novel analysis. Clin Genet. 2016 May;89(5):557-63. doi: 10.1111/cge.12716. Epub 2016 Jan 25. PMID: 26663098.

82: Reinstein E, Smirin-Yosef P, Lagovsky I, Davidov B, Peretz Amit G, Neumann D, Orr-Urtreger A, Ben-Shachar S, **Basel-Vanagaite L**. A founder mutation in ADAMTSL4 causes early-onset bilateral ectopia lentis among Jews of Bukharian origin. Mol Genet Metab. 2016 Jan;117(1):38-41. doi: 10.1016/j.ymgme.2015.11.011. Epub 2015 Nov 26. PMID: 26653794.

83: Kumar R, Corbett MA, Van Bon BW, Gardner A, Woenig JA, Jolly LA, Douglas E, Friend K, Tan C, Van Esch H, Holvoet M, Raynaud M, Field M, Leffler M, Budny B, Wisniewska M, Badura-Stronka M, Latos-Bieleńska A, Batanian J, Rosenfeld JA, **Basel-Vanagaite L**, Jensen C, Bienek M, Froyen G, Ullmann R, Hu H, Love MI, Haas SA, Stankiewicz P, Cheung SW, Baxendale A, Nicholl J, Thompson EM, Haan E, Kalscheuer VM, Gecz J. Increased STAG2 dosage defines a novel cohesinopathy with intellectual disability and behavioral problems. Hum Mol Genet. 2015 Dec 20;24(25):7171-81. doi: 10.1093/hmg/ddv414. Epub 2015 Oct 6. PMID: 26443594.

84: Mimouni-Bloch A, Yeshaya J, Kahana S, Maya I, **Basel-Vanagaite L**. A de-novo interstitial microduplication involving 2p16.1-p15 and mirroring 2p16.1-p15 microdeletion syndrome: Clinical and molecular analysis. Eur J Paediatr Neurol. 2015 Nov;19(6):711-5. doi: 10.1016/j.ejpn.2015.07.013. Epub 2015 Aug 4. PMID: 26278498.

85: Rojnueangnit K, Xie J, Gomes A, Sharp A, Callens T, Chen Y, Liu Y, Cochran M, Abbott MA, Atkin J, Babovic-Vuksanovic D, Barnett CP, Crenshaw M, Bartholomew DW, **Basel L**, Bellus G, Ben-Shachar S, Bialer MG, Bick D, Blumberg B, Cortes F, David KL, Destree A, Duat-Rodriguez A, Earl D, Escobar L, Eswara M, Ezquieta B, Frayling IM, Frydman M, Gardner K, Gripp KW, Hernández-Chico C, Heyrman K, Ibrahim J, Janssens S, Keena BA, Llano-Rivas I, Leppig K, McDonald M, Misra VK, Mulbury J, Narayanan V, Orenstein N, Galvin-Parton P, Pedro H, Pivnick EK, Powell CM, Randolph L, Raskin S, Rosell J, Rubin K, Seashore M, Schaaf CP, Scheuerle A, Schultz M, Schorry E, Schnur R, Siqveland E, Tkachuk A, Tonsgard J, Upadhyaya M, Verma IC, Wallace S, Williams C, Zackai E, Zonana J, Lazaro C,

Claes K, Korf B, Martin Y, Legius E, Messiaen L. High Incidence of Noonan Syndrome Features Including Short Stature and Pulmonic Stenosis in Patients carrying NF1 Missense Mutations Affecting p.Arg1809: Genotype-Phenotype Correlation. Hum Mutat. 2015 Nov;36(11):1052-63. doi: 10.1002/humu.22832. Epub 2015 Aug 21. PMID: 26178382; PMCID: PMC5049609.

86: **Basel-Vanagaite L**, Smirin-Yosef P, Essakow JL, Tzur S, Lagovsky I, Maya I, Pasmanik-Chor M, Yeheskel A, Konen O, Orenstein N, Weisz Hubshman M, Drasinover V, Magal N, Peretz Amit G, Zalzstein Y, Zeharia A, Shohat M, Straussberg R, Monté D, Salmon-Divon M, Behar DM. Homozygous MED25 mutation implicated in eye-intellectual disability syndrome. Hum Genet. 2015 Jun;134(6):577-87. doi: 10.1007/s00439-015-1541-x. Epub 2015 Mar 20. PMID: 25792360.

87: Masotti A, Uva P, Davis-Keppen L, **Basel-Vanagaite L**, Cohen L, Pisaneschi E, Celluzzi A, Bencivenga P, Fang M, Tian M, Xu X, Cappa M, Dallapiccola B. Keppen-Lubinsky syndrome is caused by mutations in the inwardly rectifying K+ channel encoded by KCNJ6. Am J Hum Genet. 2015 Feb 5;96(2):295-300. doi: 10.1016/j.ajhg.2014.12.011. Epub 2015 Jan 22. PMID: 25620207; PMCID: PMC4320262.

88: Straussberg R, Ganelin-Cohen E, Goldberg-Stern H, Tzur S, Behar DM, Smirin-Yosef P, Salmon-Divon M, **Basel-Vanagaite L**. Lethal neonatal rigidity and multifocal seizure syndrome--report of another family with a BRAT1 mutation. Eur J Paediatr Neurol. 2015 Mar;19(2):240-2. doi: 10.1016/j.ejpn.2014.11.004. Epub 2014 Nov 29. PMID: 25500575.

89: de Vries L, Behar DM, Smirin-Yosef P, Lagovsky I, Tzur S, **Basel-Vanagaite L**. Exome sequencing reveals SYCE1 mutation associated with autosomal recessive primary ovarian insufficiency. J Clin Endocrinol Metab. 2014 Oct;99(10):E2129-32. doi: 10.1210/jc.2014-1268. Epub 2014 Jul 25. PMID: 25062452.

90: Cohen R, **Basel-Vanagaite L**, Goldberg-Stern H, Halevy A, Shuper A, Feingold-

Zadok M, Behar DM, Straussberg R. Two siblings with early infantile myoclonic encephalopathy due to mutation in the gene encoding mitochondrial glutamate/H+ symporter SLC25A22. Eur J Paediatr Neurol. 2014 Nov;18(6):801-5. doi: 10.1016/j.ejpn.2014.06.007. Epub 2014 Jul 5. PMID: 25033742.

91: Borck G, de Vries L, Wu HJ, Smirin-Yosef P, Nürnberg G, Lagovsky I, Ishida LH, Thierry P, Wieczorek D, Nürnberg P, Foley J, Kubisch C, **Basel-Vanagaite L**. Homozygous truncating PTPRF mutation causes athelia. Hum Genet. 2014 Aug;133(8):1041-7. doi: 10.1007/s00439-014-1445-1. Epub 2014 Apr 30. PMID: 24781087.

