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Current position: Professor of Molecular Genetics
Western Australian Institute of Medical Research
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Education and professional qualifications

Medical School:

- **Graduate** - Higher Medical Institute, Sofia, Bulgaria
1964-1971
- **Postgraduate**
 - Sofia Postgraduate Medical Institute, Department of
Clinical Chemistry, 1971-1973
 - Moscow Institute of Pediatrics and Pediatric Surgery,
1976-1979

Degrees and Qualifications:

- **MD** - Sofia Higher Medical Institute, 1971
- **PhD** - Degree awarded by the Higher Accreditation
Commission of the USSR in 1979

Fellowships and Awards:

Alexander von Humboldt Research Foundation Fellowship:

- Institute of Physiological Chemistry, Medical Faculty,
University of Münster, Germany - 1979-1980
- Institute of Human Genetics, Medical Faculty,
University of Münster, Germany - 1988-1990

Spinoza Visiting Professorship

- University of Amsterdam, The Netherlands – 2003

Visiting Professor

- Medical University, Sofia, Bulgaria - 2005

Previous professional appointments:

- Associate Professor
Co-Director, Centre for Human Genetics
Edith Cowan University (ECU), Perth, Australia 1998-2002
- Senior Lecturer, Department of Human Biology, ECU
1996-1998

- Lecturer Level B, Department of Human Biology, ECU 1994-1996
- Visiting scientist, Department of Genetics, Stanford University Medical School, Stanford California 1993
- Associate Professor, Laboratory of Molecular Pathology, Faculty of Medicine, Medical Academy Sofia, Bulgaria 1989-1993
- Senior Lecturer and Head of Biochemical Laboratory, Department of Clinical Genetics, Institute of Pediatrics, Medical Academy, 1980 - 1988
- Lecturer and Head of Laboratory for Inborn Errors of Metabolism and National Neonatal Screening Program, Institute of Pediatrics, Medical Academy, Sofia 1977-1980
- Lecturer, Department of Clinical Chemistry, Sofia Postgraduate Medical Institute 1974-1977

Follows my full list of publications. Those relevant to the topic start at # 74.

Luba Kalaydjieva – Publications

Listed on the ISI Web of Knowledge “Most Cited Scientists in Molecular Biology and Genetics” (<http://esi.isiknowledge.com/home.cgi>)

1. Kalaydjieva L (1969) Laboratory diagnosis of phenylketonuria (Oligophrenia phenylpyruvica, Fölling disease): a review. *Savremenna medicina* XX: 587-591 (in Bulgarian).
2. Kalaydjieva L, Danev S (1972) Our experience with screening tests for the early detection of phenylketonuria. *Savremenna medicina* XXIII: 14-18 (in Bulgarian).
3. Stateva S, Kisselkova V, Christov L, Kalaydjieva L, Koeva N, Shuleva M (1977) First results of phenylketonuria screening in Bulgaria. *Pediatrics Sofia* XVI: 573-579 (in Bulgarian).
4. Kalaydjieva L, Stanchev Z (1978) The genetic mucopolysaccharidoses and mucopolipidoses: classification and diagnosis: a review. *Pediatrics Sofia* XVII: 230-238 (in Bulgarian).
5. Peneva L, Kalaydjieva L, Stanchev Z (1978) Generalized gangliosidosis GM1 type I (Norman Landing disease): a case report. *Pediatrics Sofia* XVII: 238-247 (in Bulgarian).
6. Kalaydjieva LV (1978) Characterization of liver 4-MU- β -galactosidase. In “*Biochemical basis of genetic disorders of carbohydrate metabolism*”, Moscow, Ministry of Health of the Russian Federation and Institute of Biological Chemistry of the Academy of Medical Sciences of the USSR, pp 75-81 (in Russian).
7. Barashnev Yu I, Shaposhnikov AM, Korneichuk VV, Khalchitsky SE, Kalaydjieva LV (1978) Phenylketonuria in combination with leukodystrophy. *Ohrana materinstva i detstva* 7:46-49 (in Russian).
8. Veltishev Yu E, Kalaydjieva LV (1979) Properties of lysosomal β -galactosidase and molecular genetics of gangliosidoses GM1. *Voprosy medicinskoj himii* 6:709-715 (in Russian)
9. Kalaydjieva LV, Kremensky IM, Petkov PE (1979) Serum and leukocyte lysosomal hydrolases in mucopolipidosis II. *Comptes rendus de l'Académie bulgare des Sciences* 32: 1299-1302.
10. Petkov PE, Kalaydjieva LV (1979) Ultrastructural characteristics of conjunctival fibrocytes in mucopolipidosis II. *Comptes rendus de l'Académie bulgare des Sciences* 32: 1437-1440.
11. Kremensky I, Kalaydjieva L, Savova R, Radivenska A, Kumanova R (1979) Laboratory diagnosis of chronic Gaucher disease. *Pediatrics Sofia* XVIII: 322-326 (in Bulgarian).
12. Simeonov E, Kalaydjieva L, Petkov P, Kremensky I (1980) Mucopolipidosis II (I-cell disease): a case report. *Pediatrics Sofia* XIX: 206-211 (in Bulgarian).

