CURRICULUM VITAE

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DATE AND PLACE OF BIRTH 03.03.1967, VILNIUS, LITHUANIA

DATE OF ARRIVAL IN ISRAEL: MAY 1991

ZAHAL (ISRAELI) MILITARY SERVICE: Not applicable

MARITAL STATUS: MARRIED +2

Education

1984-1991: Vilnius University, Faculty of Medicine, MD degree, graduated with honors

1991-1995: Tel Aviv University, Faculty of Medicine, 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: Residency in Pediatrics, Schneider Children's Medical Center of Israel, Petah Tikva, Israel, graduated with honors

2000-2002: Residency in Genetics, Rabin Medical Center and Schneider Children's Medical Center of Israel, Petah Tikva, Israel, graduated with honors

2011: Lecturer, School of Continuing Medical Education, Pediatric Cardiology

2011: Lecturer, School of Continuing Medical Education, Pediatric Endocrinology

2012-2014: Lecturer, School of Continuing Medical Education: update to physicians

2013: Member of the committee for reorganization of genetics curriculum, Sackler School of Medicine

2014-current: Lecturer, Tel Aviv University medical students "Reproduction system" 4th year medical students

2014: Member of the committee on preclinical studies of the Sackler School of Medicine

2014-current: Organizer, tutor - new MSc Program "Genetic Counseling", Sackler School of Medicine

2015: Lecturer, Tel Aviv University, Sackler School of Medicine, New York State Program

PROFESSIONAL EXPERIENCE

2002 - 2008: Senior Geneticist, Schneider Children's Medical Center of Israel and Raphael Recanati Genetic Institute, Rabin Medical Center, Beilinson Campus, Petah Tikva, Israel

2002-2008: Scientific exchange visits to the Necker hospital in Paris, France, Boston Children's hospital, Boston USA, Chicago University, Chicago, USA, Great Ormond's street hospital, London, UK

2008 - 2014: Director, Pediatric Genetics Unit, Schneider Children's Medical Center of Israel and Raphael Recanati Genetic Institute, Rabin Medical Center, Beilinson Campus, Petah Tikva, Israel

2015-current: Acting Director, The Raphael Recanati Genetics Institute, Beilinson campus and Schneider Children's Medical Center of Israel, Rabin Medical Center, Petah Tikva, Israel

2012-present: Head, Immunology and Genetics laboratory, the Felsenstein Medical Research Center, Tel Aviv University

ACTIVE PARTICIPATION IN SCIENTIFIC MEETINGS

Date	City, country	Subject	Role
2001	Vienna, Austria	10th International Congress of Human Genetics	Poster presentation
2001	San Diego, US	51st Annual Meeting of the American Society of Human Genetics	Poster presentation
2002	Strasbourg, France	34th European Human Genetics Conference	Poster presentation
2003	Los Angeles, US	53rd Annual Meeting of the American Society of Human Genetics	Poster presentation

2004	Toronto, Canada	54th Annual Meeting of the American Society of Human Genetics	Poster presentation
2005	Salt Lake City, US	55th Annual Meeting of the American Society of Human Genetics	Platform presentation
2006	Amsterdam, Netherlands	38th European Human Genetics Conference	Platform presentation
2006	Vilnius, Lithuania	8th Baltic Congress of Laboratory Medicine	Invited speaker
2006	Brisbane, Australia	11th International Congress of Human Genetics	Platform presentation
2007	Vilnius, Lithuania	9th International Conference of the Baltic Child Neurology Association	Invited speaker
2007	Strasburg, France	18th European Dysmprphology Meeting	Platform presentation
2007	New York, USA	National Institutes of Mental Health Research Workshop	Invited speaker
2008	Eilat, Israel	5th Congress of the Federation of the Israel Societies for Experimental Biology (Ilanit)	Invited speaker
2008	Ventura, USA	Gordon Conference on Plasminogen Activation and Extracellular Proteolysis	Invited speaker
2008	Barcelona, Spain	40th European Human Genetics Conference	Platform presentation
2008	Quebec, Canada	29th Annual David W. Smith Workshop on Malformations and Morphogenesis	Platform presentation
2008	Manchester, UK	13th Manchester Dysmorphology Meeting	Poster
2009	Vilnius,	6th Baltic Congress of	Invited speaker

	Lithuania	Neurology	
2009	Warwick, UK	British Human Genetics Conference	Plenary platform presentation – late breaking research
2010	Jerusalem, Israel	Annual meeting of the Israeli Society of Genetics	Platform Presentation
2010	Cambridge, UK	Genomic Disorders Meeting	Poster
2010	Manchester, UK	14th Manchester Dysmorphology Meeting	Platform presentation
2010, 2011	Jerusalem, Israel	Neurogenetic seminar, Monique and Jacques Roboh Department of Genetic Research	Invited speaker
2011	Haifa, Israel	UK-Israeli genetics conference, Rambam Health Care Campus	Invited speaker
2011	Saint-Petersburg, Russia	Second All-Russian Conference for Rare Diseases and Rarely-Used Medical Technologies	Invited speaker
2011	Amsterdam, Netherlands	43th European Human Genetics Conference	Platform presentation
2011	Saint-Petersburg, Russia	Healthy Woman – Healthy Child All-Russian Conference Healthy Child”	Invited speaker
2011	Bangkok, Thailand	The 4th Asia Pacific Congress on Controversies in Obsterics, Gynecology and Infertility	Invited speaker
2011	Montreal, Canada	12th International Congress of Human Genetics	Poster presentation
2011	Atlanta, USA	33th Annual David W. Smith Workshop on Malformations and Morphogenesis	Poster presentation
2012	Naarden,	ENMC International Workshop on SMA-RD	Invited speaker