92: Zanni G, Barresi S, Cohen R, Specchio N, **Basel-Vanagaite L**, Valente EM, Shuper A, Vigevano F, Bertini E. A novel mutation in the endosomal Na+/H+ exchanger NHE6 (SLC9A6) causes Christianson syndrome with electrical status epilepticus during slow-wave sleep (ESES). Epilepsy Res. 2014 May;108(4):811-5. doi: 10.1016/j.eplepsyres.2014.02.009. Epub 2014 Feb 19. PMID: 24630051.

93: **Basel-Vanagaite L**, Yilmaz R, Tang S, Reuter MS, Rahner N, Grange DK, Mortenson M, Koty P, Feenstra H, Farwell Gonzalez KD, Sticht H, Boddaert N, Désir J, Anyane-Yeboa K, Zweier C, Reis A, Kubisch C, Jewett T, Zeng W, Borck G. Expanding the clinical and mutational spectrum of Kaufman oculocerebrofacial syndrome with biallelic UBE3B mutations. Hum Genet. 2014 Jul;133(7):939-49. doi: 10.1007/s00439-014-1436-2. Epub 2014 Mar 11. PMID: 24615390.

94: Piras R, Chiappe F, Torraca IL, Buers I, Usala G, Angius A, Akin MA, **Basel-Vanagaite L**, Benedicenti F, Chiodin E, El Assy O, Feingold-Zadok M, Guibert J, Kamien B, Kasapkara CS, Kiliç E, Boduroğlu K, Kurtoglu S, Manzur AY, Onal EE, Paderi E, Roche CH, Tümer L, Unal S, Utine GE, Zanda G, Zankl A, Zampino G, Crisponi G, Crisponi L, Rutsch F. Expanding the mutational spectrum of CRLF1 in Crisponi/CISS1 syndrome. Hum Mutat. 2014 Apr;35(4):424-33. doi: 10.1002/humu.22522. Epub 2014 Mar 6. PMID: 24488861.

95: Peyrard-Janvid M, Leslie EJ, Kousa YA, Smith TL, Dunnwald M, Magnusson M, Lentz BA, Unneberg P, Fransson I, Koillinen HK, Rautio J, Pegelow M, Karsten A, **Basel-Vanagaite L**, Gordon W, Andersen B, Svensson T, Murray JC, Cornell RA, Kere J, Schutte BC. Dominant mutations in GRHL3 cause Van der Woude Syndrome and disrupt oral periderm development. Am J Hum Genet. 2014 Jan 2;94(1):23-32. doi: 10.1016/j.ajhg.2013.11.009. Epub 2013 Dec 19. PMID: 24360809; PMCID: PMC3882735.

96: Eytan O, Sarig O, Israeli S, Mevorah B, **Basel-Vanagaite L**, Sprecher E. A novel splice-site mutation in the AAGAB gene segregates with hereditary punctate palmoplantar keratoderma and congenital dysplasia of the hip in a large family. Clin Exp Dermatol. 2014 Mar;39(2):182-6. doi: 10.1111/ced.12213. Epub 2013 Dec 2. PMID: 24289292.

97: Behar DM, **Basel-Vanagaite L**, Glaser F, Kaplan M, Tzur S, Magal N, Eidlitz-Markus T, Haimi-Cohen Y, Sarig G, Bormans C, Shohat M, Zeharia A. Identification of a novel mutation in the PNLIP gene in two brothers with congenital pancreatic lipase deficiency. J Lipid Res. 2014 Feb;55(2):307-12. doi: 10.1194/jlr.P041103. Epub 2013 Nov 21. PMID: 24262094; PMCID: PMC3886669.

98: Goldberg-Stern H, Aharoni S, Afawi Z, Bennett O, Appenzeller S, Pendziwiat M, Kuhlenbäumer G, **Basel-Vanagaite L**, Shuper A, Korczyn AD, Helbig I. Broad phenotypic heterogeneity due to a novel SCN1A mutation in a family with genetic epilepsy with febrile seizures plus. J Child Neurol. 2014 Feb;29(2):221-6. doi: 10.1177/0883073813509016. Epub 2013 Nov 20. PMID: 24257433.

99: Hellman-Aharony S, Smirin-Yosef P, Halevy A, Pasmanik-Chor M, Yeheskel A, Har-Zahav A, Maya I, Straussberg R, Dahary D, Haviv A, Shohat M, **Basel-Vanagaite L**. Microcephaly thin corpus callosum intellectual disability syndrome caused by mutated TAF2. Pediatr Neurol. 2013 Dec;49(6):411-416.e1. doi: 10.1016/j.pediatrneurol.2013.07.017. Epub 2013 Sep 29. PMID: 24084144.

100: Koehler K, Malik M, Mahmood S, Gießelmann S, Beetz C, Hennings JC, Huebner

AK, Grahn A, Reunert J, Nürnberg G, Thiele H, Altmüller J, Nürnberg P, Mumtaz R, Babovic-Vuksanovic D, **Basel-Vanagaite L**, Borck G, Brämswig J, Mühlenberg R, Sarda P, Sikiric A, Anyane-Yeboa K, Zeharia A, Ahmad A, Coubes C, Wada Y, Marquardt T, Vanderschaeghe D, Van Schaftingen E, Kurth I, Huebner A, Hübner CA. Mutations in GMPPA cause a glycosylation disorder characterized by intellectual disability and autonomic dysfunction. Am J Hum Genet. 2013 Oct 3;93(4):727-34. doi: 10.1016/j.ajhg.2013.08.002. Epub 2013 Sep 12. PMID: 24035193; PMCID: PMC3791256.

101: **Basel-Vanagaite L**, Hershkovitz T, Heyman E, Raspall-Chaure M, Kakar N, Smirin-Yosef P, Vila-Pueyo M, Kornreich L, Thiele H, Bode H, Lagovsky I, Dahary D, Haviv A, Hubshman MW, Pasmanik-Chor M, Nürnberg P, Gothelf D, Kubisch C, Shohat M, Macaya A, Borck G. Biallelic SZT2 mutations cause infantile encephalopathy with epilepsy and dysmorphic corpus callosum. Am J Hum Genet. 2013 Sep 5;93(3):524-9. doi: 10.1016/j.ajhg.2013.07.005. Epub 2013 Aug 8. PMID: 23932106; PMCID: PMC3769928.

102: Goldenberg-Cohen N, Banin E, Zalzstein Y, Cohen B, Rotenstreich Y, Rizel L, **Basel-Vanagaite L**, Ben-Yosef T. Genetic heterogeneity and consanguinity lead to a "double hit": homozygous mutations of MYO7A and PDE6B in a patient with retinitis pigmentosa. Mol Vis. 2013 Jul 20;19:1565-71. PMID: 23882135; PMCID: PMC3718492.