13. Stantchev Z, Simeonov E, Peneva L, Kalajdjieva L, Petkov P, Kremensky I (1981) Les mucopolidoses. *Archives de l'Union Médicale Balkanique* XIX:246-247 (in French).
14. Kalaydjieva L, Cantz M (1981) Subcellular distribution of lysosomal hydrolases in mucopolidosis III and a mucopolidosis III variant. *J Inher Metab Dis* 4:145.
15. Cantz M, Mendla K, Baumkötter J, Kalaydjieva L (1981) Sialidosis: the specificity of the sialidase deficiency. *Perspectives Inher Metab Dis* 4:219-232.
16. Kremensky I, Kalaydjieva L (1981) Biochemical diagnosis of liver glycogenoses. *Pediatrics Sofia* XX: 609-613 (in Bulgarian).
17. Kalaydjieva L, Sinigerska I, Varon R, Kremensky I, Panayotova V, Simeonov E, Liharska K (1983) Laboratory diagnosis of hereditary mucopolysaccharidoses – our experience. *Pediatrics Sofia* XXII:513-519 (in Bulgarian).
18. Bobev D, Ninyo S, Kremensky I, Russanova M, Kalaydjieva L, Liharska K (1983) N-acetyl- β -glucosaminidase isoenzymes in acute lymphoblastic leukemia in childhood. *Pediatrics Sofia* XXII:520-522 (in Bulgarian).
19. Hadjiev A, Simeonov E, Kalaydjieva L, Krachunova M, Varon R, Liharska K, Kremensky I, Kincheva V (1984) Obstetrical problems in early amniocentesis: discussion of our first cases. *Obsterics and Gynecology* XXIII:1-6 (in Bulgarian).
20. Damyanova Tzv, Damyanova M, Gitzelman R, Koprivarova K, Radeva B, Kremensky I, Kalaydjieva L (1985) Clinical observations in children with galactosemia detected by the mass neonatal screening program for inborn errors of metabolism during the period 1979-1984. *Pediatrics Sofia* XXIV:13-17 (in Bulgarian).
21. Varon R, Kalaydjieva L, Simeonov E, Liharska K, Klaier W (1985) Krabbe disease – biochemical diagnosis. *Pediatrics Sofia* XXIV:58-61 (in Bulgarian).
22. Kalaydjieva L, Varon R, Kremensky I (1985) Laboratory diagnosis of metachromatic leukodystrophy. *Pediatrics Sofia* XXIV:61-64 (in Bulgarian).
23. Simeonov E, Hadzhiev A, Varon R, Kalaydjieva L, Liharska K, Brankova M (1986) Prevention of neural tube defects *Pediatrics Sofia* XXV:45-50 (in Bulgarian).
24. Kumanova R, Radivenska A, Zelev H, Varon R, Kalaydjieva L (1986) Peculiarities in the clinical course of Niemann-Pick disease. *Pediatrics Sofia* XXV:74-79 (in Bulgarian).
25. Lalov V, Issaev V, Perchinska E, Kalaydjieva L (1986) Generalized gangliosidosis: a case report. *Pediatrics Sofia* XXV:80-85 (in Bulgarian).
26. Kalaydjieva L, Simeonov E, Valov V, Zekova N (1986) A registry of congenital malformations – the first year experience of the Institute of Obstetrics and Gynecology: I. Diagnostic aspects and prevention. *Pediatrics Sofia* XXV:44-48 (in Bulgarian).
27. Kalaydjieva L (1986) A registry of congenital malformations – the first year experience of the Institute of Obstetrics and Gynecology: II. Organizational aspects. *Pediatrics Sofia* XXV:49-52 (in Bulgarian).
28. Varon R, Kalaydjieva L, Simeonov E, Hadjiev A, Krachunova M, Liharska K, Kincheva V, Pechenyakova N, Andreev A, Kremensky I (1986) Prenatal diagnosis of genetic defects. *Acta Medica Bulgarica* XIII: 37-41.
29. Kalaydjieva L (1987) Fighting the unknown: how much do practicing physicians know about clinical genetics. *Pediatrics Sofia* XXVI:83-89 (in Bulgarian).
30. Kalaydjieva L, Kremensky I, Varon R (1987) GM1 gangliosidosis. *Pediatrics Sofia* XXVI:39-45 (in Bulgarian).
31. Kalaydjieva L, Plageras P, Kremensky I (1987) Enzymatic prenatal diagnosis of Cystic Fibrosis: a review. *Savremenna Medicina* XXXVIII: 5-10 (in Bulgarian).
32. Kremensky I, Plageras P, Kalaydjieva L (1987) Prenatal enzyme diagnosis of Cystic Fibrosis: II. Assessment of the analytical reliability. *Pediatrics Sofia* XXVI:12-18 (in Bulgarian).
33. Kalaydjieva L, Kremensky I, Plageras P (1987) Prenatal enzyme diagnosis of Cystic Fibrosis: II. Assessment of the diagnostic reliability. *Pediatrics Sofia* XXVI:19-25 (in Bulgarian).