	Netherlands	Type 1	
2012	Nuerenberg, Germany	44th European Human Genetics Conference	Platform presentation
2012	Gelendzhik, Russia	Meeting of the Russian Association for Reproductive Genetics	Invited speaker
2012	Manchester, UK	15th Manchester Dysmorphology Meeting	Platform presentation
2012	San Francisco, USA	62nd Annual Meeting of the American Society of Human Genetics	Platform presentation
2012	Novosibirsk, Russia	Issues in Fertility Preservation in Oncological Patients and Novel Technologies in Genetic Investigations of Human Embryos and Neonates born after ATR	Invited speaker
2013	Tel Aviv, Israel	Annual meeting of the Genetic Society of Israel	Platform Presentation
2013	Petah Tikva, Israel	1st Joint Meeting of the Fertility Societies from Israel and CIS Countries - Russia, Ukraine, Kazakhstan and Belarus	Invited speaker
2013	Istanbul, Turkey	Dysmorphology Days with CRANIRARE-2: Educational Contribution	Invited speaker
2013	Kaunas, Lithuania	12th International Conference of the Baltic Child Neurology Association	Invited speaker
2013	Narva, Estonia	Annual meeting of the Estonian Society of Human Genetics	Invited keynote speaker
2013	Eilat, Israel	7th Congress of the Federation of the Israel Societies for Experimental Biology (Ilanit)	Session Chair and invited speaker
2014	Astana,	Master class in genetics for	Organizer and

	Kazakhstan	physicians	lecturer
2014	Novosibirsk, Russia	IVF: Predictors of Success, conference	Invited lecturer
2014	Moscow, Russia	Master class in genetics for physicians	Organizer and lecturer
2014	Manchester, UK	16th Manchester Dysmorphology Meeting	Poster presentation
2014	Tel Aviv, Israel	Annual meeting of the Israeli Society of Genetics	Platform Presentation
2015	Yekaterinburg, Russia	Master class in genetics for physicians	Organizer and lecturer
2015	Tel Aviv, Israel	Fourth Up Close and Personalized, International Congress on Personalized Medicine	Invited speaker
2015	Kiev, Ukraine	Healthy Child: From Fertilization to Birth	Invited speaker
2016	Malaga, Spain	18th ISPOG congress	Invited speaker
2016	Vancouver, Canada	65th Annual Meeting of the American Society of Human Genetics	Workshop organizer and moderator
2016	Manchester, UK	18th Manchester Dysmorphology Meeting	Poster presentation

MEMBERSHIP IN PROFESSIONAL SOCIETIES*

Years (period)	Name of Organization	Role
2003 - 2015	Israeli Society of Medical Geneticists	Member
2005-2007, 2011	Israeli Society of Pediatrics	Member
2007-2012	Examination Committee for Residency in	Member

	Medical Genetics	
2007-2014	Research Committee, Schneider Children's Medical Center of Israel	Head
2007 - 2013	Sackler School of Medicine Faculty Council	Senior lecturers' representative
2008-current	European Society of Human Genetics	Member
2008	Tel Aviv University Research Fair organization committee	Member
2008 – current	Committee evaluating scientific projects as a part of MD study program, Sackler School of Medicine	Member
2009-2013	Orphanet Europe	National Coordinator
2010-current	American Society of Human Genetics	Member
2012-present	International consortium of autosomal recessive intellectual disability (CARID)	Member
2015-current	Israeli Society of Medical Geneticists	Head

DOCTORAL STUDENTS MENTORED BY CANDIDATE

Years (period)	Name of Student	Subject	Academic Institute	Ph.D/ M.D	In collaboration with
2003-2006	Liora Muncher	Identification of gene causing bilateral infantile striatal necrosis	Tel Aviv University	PhD	Prof. Motti Shohat

2006-2011	Noa Shoshani	Studies of the biological function of the p62 protein encoded by the nup62 gene involved in IBSN	Tel Aviv University	PhD	Prof. Motti Shohat
2014-current	Pola Smirin-Yosef	Identification of genes causing intellectual disability	Ariel University	PhD	Prof. Danny Baranes Dr. Mali Salmon-Divon