103: Dyment DA, Smith AC, Alcantara D, Schwartzentruber JA, **Basel-Vanagaite L**, Curry CJ, Temple IK, Reardon W, Mansour S, Haq MR, Gilbert R, Lehmann OJ, Vanstone MR, Beaulieu CL; FORGE Canada Consortium; Majewski J, Bulman DE, O'Driscoll M, Boycott KM, Innes AM. Mutations in PIK3R1 cause SHORT syndrome. Am J Hum Genet. 2013 Jul 11;93(1):158-66. doi: 10.1016/j.ajhg.2013.06.005. Epub 2013 Jun 27. PMID: 23810382; PMCID: PMC3710754.

104: Behar DM, Adler L, **Basel-Vanagaite L**. Severe hypertriglyceridemia in an infant of Arab descent. Isr Med Assoc J. 2013 Jan;15(1):53-4. PMID: 23484243.

105: Handley MT, Morris-Rosendahl DJ, Brown S, Macdonald F, Hardy C, Bem D, Carpanini SM, Borck G, Martorell L, Izzi C, Faravelli F, Accorsi P, Pinelli L, **Basel-Vanagaite L**, Peretz G, Abdel-Salam GM, Zaki MS, Jansen A, Mowat D, Glass I, Stewart H, Mancini G, Lederer D, Roscioli T, Giuliano F, Plomp AS, Rolfs A, Graham JM, Seemanova E, Poo P, García-Cazorla A, Edery P, Jackson IJ, Maher ER, Aligianis IA. Mutation spectrum in RAB3GAP1, RAB3GAP2, and RAB18 and genotype-phenotype correlations in warburg micro syndrome and Martsolf syndrome. Hum Mutat. 2013 May;34(5):686-96. doi: 10.1002/humu.22296. PMID: 23420520.

106: Abu-Rashid M, Mahajnah M, Jaber L, Kornreich L, Bar-On E, **Basel-Vanagaite L**, Soffer D, Koenig M, Straussberg R. A novel mutation in the GAN gene causes an intermediate form of giant axonal neuropathy in an Arab-Israeli family. Eur J Paediatr Neurol. 2013 May;17(3):259-64. doi: 10.1016/j.ejpn.2012.10.012. Epub 2013 Jan 16. PMID: 23332420.

107: Gordon CT, Vuillot A, Marlin S, Gerkes E, Henderson A, AlKindy A, Holder-Espinasse M, Park SS, Omarjee A, Sanchis-Borja M, Bdira EB, Oufadem M, Sikkema-Raddatz B, Stewart A, Palmer R, McGowan R, Petit F, Delobel B, Speicher MR, Aurora P, Kilner D, Pellerin P, Simon M, Bonnefont JP, Tobias ES, García-Miñaúr S, Bitner-Glindzicz M, Lindholm P, Meijer BA, Abadie V, Denoyelle F, Vazquez MP, Rotky-Fast C, Couloigner V, Pierrot S, Manach Y, Breton S, Hendriks YM, Munnich A, Jakobsen L, Kroisel P, Lin A, Kaban LB, **Basel-Vanagaite L**, Wilson L, Cunningham ML, Lyonnet S, Amiel J. Heterogeneity of mutational mechanisms and modes of inheritance in auriculocondylar syndrome. J Med Genet. 2013 Mar;50(3):174-86. doi: 10.1136/jmedgenet-2012-101331. Epub 2013 Jan 12. PMID: 23315542.

108: Michaelovsky E, Frisch A, Carmel M, Patya M, Zarchi O, Green T, **Basel-Vanagaite L**, Weizman A, Gothelf D. Genotype-phenotype correlation in 22q11.2 deletion syndrome. BMC Med Genet. 2012 Dec 17;13:122. doi: 10.1186/1471-2350-13-122. PMID: 23245648; PMCID: PMC3548696.

109: **Basel-Vanagaite L**, Dallapiccola B, Ramirez-Solis R, Segref A, Thiele H, Edwards A, Arends MJ, Miró X, White JK, Désir J, Abramowicz M, Dentici ML, Lepri F, Hofmann K, Har-Zahav A, Ryder E, Karp NA, Estabel J, Gerdin AK, Podrini C, Ingham NJ, Altmüller J, Nürnberg G, Frommolt P, Abdelhak S, Pasmanik-Chor M, Konen O, Kelley RI, Shohat M, Nürnberg P, Flint J, Steel KP, Hoppe T, Kubisch C, Adams DJ, Borck G. Deficiency for the ubiquitin ligase UBE3B in a blepharophimosis-ptosis-intellectual-disability syndrome. Am J Hum Genet. 2012 Dec 7;91(6):998-1010. doi: 10.1016/j.ajhg.2012.10.011. Epub 2012 Nov 29. PMID: 23200864; PMCID: PMC3516591.

110: Borck G, Shin BS, Stiller B, Mimouni-Bloch A, Thiele H, Kim JR, Thakur M, Skinner C, Aschenbach L, Smirin-Yosef P, Har-Zahav A, Nürnberg G, Altmüller J, Frommolt P, Hofmann K, Konen O, Nürnberg P, Munnich A, Schwartz CE, Gothelf D, Colleaux L, Dever TE, Kubisch C, **Basel-Vanagaite L**. eIF2γ mutation that disrupts eIF2 complex integrity links intellectual disability to impaired translation initiation. Mol Cell. 2012 Nov 30;48(4):641-6. doi: 10.1016/j.molcel.2012.09.005. Epub 2012 Oct 11. PMID: 23063529; PMCID: PMC3513554.

111: Halevy A, **Basel-Vanagaite L**, Shuper A, Helman S, Har-Zahav A, Birk E, Maya I, Kornreich L, Inbar D, Nürnberg G, Nürnberg P, Steinberg T, Straussberg R. Microcephaly-thin corpus callosum syndrome maps to 8q23.2-q24.12. Pediatr Neurol. 2012 Jun;46(6):363-8. doi: 10.1016/j.pediatrneurol.2012.03.014. PMID: 22633631.

112: **Basel-Vanagaite L**, Zevit N, Har Zahav A, Guo L, Parathath S, Pasmanik-Chor M, McIntyre AD, Wang J, Albin-Kaplanski A, Hartman C, Marom D, Zeharia A, Badir A, Shoerman O, Simon AJ, Rechavi G, Shohat M, Hegele RA, Fisher EA, Shamir R. Transient infantile hypertriglyceridemia, fatty liver, and hepatic fibrosis caused by mutated GPD1, encoding glycerol-3-phosphate dehydrogenase 1. Am J Hum Genet. 2012 Jan 13;90(1):49-60. doi: 10.1016/j.ajhg.2011.11.028. Epub 2012 Jan

5. PMID: 22226083; PMCID: PMC3257852.

113: Green T, Avda S, Dotan I, Zarchi O, **Basel-Vanagaite L**, Zalsman G, Weizman A, Gothelf D. Phenotypic psychiatric characterization of children with Williams syndrome and response of those with ADHD to methylphenidate treatment. Am J Med Genet B Neuropsychiatr Genet. 2012 Jan;159B(1):13-20. doi: 10.1002/ajmg.b.31247. Epub 2011 Nov 3. PMID: 22052570.