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35. Kalaydjieva L, Argiris E, Ninyo S (1989) Molecular genetics of β -thalassemia in Bulgaria. *Savremenna Medicina* XL: 25-30 (in Bulgarian).
36. Kalaydjieva L, Eigel A, Horst J (1989) The molecular basis of β -thalassemia in Bulgaria. *J Med Genet* 26: 614-618.
37. Plageras P, Kalaydjieva L (1989) Polymorphic DNA analysis in Bulgarian families with Cystic Fibrosis. *Savremenna Medicina* XL: 9-14 (in Bulgarian).
38. The Cystic Fibrosis Genetic Analysis Consortium (1990) Worldwide survey of the delF508 mutation. *Amer J Hum Genet* 47: 354-359.
39. European Working Group on CF Genetics (1990) Gradient of distribution in Europe of the major CF mutation and its associated haplotype. *Hum Genet* 85: 450-454.
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41. Kalaydjieva L, Dworniczak B, Aulehla-Scholz C, Kremensky I, Bronzova J, Eigel A, Horst J (1990) Classical phenylketonuria in Bulgaria: RELP haplotypes and frequency of the major mutations. *J Med Genet* 27: 742-745.
42. Dworniczak B, Aulehla-Scholz C, Kalaydjieva L, Bartholome K, Grudda K, Horst J (1991) Aberrant splicing of phenylalanine hydroxylase mRNA: the major cause for phenylketonuria in parts of southern Europe. *Genomics* 11: 242-246.
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45. Kalaydjieva L, Plageras P, Horst J (1991) Further evidence of cystic fibrosis heterogeneity in southern Europe. *Hum Hered* 41: 65-67.
46. Kalaydjieva L, Dworniczak B, Kucinkas V, Yurgeliavicius V, Kunnert E, Horst J (1991) Geographical distribution gradients of the major PKU mutations and the linked haplotypes. *Hum Genet* 86: 411-413.
47. Kalaydjieva L, Dworniczak B, Aulehla-Scholz C, Devoto M, Romeo G, Stuhmann M, Horst J (1991) Phenylketonuria mutation in southern Europeans. *Lancet* 337: 865.
48. Kalaydjieva L, Dworniczak B, Aulehla-Scholz C, Devoto M, Romeo G, Stuhmann M, Kucinkas V, Yurgelyavicius V, Horst J (1991) Silent mutations in the phenylalanine hydroxylase gene as an aid to the diagnosis of phenylketonuria. *J Med Genet* 28: 686-690.
49. Kalaydjieva L, Angelicheva D, Galeva I, Konstantinova K, Lalov V (1991) Cystic fibrosis in Bulgaria. *J Med Genet* 28: 807.
50. Dworniczak B, Kalaydjieva L, Pankoke S, Aulehla-Scholz C, Allen G, Horst J (1992). Analysis of exon 7 of the human phenylalanine hydroxylase gene: a mutation hot spot? *Hum Mut* 1: 138-146.
51. Kalaydjieva L, Dworniczak B, Kremensky I, Koprivarova K, Radeva B, Milusheva R, Aulehla-Scholz C, Horst J (1992). Heterogeneity of mutations in Bulgarian phenylketonuria haplotype 1 and 4 alleles. *Clin Genet* 41: 123-128.
52. Kalaydjieva L, Kremensky I (1992) Screening for phenylketonuria in a totalitarian state. *J Med Genet* 29: 656-658.