E. M.A./M.Sc. Students

Years (period)	Name of Student	Subject	Academic Institute	EMA/MSc	In collaboration with
2002-2003	Ana Alkelai	Genetic linkage study of the gene causing nonspecific mental retardation	Tel Aviv University	MSc	Prof. Motti Shohat
2003-2004	Rachel Berger	Detection of chromosomal imbalances in patients with mental retardation using comparative genome hybridization	Tel Aviv University	MSc	Prof. Motti Shohat
2007-2010	Efrat Birk	Genetic linkage study of the gene causing MRAMS syndrome	Tel Aviv University	MSc	Prof. Motti Shohat
2009-2011	Shlomit Hellman	Identification of the gene causing microcephaly-thin corpus callosum syndrome	Tel Aviv University	MSc	Prof. Motti Shohat

Medical residents

2011-2012	Gila Nelkenbaum - Isman	Genetic basis of generalized basaloid follicular hamartoma syndrome	Tel Aviv Sourasky Medical Center	Research in basic sciences as a part of the medical residency program	Prof Eli Sprecher
2013-2014	Goldsmith Tomer	Identification of genetic basis of autosomal recessive congenital ichthyosis	Tel Aviv Sourasky Medical Center	Research in basic sciences as a part of the medical residency program	Prof Eli Sprecher
2015	Lital Cohen-Weig	Retrospective study of the diagnostic yield of chromosomal microarray testing in prenatal and postnatal samples	Schneider Children's Medical Center of Israel	Research in basic sciences as a part of the medical residency program	-

Postdoctoral Students

2013	Dr. Sky Gross	Hearing and listening: ethical and sociological aspects of genetic testing and next-generation sequencing technologies as applied to deafness and hearing disabilities	Tel Aviv University	Postdoctoral studies	Prof. Karen B. Avraham Prof. Amos Shapira

Grants

Years	Investigator (Principal/other)	Granted by Institute/Company	Total Amount
2004-2007	PI	Israeli Science Foundation	600,000 NIS
2006-2007	PI	Ministry of Health - Chief Scientist's Foundation	80,000 NIS
2009-2012	PI	Ministry of Health - Chief Scientist's Foundation	300,000 NIS
2009-2010	PI	Tel Aviv University Margaret Stolz Foundation	25,000 NIS
2009-2014	PI	Israeli Science Foundation	900,000 NIS
2011-2012	PI	Tel Aviv University Sackler School of Medicine	40,000 NIS
2012-2013	PI	Beilinson - Bar-Ilan research grant	60,000 NIS
2013-2016	PI for TAU	European community TEMPUS educational grant	750,000 NIS
2016-current	PI	Research collaboration grant, Regeneron biotechnology company	400,000 NIS

AWARDS

1992-1994: Branco Weiss Foundation Fellowship

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

2013: European EDEN scholarship for lecturing on Medical Genetics as a visiting professor at the Vilnius University

SCIENTIFIC PUBLICATIONS

B.1. ORIGINAL ARTICLES

1.Kleiman S, Vanagaite L, Bernstein J, Schwartz G, Brand N, Elitzur A, Woo SL, Shiloh Y. Phenylketonuria: variable phenotypic outcomes of the R261Q mutation and maternal PKU in the offspring of a healthy homozygote. Journal of Medical Genetics, 30, pp.284-288, 1993.

2. McConville CM, Byrd PJ, Ambrose HJ, Stankovic T, Ziv Y, Bar-Shira A, Vanagaite L, Rotman G, Shiloh Y, Gillet GT, Riley JH and Taylor MR.
Paired STSs amplified from radiation hybrids, and from associated YACs, identify highly polymorphic loci flanking the ataxia telangiectasia locus on chromosome 11q22-23. Human Molecular Genetics, 2, pp. 969-974, 1993.

3. Vanagaite L, Savitsky K, Rotman G, Ziv Y, Gerken SC, White R, Weissenbach J, Gillett G, Benham FJ, Richard CW, James MR, Collins FS, and Shiloh Y. Physical localization of microsatellite markers at the ataxia-telangiectasia locus at 11q22-23. Genomics, 22, pp. 231-233, 1994.

4. Kleiman S, Avigad S, Vanagaite L, Shmuelevitz A, David M, Eisensmith RC, Brand N, Schwartz G, Rey F, Munnich A, Woo SLC, and Shiloh, Y. Origins of hyperphenylalaninemia in Israel.
European Journal of Human Genetics, 2, pp.24-34, 1994.

5. Rotman G, Savitsky K, Ziv Y, Cole CG, Higgins MJ, Bar-Am I, Dunham I, Bar-Shira A, Vanagaite L, Shinzen Q, Zhang J, Nowak NJ, Chandrasekharappa SC, Lehrach H, Avivi L, Shows TB, Collins FS, Bentley DR and Shiloh Y. A YAC contig spanning the ataxia-telangiectasia locus (groups A and C) at 11q22-q23. Genomics, 24, pp.234-242, 1994.

6. Rotman G, Savitsky K, Vanagaite L, Bar-Shira A, Ziv Y, Gilad S, Uchenik V, Smith S and Shiloh Y. Physical and genetic mapping at the ATA/ATC locus on chromosome 11q22-23.
International Journal of Radiation Biology, 66, S63-S66, 1994.

7. Rotman G, Vanagaite L, Collins FS, and Shiloh Y. Three dinucleotide repeat polymorphisms at the ataxia-telangiectasia locus.
Human Molecular Genetics, 3, pp.2079, 1994.