114: **Basel-Vanagaite L**, Sprecher E, Gat A, Merlob P, Albin-Kaplanski A, Konen O, Solomon BD, Muenke M, Grzeschik KH, Sirota L. New syndrome of congenital circumferential skin folds associated with multiple congenital anomalies. Pediatr Dermatol. 2012 Jan-Feb;29(1):89-95. doi: 10.1111/j.1525-1470.2011.01403.x. Epub 2011 Oct 13. PMID: 21995818; PMCID: PMC4131925.

115: Marom D, Albin A, Schwartz C, Har-Zahav A, Straussberg R, Bartel F, Birk E, Inbar D, **Basel-Vanagaite L**. X-linked mental retardation with alacrima and achalasia-Triple A syndrome or a new syndrome? Am J Med Genet A. 2011 Aug;155A(8):1959-63. doi: 10.1002/ajmg.a.34121. Epub 2011 Jul 8. PMID: 21744492.

116: Borck G, Rainshtein L, Hellman-Aharony S, Volk AE, Friedrich K, Taub E, Magal N, Kanaan M, Kubisch C, Shohat M, **Basel-Vanagaite L**. High frequency of autosomal-recessive DFNB59 hearing loss in an isolated Arab population in Israel. Clin Genet. 2012 Sep;82(3):271-6. doi: 10.1111/j.1399-0004.2011.01741.x. Epub 2011 Jul 18. PMID: 21696384.

117: Maydan G, Noyman I, Har-Zahav A, Neriah ZB, Pasmanik-Chor M, Yeheskel A, Albin-Kaplanski A, Maya I, Magal N, Birk E, Simon AJ, Halevy A, Rechavi G, Shohat M, Straussberg R, **Basel-Vanagaite L**. Multiple congenital anomalies-hypotonia-seizures syndrome is caused by a mutation in PIGN. J Med Genet. 2011 Jun;48(6):383-9. doi: 10.1136/jmg.2010.087114. Epub 2011 Apr 14. PMID: 21493957.

118: Shkalim V, Ben-Sira L, Inbar D, Kaadan W, **Basel-Vanagaite L**, Straussberg R. Three sibs with microcephaly, clubfeet and agenesis of corpus callosum: a new genetic syndrome? Am J Med Genet A. 2011 May;155A(5):1060-5. doi: 10.1002/ajmg.a.33978. Epub 2011 Apr 4. PMID: 21465661.

119: Balasubramanian M, Smith K, **Basel-Vanagaite L**, Feingold MF, Brock P, Gowans GC, Vasudevan PC, Cresswell L, Taylor EJ, Harris CJ, Friedman N, Moran R, Feret H, Zackai EH, Theisen A, Rosenfeld JA, Parker MJ. Case series: 2q33.1 microdeletion syndrome--further delineation of the phenotype. J Med Genet. 2011 May;48(5):290-8. doi: 10.1136/jmg.2010.084491. Epub 2011 Feb 22. PMID: 21343628.

120: **Basel-Vanagaite L**, Goldberg-Stern H, Mimouni-Bloch A, Shkalim V, Böhm D, Kohlhase J. An emerging 1q21.1 deletion-associated neurodevelopmental phenotype. J Child Neurol. 2011 Jan;26(1):113-6. doi: 10.1177/0883073810377658. PMID: 21212457.

121: Birk E, Har-Zahav A, Manzini CM, Pasmanik-Chor M, Kornreich L, Walsh CA, Noben-Trauth K, Albin A, Simon AJ, Colleaux L, Morad Y, Rainshtein L, Tischfield DJ, Wang P, Magal N, Maya I, Shoshani N, Rechavi G, Gothelf D, Maydan G, Shohat M, **Basel-Vanagaite L**. SOBP is mutated in syndromic and nonsyndromic intellectual disability and is highly expressed in the brain limbic system. Am J Hum Genet. 2010 Nov 12;87(5):694-700. doi: 10.1016/j.ajhg.2010.10.005. Epub 2010 Oct 28. PMID: 21035105; PMCID: PMC2978971.

122: **Basel-Vanagaite L**, Raas-Rotchild A, Kornreich L, Har-Zahav A, Yeshaya J, Latarowski V, Lerer I, Dobyns WB, Shohat M. Familial hydrocephalus with normal cognition and distinctive radiological features. Am J Med Genet A. 2010 Nov;152A(11):2743-8. doi: 10.1002/ajmg.a.33688. PMID: 20979187.

123: Namavar Y, Barth PG, Kasher PR, van Ruissen F, Brockmann K, Bernert G, Writzl K, Ventura K, Cheng EY, Ferriero DM, **Basel-Vanagaite L**, Eggens VR, Krägeloh-Mann I, De Meirleir L, King M, Graham JM Jr, von Moers A, Knoers N,

Sztriha L, Korinthenberg R; PCH Consortium; Dobyns WB, Baas F, Poll-The BT. Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. Brain. 2011 Jan;134(Pt 1):143-56. doi: 10.1093/brain/awq287. Epub 2010 Oct 15. PMID: 20952379; PMCID: PMC9136852.

124: Zarchi O, Attias J, Raveh E, **Basel-Vanagaite L**, Saporta L, Gothelf D. A comparative study of hearing loss in two microdeletion syndromes: velocardiofacial (22q11.2 deletion) and Williams (7q11.23 deletion) syndromes. J Pediatr. 2011 Feb;158(2):301-6. doi: 10.1016/j.jpeds.2010.07.056. Epub 2010 Sep 16. PMID: 20846670.

125: **Basel-Vanagaite L**, Dobyns WB. Clinical and brain imaging heterogeneity of severe microcephaly. Pediatr Neurol. 2010 Jul;43(1):7-16. doi: 10.1016/j.pediatrneurol.2010.02.015. PMID: 20682196.

126: Sun Y, Almomani R, Aten E, Celli J, van der Heijden J, Venselaar H, Robertson SP, Baroncini A, Franco B, **Basel-Vanagaite L**, Horii E, Drut R, Ariyurek Y, den Dunnen JT, Breuning MH. Terminal osseous dysplasia is caused by a single recurrent mutation in the FLNA gene. Am J Hum Genet. 2010 Jul 9;87(1):146-53. doi: 10.1016/j.ajhg.2010.06.008. PMID: 20598277; PMCID: PMC2896768.