8. Rotman G, Vanagaite L, Collins FS, and Shiloh Y. Rapid identification of polymorphic CA repeats in YAC clones.
Molecular Biotechnology, 3, pp.85-92, 1995. IF - 2.091, RF – 73/160.

9. Vanagaite L, James MR, Rotman G, Savitsky K, Bar-Shira A, Gilad S, Ziv Y, Uchenik V, Sartiel A, Collins FS, Sheffield VC, Weissenbach J, and Shiloh Y. A high-density microsatellite map of the ataxia-telangiectasia locus.
Human Genetics, 95, pp. 451-454, 1995.

10. Lange E, Borreson AL, Chen X, Chessa L, Chiplunkar S, Concannon P, Dandekar S, Gerken S, Lange K, Liang T, McConville C, Polakow J, Porras O, Rotman G, Sanal O, Telatar M, Sheikhavandi S, Shiloh Y, Sobel E, Taylor M, Udar N, Uhrhammer N, Vanagaite L, Wang Z, Yang HM, Yang L, Ziv Y, and Gatti RA. Localization of an ataxia-telangiectasia gene to a 500 kb interval on chromosome 11q23.1 by linkage analysis of 176 families in an international consortium.
American Journal of Human Genetics, 57, pp.112-119, 1995.

11. Savicky K, Bar-Shira A, Gilad S, Rotman G, Ziv Y, Vanagaite L, Tagle D, Smith S, Uziel T, Sfez S, Ashkenazi M, Pecker I, Frydman M, Harnik R, Patanjali SR, Simmons A, Clines G, Sartiel A, Gatti RA, Chessa L, Sanal O, Lavin MF, Jaspers NGJ, Taylor AMR, 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, 268, pp.1749-1753, 1995.

12. 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.
Human Molecular Genetics, 5, pp.2033-2037, 1996.

13. 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.
American Journal of Human Genetics, 69, pp.351-60, 2001.

14. Wolf B, Jensen K, Huner G, Demirkol M, Baykal T, Divry P, Rolland M, Perez-Cerda C, Ugarte M, Straussberg R, Basel-Vanagaite L, Baumgartner E, Suormala T, Scholl S, Das A, Schweitzer S, Pronicka E, Sykut-Cegielska J. Seventeen novel mutations that cause profound biotinidase deficiency.
Molecular Genetics & Metabolism, 77, pp.108-111, 2002.

15. Basel-Vanagaite L, Shohat M, Udler Y, Karmazin B, Levit O and Merlob P. Branchial cyst, deafness, congenital heart disease and skeletal abnormalities: branchio-oto-cardio-skeletal (BOCS) syndrome?
American Journal of Medical Genetics, 113, pp. 78-81, 2002.

16. 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.
American Journal of Human Genetics, 70, pp.1028-33, 2002.
*Shared first coauthorship

17. Aganna E, Zeharia A, Hitman GA, Basel-Vanagaite L, Majeed HA, Allotey RA, Booth DR, Hawkins PN, Thacker C, Syndercombe-Court D, McDermott MF. An Israeli Arab patient with a de novo TNFRSF1A mutation causing TNF receptor associated periodic syndrome (TRAPS).
Arthritis & Rheumatism, 46, pp.245-9, 2002.

18. Basel-Vanagaite L, Marcus N, Klinger G, Shohat M, Levit O, Karmazin B, and Sirota L. A new syndrome of simplified gyral pattern, micromelia, dysmorphic features, hypoparathyroidism and early death.

American Journal of Medical Genetics, 119A, pp. 200-206, 2003.

19. 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.
Annals of Neurology, 53, pp. 596-606, 2003.

20. Claes L, Ceulemans B, Audenaert D, Smets K, Lofgren 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.
Human Mutation, 21, pp. 615-21, 2003.

21. 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.
Journal of Medical Genetics, 40, pp. 729-32, 2003.

22. 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.
Human Mutation, 23, pp. 47-56, 2004.

23. 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, 62, pp. 87-90, 2004.

24. 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, Descarie JC, Barkovich AJ, Walsh CA. G protein-coupled receptor-dependent development of human frontal cortex.
Science, 303, pp. 2033-6, 2004.

25. 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 & Development, 26, pp. 326-34, 2004.

26. 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 nonsyndromic mental retardation.
Journal of Medical Genetics, 43, pp. 203-10, 2006.
*Shared first coauthorship

27. 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.
Human Mutation, 27, pp. 290, 2006.

28. 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.
Annals of Neurology, 60, pp. 214-2, 2006.
*Shared first coauthorship

29. Basel-Vanagaite L, Davidov B, Friedman J, Yeshaya Y, Magal N, Drasinover V, Shohat M. Amniotic trisomy 11 mosaicism - is it a benign finding?
Prenatal Diagnosis, 26, pp. 778-81, 2006.

30. Basel-Vanagaite L, Straussberg R, Friez MJ, Inbar D, Korenreich L, Shohat M, Schwartz CE. Expanding the phenotypic spectrum of L1CAM-associated disease.
Clinical Genetics, 69, pp. 414-9, 2006.

31. Hinkes B, Wiggins RC, Gbadegesin R, Vlangos CN, Seelow D, Nurnberg 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 of PLCE1 mutations as the first cause of a nephrotic syndrome variant which may be reversible.
Nature Genetics, 38, pp. 1397-405, 2006.

32. Basel-Vanagaite L, Pelet A, Steiner Z, Munnich A, Rozenbach Y, Shohat M, Lyonnet S. Allele dosage dependent penetrance of RET protooncogene in Israeli Arab inbred families segregating Hirschsprung disease.
European Journal of Human Genetics, 15, pp. 242-5, 2007.

33. 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.
European Journal of Human Genetics, 15, pp. 250-3, 2007.

34. 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 (ARIH) caused by a mutation in ST14, encoding type II transmembrane serine protease matriptase.
American Journal of Human Genetics, 80, pp.467-77, 2007.

35. Basel-Vanagaite L, Rainshtein L, Inbar D, Gothelf D, Hennekam R, and Straussberg R. Autosomal Recessive Mental Retardation Syndrome with Anterior Maxillary Protrusion and Strabismus: MRAMS Syndrome.

American Journal of Medical Genetics A, 143, pp. 1687-1691, 2007.

36. Salmon A, Amikam D, Sodha N, Davidson S, Basel-Vanagaite L, Eeles RE, Abeliovich D, and Peretz T. Rapid development of post radiotherapy sarcoma and breast cancer in a patient with a novel germline 'de-novo' TP53 mutation.
Clinical Oncology, 19, pp. 490-493, 2007.

37. Gothelf D, Goraly O, Avni S, Stawsky M, Dotan 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.
Journal of Neural Transmission, 115, pp. 929-936, 2008.

38. 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.
Genetic testing, 12, pp. 53-56, 2008.

39. Basel-Vanagaite L, Dokal I, Tamary H, Avigdor A, Garty BZ and Vulliamy T. Expanding the clinical phenotype of autosomal dominant dyskeratosis congenita caused by TERT mutations.
Haematologica, 93, pp. 943-944, 2008.

40. Avrahami L, Maas S, Pasmanik-Chor M, Rainshtein L, Magal N, Sillevis Smitt JH, Shohat M, Basel-Vanagaite L. Autosomal recessive ichthyosis with hypotrichosis (ARIH) syndrome: further delineation of the phenotype.
Clinical Genetics, 74, pp. 47-53, 2008.

41. Budde BS, Namavar Y, Barth PG, Poll-The BT, Nürnberg G, Becker C, van Ruissen F, Weterman MAJ, te Beek E, van der Knaap MS, Höhne W, Toliat MR, Crow Y, Steinlin M, Voit T, Roelens F, Brussel W, Brockmann K, Kyllerman M, Boltshauser E, Hammersen G, Brunner HG, Basel-Vanagaite L, Krägeloh-Mann I, de Vries L, Sztriha L, Muntoni F, Ferrie CD, Battini R, Hennekam RCM, Grillo E, Beemer FA, Stoets LME, Nürnberg P, Baas F. tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia.
Nature Genetics, 40, pp.1113-1118, 2008.

42. Van Maldergem L, Yuksel-Apak M, Kayserili H, Seemanova E, Giurgea S, Vigneron J, Greally M, Salih M, Quelhas D, Basel-Vanagaite L, Jaeken J, Mundlos S, Dobyns WB. Cobblestone-like brain dysgenesis and altered glycosylation in congenital cutis laxa, Debre type.
Neurology, 71, pp.1602-1608, 2008.

43. Manzini CM, Gleason D, Chang BS, Hill CR, Barry BJ, Partlow JN, Poduri A, Currier S, Galvin-Parton P, Shapiro LR, Schmidt K, Davis JG, Basel-Vanagaite L, Seidahmed MZ, Salih MAM, Dobyns WB, Walsh CA. Ethnically diverse causes of Walker Warburg Syndrome (WWS): fukutin mutations are a more common cause of WWS outside of the Middle East.
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B.2. CASE REPORTS

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B.3. REVIEW ARTICLES

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C. CHAPTERS IN BOOKS

Chapter 3: *Lina Basel-Vanagaite, Gabrielle J. Halpern and Lutfi Jaber*. General Health Topics Associated with Consanguinity; Genetic Disorders and Congenital Malformations; Benefits. Consanguinity – its Impact, Consequences and Management, Bentham Science Publishers, Editors Lutfi A. Jaber, Gabrielle J. Halpern, 2014

Chapter 8: *Gabrielle J. Halpern, Lina Basel-Vanagaite and Lutfi Jaber*. Future Strategies 2 – Genetic Perspectives – Counseling, Screening, Testing, Research, and Intervention. Consanguinity – its Impact, Consequences and Management, Bentham Science Publishers, Editors Lutfi A. Jaber, Gabrielle J. Halpern, 2014

D.1.INVITED PAPERS IN SCIENTIFIC MEETINGS

1. Basel-Vanagaite L. Laboratory genetic testing services in Israel – an overview (2006). 8th Baltic Congress of Laboratory Medicine, Vilnius, Lithuania.