127: **Basel-Vanagaite L**. Acute lymphoblastic leukemia in Weaver syndrome. Am J Med Genet A. 2010 Feb;152A(2):383-6. doi: 10.1002/ajmg.a.33244. PMID: 20101679.

128: Senecky Y, Inbar D, Diamond G, **Basel-Vanagaite L**, Rigler S, Chodick G. Fetal alcohol spectrum disorder in Israel. Isr Med Assoc J. 2009 Oct;11(10):619-22. PMID: 20077950.

129: **Basel-Vanagaite L**, Pasmanik-Chor M, Lurie R, Yeheskel A, Kjaer KW. CDH3-Related Syndromes: Report on a New Mutation and Overview of the Genotype-Phenotype Correlations. Mol Syndromol. 2010;1(5):223-230. doi:

10.1159/000327156. Epub 2011 Apr 7. PMID: 22140374; PMCID: PMC3214945.

130: Mochida GH, Mahajnah M, Hill AD, **Basel-Vanagaite L**, Gleason D, Hill RS, Bodell A, Crosier M, Straussberg R, Walsh CA. A truncating mutation of TRAPPC9 is associated with autosomal-recessive intellectual disability and postnatal microcephaly. Am J Hum Genet. 2009 Dec;85(6):897-902. doi: 10.1016/j.ajhg.2009.10.027. PMID: 20004763; PMCID: PMC2790576.

131: **Basel-Vanagaite L**, Sarig O, Hershkovitz D, Fuchs-Telem D, Rapaport D, Gat A, Isman G, Shirazi I, Shohat M, Enk CD, Birk E, Kohlhase J, Matysiak-Scholze U, Maya I, Knopf C, Peffekoven A, Hennies HC, Bergman R, Horowitz M, Ishida-Yamamoto A, Sprecher E. RIN2 deficiency results in macrocephaly, alopecia, cutis laxa, and scoliosis: MACS syndrome. Am J Hum Genet. 2009 Aug;85(2):254-63. doi: 10.1016/j.ajhg.2009.07.001. Epub 2009 Jul 23. PMID: 19631308; PMCID: PMC2725231.

132: **Basel-Vanagaite L**, Shaffer L, Chitayat D. Keppen-Lubinsky syndrome: Expanding the phenotype. Am J Med Genet A. 2009 Aug;149A(8):1827-9. doi: 10.1002/ajmg.a.32975. PMID: 19610118.

133: Hucthagowder V, Morava E, Kornak U, Lefeber DJ, Fischer B, Dimopoulou A, Aldinger A, Choi J, Davis EC, Abuelo DN, Adamowicz M, Al-Aama J, **Basel-Vanagaite L**, Fernandez B, Greally MT, Gillessen-Kaesbach G, Kayserili H, Lemyre E, Tekin M, Türkmen S, Tuysuz B, Yüksel-Konuk B, Mundlos S, Van Maldergem L, Wevers RA, Urban Z. Loss-of-function mutations in ATP6V0A2 impair vesicular trafficking, tropoelastin secretion and cell survival. Hum Mol Genet. 2009 Jun 15;18(12):2149-65. doi: 10.1093/hmg/ddp148. Epub 2009 Mar 25. PMID: 19321599; PMCID: PMC2685755.

134: Fischlowitz S, Merlob P, **Basel-Vanagaite L**. Isolated familial posterior earlobe indentations. Am J Med Genet A. 2009 Feb 15;149A(4):800-1. doi: 10.1002/ajmg.a.32746. PMID: 19283852.

135: **Basel-Vanagaite L**. Clinical approaches to genetic mental retardation. Isr Med Assoc J. 2008 Nov;10(11):821-6. PMID: 19070297.

136: Manzini MC, Gleason D, Chang BS, Hill RS, Barry BJ, Partlow JN, Poduri A, Currier S, Galvin-Parton P, Shapiro LR, Schmidt K, Davis JG, **Basel-Vanagaite L**, Seidahmed MZ, Salih MA, Dobyns WB, Walsh CA. Ethnically diverse causes of Walker-Warburg syndrome (WWS): FCMD mutations are a more common cause of WWS outside of the Middle East. Hum Mutat. 2008 Nov;29(11):E231-41. doi: 10.1002/humu.20844. PMID: 18752264; PMCID: PMC2577713.

137: Van Maldergem L, Yuksel-Apak M, Kayserili H, Seemanova E, Giurgea S, **Basel-Vanagaite L**, Leao-Teles E, Vigneron J, Foulon M, Greally M, Jaeken J, Mundlos S, Dobyns WB. Cobblestone-like brain dysgenesis and altered glycosylation in congenital cutis laxa, Debre type. Neurology. 2008 Nov 11;71(20):1602-8. doi: 10.1212/01.wnl.0000327822.52212.c7. Epub 2008 Aug 20. PMID: 18716235.

138: Budde BS, Namavar Y, Barth PG, Poll-The BT, Nürnberg G, Becker C, van Ruissen F, Weterman MA, Fluiter K, te Beek ET, Aronica E, van der Knaap MS, Höhne W, Toliat MR, Crow YJ, Steinling M, Voit T, Roelenso F, Brussel W, Brockmann K, Kyllerman M, Boltshauser E, Hammersen G, Willemsen M, **Basel-Vanagaite L**, Krägeloh-Mann I, de Vries LS, Sztriha L, Muntoni F, Ferrie CD, Battini R, Hennekam RC, Grillo E, Beemer FA, Stoets LM, Wollnik B, Nürnberg P, Baas F. tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. Nat Genet. 2008 Sep;40(9):1113-8. doi: 10.1038/ng.204. PMID: 18711368.

139: Shkalim V, Eliaz N, Linder N, Merlob P, **Basel-Vanagaite L**. Autosomal dominant isolated question mark ear. Am J Med Genet A. 2008 Sep 1;146A(17):2280-3. doi: 10.1002/ajmg.a.32452. PMID: 18680186.

140: **Basel-Vanagaite L**, Dokal I, Tamary H, Avigdor A, Garty BZ, Volkov A, Vulliamy T. Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations. Haematologica. 2008 Jun;93(6):943-4. doi:

10.3324/haematol.12317. Epub 2008 May 6. PMID: 18460650.

141: Avrahami L, Maas S, Pasmanik-Chor M, Rainshtein L, Magal N, Smitt J, van Marle J, Shohat M, **Basel-Vanagaite L**. Autosomal recessive ichthyosis with hypotrichosis syndrome: further delineation of the phenotype. Clin Genet. 2008 Jul;74(1):47-53. doi: 10.1111/j.1399-0004.2008.01006.x. Epub 2008 Apr 28. PMID: 18445049.

142: Gothelf D, Goraly O, Avni S, Stawski M, Hartmann I, **Basel-Vanagaite L**, Apter A. Psychiatric morbidity with focus on obsessive-compulsive disorder in an Israeli cohort of adolescents with mild to moderate mental retardation. J Neural Transm (Vienna). 2008 Jun;115(6):929-36. doi: 10.1007/s00702-008-0037-4. Epub 2008 Mar 20. PMID: 18351287.