2. Basel-Vanagaite L. Working with specific populations: lessons learned (2007). NIH Mental Health workshop, New York, USA.

3. Basel-Vanagaite L. (2007) Genetics of mental retardation: present and future. 9th International Conference of the Baltic Child Neurology Association, Vilnius, Lithuania.

4. Basel-Vanagaite L, Attia R, Muncher L, Straussberg R, Pasmanik-Chor M, Rainshtein L, Magal N, Taub E, Rechavi G, Simon AJ, Shohat M (2008). Identification of two novel genes causing cognitive impairment and an overview of molecular mechanisms contributing to mental retardation. 5th Congress of the Federation of the Israel Societies for Experimental Biology (FISEB).

5. 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 (2008). Autosomal recessive ichthyosis with hypotrichosis (ARIH) caused by a mutation in ST14, encoding type II transmembrane serine protease matriptase.

Gordon research conference, Ventura, USA

6. Basel-Vanagaite L. Genetics of mental retardation (2009). 6th Baltic Congress of Neurology, Vilnius, Lithuania

D.2. PAPERS PRESENTED AT SCIENTIFIC MEETINGS PUBLISHED AS PROCEEDINGS

1. Oskato R, Bar Shira A, Vanagaite L, Ziv Y, Ehrlich S, Rotman G, McConville CM, Chakravarti A, and Shiloh Y (1993). Ataxia-telangiectasia: allelic association with 11q22-23 markers in Moroccan-Jewish patients. 43rd Annual Meeting of the American Society of Human Genetics, New Orleans, LA.

2. Kleiman S, Avigad S, Vanagaite L, David M, Shmuelevitz A, Eisensmith RC, Brand N, Schwartz G, Woo SLC, and Shiloh Y (1993). Origins of hyperphenylalaninemia in Israel. 43rd Annual Meeting of the American Society of Human Genetics, New Orleans, LA.

3. Shiloh Y, Ziv Y, Savitsky K, Bar-Shira A, Gilad S, Vanagaite L, Uchenik V, Smit S, Patanjali SR, Tagle D, Simmons A, Clines G, Collins FS, Weissman S, Lovett M and Rotman G (1994). Genetic, physical and functional analysis of the ataxia-telangiectasia locus on chromosome 11q22-23. 44th Annual Meeting of the American Society of Human Genetics, Montreal, Canada.

4. Shiloh Y, Savitsky K, Barshira A, Gilad S, Ziv Y, Tagle DA, Sfez S, Uziel T, Pecker I, Vanagaite L, Smith S, Harnik R, Collins FS, Rotman G (1995). Ataxia-telangiectasia – from phenotype to gene, on to biology and back to the patient. 45th Annual Meeting of the American Society of Human Genetics, New Orleans, LA

5. Shenhav R, Sayag E, Basel L, Halpern G, Rotchild A, Frydman M and Shohat M. (2001). Coarse face, dysostosis multiplex, progressive laryngeal obstruction and severe psychomotor retardation - a new type of autosomal recessive storage disease? 51th Annual Meeting of the American Society of Human Genetics, San Diego, US.

6. Basel-Vanagaite L, Klinger G, Shohat M, Udler Y, Levit O, Linder N and Sirota L. (2001) Congenital hydrocephalus, ambiguous genitalia in males and early lethality: a new syndrome? 51th Annual Meeting of the American Society of Human Genetics, San Diego, US.

7. Basel-Vanagaite L, Marcus N, Klinger G, Shohat M, Levit O, Karmazin B, Legum C and Sirota L. (2001). A new syndrome of simplified gyral pattern, micromelia, dysmorphic features, hypoparathyroidism and early death. 10th International Congress of Human Genetics, Vienna, Austria

8. Basel-Vanagaite L, Kornecki A, Ludman M, Ben-Ari Y, Merlob P. (2002) Single central incisor, pyriform aperture stenosis, midfacial hypoplasia and limb abnormalities: a new syndrome? 34th European Human Genetics Conference, Strasbourg, France.

9. Levin L, Magal N, Basel-Vanagaite L, Halpern G and Shohat M. (2002). Autosomal recessive cerebellar agenesis maps to chromosome 22q11 in an Israeli Arab family. 52th Annual Meeting of the American Society of Human Genetics, Baltimore, US

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11. Shohat M, Straussberg R, Ovadia H, Kaplan A, Magal N, Shorer Z, Shalev H, Walsh CA and Basel-Vanagaite L. (2003) Mapping of a new locus for autosomal recessive nonsyndromic mental retardation in the chromosomal region 19p13.12-p13.2: further genetic heterogeneity. 53th Annual Meeting of the American Society of Human Genetics, Los Angeles, US.

12. Basel-Vanagaite L, Atia R, Magal N, Shohat T, Taub E, Shohat M. (2004) Mapping of a new autosomal recessive non-syndromic hearing loss locus to chromosome 5q13. 54th Annual Meeting of the American Society of Human Genetics, Toronto, Canada.

13. Shoshani N, Basel-Vanagaite L, Liraz-Zaltsman S, Biegon A, Rechavi G, Amariglio N, Simon AJ and Shohat M. Distribution and immuno-localization of the NPC protein p62 in human and mouse brains (2007). 57th Annual Meeting of the American Society of Human Genetics, San Diego, US.

14. 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. (2006) Mutated nup62 causes autosomal recessive infantile bilateral striatal necrosis. 11th International Congress of Human Genetics, Brisbane, Australia.

15. 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.(2005) The CC2D1A, a member of a new gene family with C2 domains, is involved in autosomal recessive nonsyndromic mental retardation. 55th Annual Meeting of the American Society of Human Genetics, Salt Lake City, US.

16. 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. (2006) Mutated nup62 causes autosomal recessive infantile bilateral striatal necrosis. 38th European Human Genetics Conference, Amsterdam, Netherlands.

17. Van Maldergem L, Yuksel-Apak M, Kayserili H, Seemanova E, Giurgea S, Vigneron J, Greally M, Leao-Teles E, Basel-Vanagaite L, Jaeken J, Mundlos S, Dobyns WB (2007). The Debré type of autosomal recessive cutis laxa is associated with brain dysgenesis or neurodegeneration and defective N-glycosylation. 56th

Annual Meeting of the American Society of Human Genetics, San Diego, US.

18. Basel-Vanagaite L, Taub E, Rainshtein L, Drasinover V, Magal N, Zlotogora J, Shohat M. (2008) Preventive genetic screening in the isolated community: lessons learned. 40th European Human Genetics Conference, Barcelona, Spain

19. Maydan G, Noyman I, Har-Zahav A, Ben Neriah Z, 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 (2010). A novel GPI-deficiency related disorder: multiple congenital anomalies-hypotonia-seizures syndrome is caused by a mutation in PIGN. 43th European Human Genetics Conference, Amsterdam, Netherlands

20. Basel-Vanagaite L, Karlinsky L, Wolf L, Shohat M. Computer assisted estimation of the prevalence of dysmorphic features in the general population (2011). 12th International Congress of Human Genetics/61st ASHG Annual Meeting, Montreal, Canada

21. Wolf L, Basel-Vanagaite L, Karlinsky L, Shohat M, Allanson J. Recognizing adult Aarskog-Scott syndrome carrier females based on craniofacial measurements using interaction variables and a surrogate covariance matrix (2012). European Conference of Human Genetics

22. Basel-Vanagaite L, Karlinsky L, Wolf L, Shohat M, Skinner C, Rogers C, Stevenson R, Schwartz CM, Graham JM. Computer-aided facial recognition of individuals with FG (Opitz-Kaveggia) syndrome caused by p.Arg961Trp mutation in MED12 (2012). 62nd Annual Meeting of the American Society of Human Genetics, San Francisco, US

23. Basel-Vanagaite L, Karlinsky L, Wolf L, Deardorff M, Shohat M, Krantz I. Computer-aided facial recognition of Cornelia de Lange syndrome: a comparison to the recognition by human experts (2012). David W. Smith workshop, Buford Georgia, US

24. Borck G, Dallapiccola B, Ramirez-Solis R, Segref A, Thiele H, Edwards A, Arends MJ, Miro X, White JK, Desir J, Abramowicz M, Dentici ML, Hofmann K, Har-Zahav A, Ryder E, Karp NA, Ingham NJ, Nuernberg G, Abdelhak S, Pasmanik-Chor M, Konen O, Kelley R, Shohat M, Nuernberg P, Flint J, Steel KP, Hoppe T, Kubisch C, Adams DJ, Basel-Vanagaite L. Biallelic mutations of a ubiquitin-ligase-encoding gene cause an Ohdo-like intellectual disability syndrome (2013). 62nd Annual Meeting of the American Society of Human Genetics, San Francisco, US

25. Basel-Vanagaite L, Wolf L. Is a computer-based facial dysmorphology analysis ready for the clinic (2014)? 63rd Annual Meeting of the American Society of Human Genetics, San Diego, US

26. Cohen L, Orenstein N, Weisz- Hubshman M, Maya I, Behar D, Kahana S, Tene T, Feingold-Zadok M, Shohat M, Basel-Vanagaite L. Homozygosity mapping using SNP microarray as a useful diagnostic tool in consanguineous populations (2015). European Human Genetics Conference, Glasgow, Scotland

27. Basel-Vanagaite L, Lyonnet S, Cormier-Daire V, Rio M, Amiel J, Wolf L. FDNA Performance of Facial Dysmorphology Novel Analysis: comparison to molecular testing-based diagnostics. 64th Annual Meeting of the American Society of Human Genetics, Baltimore, US

28. Maya I, Kahana S, Tene T, Jakobson S, Yeshaya J, Shohat M, Basel-Vanagaite L. Genomic imbalances in fetuses and patients with congenital heart malformation. 64th Annual Meeting of the American Society of Human Genetics, Baltimore, US

D.3. ABSTRACTS

1. Shiloh Y, Ziv Y, Bar-Shira A, Vanagaite L, Oskato R, Savitsky K, Shor Y, Bar-Am I, Avivi L, Bentley D, and Rotman G. (1993) Ataxia-telangiectasia: Positional and functional cloning converge. International Seminar on Human Genome Diversity and Congenital Disorders. Kfar-Saba, Israel.