143: **Basel-Vanagaite L**, Taub E, Drasinover V, Magal N, Brudner A, Zlotogora J, Shohat M. Genetic carrier screening for spinal muscular atrophy and spinal muscular atrophy with respiratory distress 1 in an isolated population in Israel. Genet Test. 2008 Mar;12(1):53-6. doi: 10.1089/gte.2007.0030. PMID: 18298318.

144: **Basel-Vanagaite L**, Kornreich L, Schiller O, Yacobovich J, Merlob P. Yunis-Varon syndrome: further delineation of the phenotype. Am J Med Genet A. 2008 Feb 15;146A(4):532-7. doi: 10.1002/ajmg.a.32135. PMID: 18203163.

145: Kornak U, Reynders E, Dimopoulou A, van Reeuwijk J, Fischer B, Rajab A, Budde B, Nürnberg P, Foulquier F; ARCL Debré-type Study Group; Lefeber D, Urban Z, Gruenewald S, Annaert W, Brunner HG, van Bokhoven H, Wevers R, Morava E, Matthijs G, Van Maldergem L, Mundlos S. Impaired glycosylation and cutis laxa caused by mutations in the vesicular H+-ATPase subunit ATP6V0A2. Nat Genet. 2008 Jan;40(1):32-4. doi: 10.1038/ng.2007.45. Epub 2007 Dec 23. PMID: 18157129.

146: **Basel-Vanagaite L**. Genetics of autosomal recessive non-syndromic mental

retardation: recent advances. Clin Genet. 2007 Sep;72(3):167-74. doi: 10.1111/j.1399-0004.2007.00881.x. PMID: 17718851.

147: **Basel-Vanagaite L**, Rainshtein L, Inbar D, Gothelf D, Hennekam R, Straussberg R. Autosomal recessive mental retardation syndrome with anterior maxillary protrusion and strabismus: MRAMS syndrome. Am J Med Genet A. 2007 Aug 1;143A(15):1687-91. doi: 10.1002/ajmg.a.31810. PMID: 17618476.

148: Salmon A, Amikam D, Sodha N, Davidson S, **Basel-Vanagaite L**, Eeles RA, Abeliovich D, Peretz T. Rapid development of post-radiotherapy sarcoma and breast cancer in a patient with a novel germline 'de-novo' TP53 mutation. Clin Oncol (R Coll Radiol). 2007 Sep;19(7):490-3. doi: 10.1016/j.clon.2007.05.001. Epub 2007 Jun 14. PMID: 17572079.

149: **Basel-Vanagaite L**, Attia R, Ishida-Yamamoto A, Rainshtein L, Ben Amitai D, Lurie R, Pasmanik-Chor M, Indelman M, Zvulunov A, Saban S, Magal N, Sprecher E, Shohat M. Autosomal recessive ichthyosis with hypotrichosis caused by a mutation in ST14, encoding type II transmembrane serine protease matriptase. Am J Hum Genet. 2007 Mar;80(3):467-77. doi: 10.1086/512487. Epub 2007 Jan 23. PMID: 17273967; PMCID: PMC1821100.

150: **Basel-Vanagaite L**, Taub E, Halpern GJ, Drasinover V, Magal N, Davidov B, Zlotogora J, Shohat M. Genetic screening for autosomal recessive nonsyndromic mental retardation in an isolated population in Israel. Eur J Hum Genet. 2007 Feb;15(2):250-3. doi: 10.1038/sj.ejhg.5201750. Epub 2006 Dec 6. PMID: 17149387.

151: **Basel-Vanagaite L**, Pelet A, Steiner Z, Munnich A, Rozenbach Y, Shohat M, Lyonnet S. Allele dosage-dependent penetrance of RET proto-oncogene in an Israeli-Arab inbred family segregating Hirschsprung disease. Eur J Hum Genet. 2007 Feb;15(2):242-5. doi: 10.1038/sj.ejhg.5201733. Epub 2006 Nov 8. PMID: 17091122.

152: Hinkes B, Wiggins RC, Gbadegesin R, Vlangos CN, Seelow D, Nürnberg G, Garg P, Verma R, Chaib H, Hoskins BE, Ashraf S, Becker C, Hennies HC, Goyal M, Wharram BL, Schachter AD, Mudumana S, Drummond I, Kerjaschki D, Waldherr R, Dietrich A, Ozaltin F, Bakkaloglu A, Cleper R, **Basel-Vanagaite L**, Pohl M, Griebel M, Tsygin AN, Soylu A, Müller D, Sorli CS, Bunney TD, Katan M, Liu J, Attanasio M, O'toole JF, Hasselbacher K, Mucha B, Otto EA, Airik R, Kispert A, Kelley GG, Smrcka AV, Gudermann T, Holzman LB, Nürnberg P, Hildebrandt F. Positional cloning uncovers mutations in PLCE1 responsible for a nephrotic syndrome variant that may be reversible. Nat Genet. 2006 Dec;38(12):1397-405. doi: 10.1038/ng1918. Epub 2006 Nov 5. PMID: 17086182.

153: **Basel-Vanagaite L**, Davidov B, Friedman J, Yeshaya Y, Magal N, Drasinover V, Shohat M. Amniotic trisomy 11 mosaicism--is it a benign finding? Prenat Diagn. 2006 Sep;26(9):778-81. doi: 10.1002/pd.1501. PMID: 16810710.

154: **Basel-Vanagaite L**, Muncher L, Straussberg R, Pasmanik-Chor M, Yahav M, Rainshtein L, Walsh CA, Magal N, Taub E, Drasinover V, Shalev H, Attia R, Rechavi G, Simon AJ, Shohat M. Mutated nup62 causes autosomal recessive infantile bilateral striatal necrosis. Ann Neurol. 2006 Aug;60(2):214-22. doi: 10.1002/ana.20902. PMID: 16786527.

155: **Basel-Vanagaite L**, Straussberg R, Friez MJ, Inbar D, Korenreich L, Shohat M, Schwartz CE. Expanding the phenotypic spectrum of L1CAM-associated disease. Clin Genet. 2006 May;69(5):414-9. doi: 10.1111/j.1399-0004.2006.00607.x. PMID: 16650080.

156: Hellemans J, Debeer P, Wright M, Janecke A, Kjaer KW, Verdonk PC, Savarirayan R, **Basel L**, Moss C, Roth J, David A, De Paepe A, Coucke P, Mortier GR. Germline LEMD3 mutations are rare in sporadic patients with isolated melorheostosis. Hum Mutat. 2006 Mar;27(3):290. doi: 10.1002/humu.9403. PMID: 16470551.

157: **Basel-Vanagaite L**, Attia R, Yahav M, Ferland RJ, Anteki L, Walsh CA, Olender T, Straussberg R, Magal N, Taub E, Drasinover V, Alkelai A, Bercovich D, Rechavi G, Simon AJ, Shohat M. The CC2D1A, a member of a new gene family with C2 domains, is involved in autosomal recessive non-syndromic mental retardation. J Med Genet. 2006 Mar;43(3):203-10. doi: 10.1136/jmg.2005.035709. Epub 2005 Jul 20. PMID: 16033914; PMCID: PMC2563235.

158: Mahajnah M, **Basel-Vanagaite L**, Inbar D, Kornreich L, Weitz R, Straussberg R. Familial cognitive impairment with ataxia with oculomotor apraxia. J Child Neurol. 2005 Jun;20(6):523-5. doi: 10.1177/08830738050200610. PMID: 15996403.

159: Straussberg R, **Basel-Vanagaite L**, Kivity S, Dabby R, Cirak S, Nurnberg P, Voit T, Mahajnah M, Inbar D, Saifi GM, Lupski JR, Delague V, Megarbane A, Richter A, Leshinsky E, Berkovic SF. An autosomal recessive cerebellar ataxia syndrome with upward gaze palsy, neuropathy, and seizures. Neurology. 2005 Jan 11;64(1):142-4. doi: 10.1212/01.WNL.0000148600.60470.E6. PMID: 15642921.

160: Sheen VL, **Basel-Vanagaite L**, Goodman JR, Scheffer IE, Bodell A, Ganesh VS, Ravenscroft R, Hill RS, Cherry TJ, Shugart YY, Barkovich J, Straussberg R, Walsh CA. Etiological heterogeneity of familial periventricular heterotopia and hydrocephalus. Brain Dev. 2004 Aug;26(5):326-34. doi: 10.1016/j.braindev.2003.09.004. PMID: 15165674.

161: Piao X, Hill RS, Bodell A, Chang BS, **Basel-Vanagaite L**, Straussberg R, Dobyns WB, Qasrawi B, Winter RM, Innes AM, Voit T, Ross ME, Michaud JL, Déscarie JC, Barkovich AJ, Walsh CA. G protein-coupled receptor-dependent development of human frontal cortex. Science. 2004 Mar 26;303(5666):2033-6. doi: 10.1126/science.1092780. PMID: 15044805.

162: **Basel-Vanagaite L**, Straussberg R, Ovadia H, Kaplan A, Magal N, Shorer Z, Shalev H, Walsh C, Shohat M. Infantile bilateral striatal necrosis maps to chromosome 19q. Neurology. 2004 Jan 13;62(1):87-90. doi:

10.1212/01.wnl.0000101680.49036.69. PMID: 14718703.

163: Hermans MM, van Leenen D, Kroos MA, Beesley CE, Van Der Ploeg AT, Sakuraba H, Wevers R, Kleijer W, Michelakakis H, Kirk EP, Fletcher J, Bosshard N, **Basel-Vanagaite L**, Besley G, Reuser AJ. Twenty-two novel mutations in the lysosomal alpha-glucosidase gene (GAA) underscore the genotype-phenotype correlation in glycogen storage disease type II. Hum Mutat. 2004 Jan;23(1):47-56. doi: 10.1002/humu.10286. PMID: 14695532.

164: **Basel-Vanagaite L**, Alkelai A, Straussberg R, Magal N, Inbar D, Mahajna M, Shohat M. Mapping of a new locus for autosomal recessive non-syndromic mental retardation in the chromosomal region 19p13.12-p13.2: further genetic heterogeneity. J Med Genet. 2003 Oct;40(10):729-32. doi: 10.1136/jmg.40.10.729. PMID: 14569116; PMCID: PMC1735276.

165: Claes L, Ceulemans B, Audenaert D, Smets K, Löfgren A, Del-Favero J, Ala-Mello S, **Basel-Vanagaite L**, Plecko B, Raskin S, Thiry P, Wolf NI, Van Broeckhoven C, De Jonghe P. De novo SCN1A mutations are a major cause of severe myoclonic epilepsy of infancy. Hum Mutat. 2003 Jun;21(6):615-21. doi: 10.1002/humu.10217. PMID: 12754708.

166: **Basel-Vanagaite L**, Marcus N, Klinger G, Shohat M, Levit O, Karmazin B, Taub E, Sirota L. New syndrome of simplified gyral pattern, micromelia, dysmorphic features and early death. Am J Med Genet A. 2003 Jun 1;119A(2):200-6. doi: 10.1002/ajmg.a.20133. PMID: 12749064.

167: Chang BS, Piao X, Bodell A, **Basel-Vanagaite L**, Straussberg R, Dobyns WB, Qasrawi B, Winter RM, Innes AM, Voit T, Grant PE, Barkovich AJ, Walsh CA. Bilateral frontoparietal polymicrogyria: clinical and radiological features in 10 families with linkage to chromosome 16. Ann Neurol. 2003 May;53(5):596-606. doi: 10.1002/ana.10520. PMID: 12730993.

168: **Basel-Vanagaite L**, Shohat M, Udler Y, Karmazin B, Levit O, Merlob P. Branchial cyst, sensorineural deafness, congenital heart defect, and skeletal abnormalities: Branchio-oto-cardio-skeletal (BOCS) syndrome? Am J Med Genet. 2002 Nov 15;113(1):78-81. doi: 10.1002/ajmg.10723. PMID: 12400069.

169: Straussberg R, Shorer Z, Weitz R, **Basel L**, Kornreich L, Corie CI, Harel L, Djaldetti R, Amir J. Familial infantile bilateral striatal necrosis: clinical features and response to biotin treatment. Neurology. 2002 Oct 8;59(7):983-9. doi: 10.1212/wnl.59.7.983. PMID: 12374138.

170: Wolf B, Jensen K, Hüner G, Demirkol M, Baykal T, Divry P, Rolland MO, Perez-Cerdá C, Ugarte M, Straussberg R, **Basel-Vanagaite L**, Baumgartner ER, Suormala T, Scholl S, Das AM, Schweitzer S, Pronicka E, Sykut-Cegielska J. Seventeen novel mutations that cause profound biotinidase deficiency. Mol Genet Metab. 2002 Sep-Oct;77(1-2):108-11. doi: 10.1016/s1096-7192(02)00149-x. PMID: 12359137.

171: Piao X, **Basel-Vanagaite L**, Straussberg R, Grant PE, Pugh EW, Doheny K, Doan B, Hong SE, Shugart YY, Walsh CA. An autosomal recessive form of bilateral frontoparietal polymicrogyria maps to chromosome 16q12.2-21. Am J Hum Genet. 2002 Apr;70(4):1028-33. doi: 10.1086/339552. Epub 2002 Feb 13. PMID: 11845408; PMCID: PMC379097.