2. Shiloh Y, Ziv Y, Bar-Shira A, Vanagaite L, Savitsky K, Oskato R, Aharonson S, Ochnik V, Bar-Am I, Avivi L and Rotman G. (1994) Ataxia-telangiectasia: The race for the gene. Annual Meeting of the Israel Society for Biochemistry and Molecular Biology, Ramat Gan, Israel.

3. Rotman G, Savitsky K, Bar-Shira A, Vanagaite L, Ziv Y, Aharonson S, Smith S, Ochnik V, and Shiloh Y. (1994) Genetic and physical mapping of the A-T locus on chromosome 11q22-23. 6th International Workshop on Ataxia-telangiectasia, Birmingham, England.

4. Shiloh Y, Ziv Y, Bar-Shira A, Savitsky K, Vanagaite L, Gilad S, Smith S, Ochnik V, and Rotman G. (1994) Ataxia-telangiectasia: Positional and complementation cloning converge. 10th Meeting of the European Society for Neurochemistry, Jerusalem, Israel. J. Neurochem, Vol. 63, Suppl. 1, p. S4.

5. Bar-Shira A, Savitsky K, Gilad S, Patanjali SR, Rotman G, Ziv Y, Vanagaite L, Simmons A, Clines G, Tagle D, Collins FS, Weissman S, Lovett M, and Shiloh Y. (1994) A transcriptional map of the ataxia-telangiectasia locus on chromosome 11q22-23. 4th International Workshop on Transcribed Sequences, Montreal, Canada.

6. Shiloh Y, Bar-Shira A, Savitsky K, Ziv Y, Vanagaite L, Gilad S, Uchenik V, Smith S, and Rotman G. (1994) Ataxia-telangiectasia: From Phenotype to genes. 2nd France-Israel Binational Meeting on Human Genetic Diseases and Gene Therapy, Jerusalem, Israel.

7. Shiloh Y, Kleiman S, Avigad S, Vanagaite L, Eisensmith RC, and Woo SLC. (1993) Genetic diversity of hyperphenylalaninemia in Israel. International Symposium on Hyperphenylalaninemia in the Mediterranean Countries, Torina, Sicily.

8. Williams LJ, O'Rourke AW, Hurst JA, Basel-Vanagaite L, Seller A, Wilkie AOM, Lester T. (2007). A Molecular Diagnostic Service for Craniofrontonasal Syndrome. BSHG meeting, York, UK.

9. Basel-Vanagaite L, Rainshtein L, Inbar D, Gothelf D, Hennekam R, and Straussberg R. Autosomal Recessive Mental Retardation Syndrome with Anterior Maxillary Protrusion and Strabismus: MRAMS Syndrome (2007). 18th European Meeting on Dysmorphology, Strasburg, France

10. 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. (2010), British Society of Human Genetics annual conference, Warwick, UK

11. Basel-Vanagaite L, Karlinsky L, Wolf L, Deardorff M, Shohat M, Krantz I. Computer-aided facial recognition of Cornelia de Lange syndrome: a comparison to the recognition by human experts (2012). David W. Smith workshop, Atlantic city, 2012

12. 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 (2012). 15th Manchester dysmorphology meeting, Manchester, UK

13. Basel-Vanagaite L, Wolf L. Is a computer-based facial dysmorphology analysis ready for the clinic (2014)? 16th Manchester dysmorphology meeting, Manchester, UK

14. Basel-Vanagaite L, Wolf L. Performance of facial dysmorphology novel analysis: comparison to molecular testing-based diagnostics (2016)? 17th Manchester dysmorphology meeting, Manchester, UK

E. ITEMS IN ENCYCLOPEDIAS

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

H. Member of Editorial Board/ Reviewer (International Journals)

Reviewer	Clinical Genetics
Reviewer	Journal of Neuroscience Research
Reviewer	Journal of Perinatology
Reviewer	European Journal of Neuroscience
Reviewer	American Journal of Human Genetics

Reviewer	BMC Medical Genetics Journal
Reviewer	Psychiatry Research
Reviewer	British Journal of Dermatology
Reviewer	Clinical Pediatrics
Reviewer	Molecular Diagnosis & Therapy
Reviewer	Journal of Medical Genetics
Reviewer	Clinical and Experimental Dermatology
Reviewer	Pediatric Endocrinology Reviews
Reviewer	Genetics in Medicine
Reviewer	National Organization for Rare Disorders, Inc (NORD), USA
Reviewer	Annals of Human Genetics
Reviewer	American Journal of Medical Genetics
Reviewer	European Journal of Human Genetics
Reviewer	Neurogenetics
Reviewer	Neuropediatrics
Reviewer	Human Genetics
Reviewer	European Journal of Medical Genetics
Reviewer	Orphanet Journal of Rare Diseases
Reviewer	Human Hereditary
Reviewer	Israeli Science foundation
Reviewer	Chief Scientist foundation, Israeli Ministry of Health
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