172: Aganna E, Zeharia A, Hitman GA, **Basel-Vanagaite L**, Allotey RA, Booth DR, Hawkins PN, Thacker C, Syndercombe-Court D, McDermott MF. An Israeli Arab patient with a de novo TNFRSF1A mutation causing tumor necrosis factor receptor-associated periodic syndrome. Arthritis Rheum. 2002 Jan;46(1):245-9. doi: 10.1002/1529-0131(200201)46:1<245::AID-ART10038>3.0.CO;2-6. PMID: 11817598.

173: Toledano-Alhadef H, **Basel-Vanagaite L**, Magal N, Davidov B, Ehrlich S, Drasinover V, Taub E, Halpern GJ, Ginott N, Shohat M. Fragile-X carrier screening and the prevalence of premutation and full-mutation carriers in

Israel. Am J Hum Genet. 2001 Aug;69(2):351-60. doi: 10.1086/321974. Epub 2001 Jul 6. PMID: 11443541; PMCID: PMC1235307.

174: **Basel-Vanagaite L**, Zeharia A, Amir J, Mimouni M. Edema associated with valproate therapy. Ann Pharmacother. 1999 Dec;33(12):1370-1. doi: 10.1345/aph.19070. PMID: 10630841.

175: Gilad S, Bar-Shira A, Harnik R, Shkedy D, Ziv Y, Khosravi R, Brown K, **Vanagaite L**, Xu G, Frydman M, Lavin MF, Hill D, Tagle DA, Shiloh Y. Ataxia-telangiectasia: founder effect among north African Jews. Hum Mol Genet. 1996 Dec;5(12):2033-7. doi: 10.1093/hmg/5.12.2033. PMID: 8968760.

176: Savitsky K, Bar-Shira A, Gilad S, Rotman G, Ziv Y, **Vanagaite L**, Tagle DA, Smith S, Uziel T, Sfez S, Ashkenazi M, Pecker I, Frydman M, Harnik R, Patanjali SR, Simmons A, Clines GA, Sartiel A, Gatti RA, Chessa L, Sanal O, Lavin MF, Jaspers NG, Taylor AM, Arlett CF, Miki T, Weissman SM, Lovett M, Collins FS, Shiloh Y. A single ataxia telangiectasia gene with a product similar to PI-3 kinase. Science. 1995 Jun 23;268(5218):1749-53. doi: 10.1126/science.7792600. PMID: 7792600.

177: **Vanagaite L**, James MR, Rotman G, Savitsky K, Bar-Shira A, Gilad S, Ziv Y, Uchenik V, Sartiel A, Collins FS, Sheffield VC, Richard CW 3rd, Weissenbach J, Shiloh Y. A high-density microsatellite map of the ataxia-telangiectasia locus. Hum Genet. 1995 Apr;95(4):451-4. doi: 10.1007/BF00208975. PMID: 7705845.

178: Rotman G, **Vanagaite L**, Collins FS, Shiloh Y. Rapid identification of polymorphic CA-repeats in YAC clones. Mol Biotechnol. 1995 Apr;3(2):85-92. doi: 10.1007/BF02789104. PMID: 7620980.

179: Rotman G, Savitski K, **Vanagaite L**, Bar-Shira A, Ziv Y, Gilad S, Uchenik V, Smith S, Shiloh Y. Physical and genetic mapping at the ATA/ATC locus on chromosome 11q22-23. Int J Radiat Biol. 1994 Dec;66(6 Suppl):S63-6. PMID:

7836854.

180: Rotman G, Savitsky K, Ziv Y, Cole CG, Higgins MJ, Bar-Am I, Dunham I, Bar-Shira A, **Vanagaite L**, Qin S, Zhang J, Nowak NJ, Chandrasekharappa SC, Lehrach H, Avivi L, Shows TB, Collins FS, Bentley DR, Shiloh Y. A YAC contig spanning the ataxia-telangiectasia locus (groups A and C) at 11q22-q23. Genomics. 1994 Nov 15;24(2):234-42. doi: 10.1006/geno.1994.1611. PMID: 7698744.

181: Rotman G, **Vanagaite L**, Collins FS, Shiloh Y. Three dinucleotide repeat polymorphisms at the ataxia-telangiectasia locus. Hum Mol Genet. 1994 Nov;3(11):2079. PMID: 7874134.

182: **Vanagaite L**, Savitsky K, Rotman G, Ziv Y, Gerken SC, White R, Weissenbach J, Gillett G, Benham FJ, Richard CW 3rd, James MR, Collins FS, Shiloh Y. Physical localization of microsatellite markers at the ataxia-telangiectasia locus at 11q22-q23. Genomics. 1994 Jul 1;22(1):231-3. doi: 10.1006/geno.1994.1370. PMID: 7959777.

183: Kleiman S, Avigad S, **Vanagaite L**, Shmuelevitz A, David M, Eisensmith RC, Brand N, Schwartz G, Rey F, Munnich A, Woo SL, Shiloh Y. Origins of hyperphenylalaninemia in Israel. Eur J Hum Genet. 1994;2(1):24-34. doi: 10.1159/000472338. PMID: 7913865.

184: McConville CM, Byrd PJ, Ambrose HJ, Stankovic T, Ziv Y, Bar-Shira A, **Vanagaite L**, Rotman G, Shiloh Y, Gillett GT, et al. Paired STSs amplified from radiation hybrids, and from associated YACs, identify highly polymorphic loci flanking the ataxia telangiectasia locus on chromosome 11q22-23. Hum Mol Genet. 1993 Jul;2(7):969-74. doi: 10.1093/hmg/2.7.969. PMID: 8364579.

CHAPTERS IN BOOKS

1. **Basel-Vanagaite L**, Halpern GJ, Jaber L. General health topics associated with consanguinity; genetic disorders and congenital malformations; benefits.

In: Consanguinity – its Impact, Consequences and Management
Editors: Editors Lutfi A. Jaber, Gabrielle J. Halpern
Bentham Science Publishers, Sharjah, United Arab Emirates**,** pp. 50-74, 2014.

2. Halpern GJ, **Basel-Vanagaite L**, Jaber L. Future Strategies 2 – Genetic Perspectives – Counseling, Screening, Testing, Research, and Intervention.
In: Consanguinity – its Impact, Consequences and Management
Editors: Editors Lutfi A. Jaber, Gabrielle J. Halpern
Bentham Science Publishers, Sharjah, United Arab Emirates, pp**.** 158-176**,** 2014.

ITEMS IN ENCYCLOPEDIAS

1. **Basel-Vanagaite L**.
Molecular Genetics of Mental Retardation.
In: ENCYCLOPEDIA OF LIFE SCIENCES.
John Wiley & Sons, Ltd., Chichester http://www.els.net/, December 2008.