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55. Savov A, Angelicheva D, Jordanova A, Eigel A, Kalaydjieva L (1992) High percentage acrylamide gels improve resolution in SSCP analysis. *Nucl Acids Res* 20: 6741-6742.
56. The Cystic Fibrosis Genotype-Phenotype Consortium (1993) Correlation between genotype and phenotype in cystic fibrosis: analysis of seven common mutations. *N Engl J Med* 329: 1308-1313.
57. Horst J, Eigel A, Kalaydjieva L, Dworniczak B (1993). Phenylketonuria in Germany - molecular heterogeneity and diagnostic implications. *Dev Brain Dysfunc* 6: 32-39.
58. Kalaydjieva L, Dworniczak B, Kremensky I, Radeva B, Horst J (1993). Population genetics of phenylketonuria in Bulgaria. *Dev Brain Dysfunc* 6: 39-46.
59. Ramsay M, Williamson R, Estivill X, Wainwright BJ, Ho M-F, Halford S, Kere J, Savilahti E, de la Chapelle A, Schwartz M, Schwartz M, Super M, Farndon P, Harding C, Meredith L, Al-Jader L, Ferec C, Claustres M, Casals T, Nunes V, Gasparini P, Savoia A, Pignatti PF, Novelli G, Bennarelli M, Dallapiccola B, Kalaydjieva L, Scambler PJ (1993). Haplotype analysis to determine the position of a mutation among closely linked DNA markers. *Hum Molec Genet* 2: 1007-1014.
60. Morral N, Bertranpetit J, Estivill X, Nunes V, Casals T, Gimenez J, Reis A, Varon-Mateeva R, Macek M, Kalaydjieva L, Angelicheva D, Dancheva R, Romeo G, Rossu MP, Garneone S, Restagno G, Ferrari M, Magnani C, Claustres M, Desgeorges M, Schwartz M, Dallapiccola B, Novelli G, Ferec C, de Arce M, Nermeti M, Kere J, Anvret M, Dahl N, Kadasi L (1994) The origin of the major cystic fibrosis mutation (delF508) in European populations. *Nature Genet* 7: 169-175.
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62. Bronzova J, Todorova A, Kalaydjieva L (1994) Detection of carriers of deletions in the dystrophin gene in Bulgarian DMD/BMD families. *Hum Genet* 93: 170-174.
63. The Cystic Fibrosis Genetic Analysis Consortium (1994) Population variation of common CF mutations. *Hum Mut* 4:167-177.
64. Mercier B, Lissens W, Novelli G, Kalaydjieva L, De Arce M, Kapranov N, Canki Klain N, Estivill X, Palacio A, Cashman S (1994) A cluster of cystic fibrosis mutations in exon 17b of the CFTR gene: a site for rare mutations. *J Med Genet* 31: 731-734.
65. Savov A, Mercier B, Kalaydjieva L, Ferec (1994) Identification of six novel mutations in the CFTR gene of patients from Bulgaria by screening the twenty seven exons and exon/intron boundaries using DGGE and direct DNA sequencing. *Hum Molec Genet* 3: 57-60.
66. Savov A, Jordanova A, Gavrilov D, Angelicheva D, Kalaydjieva L (1994) A novel missense mutation in the second nucleotide binding domain of the CFTR gene in a Bulgarian cystic fibrosis patient. *Hum Molec Genet* 3: 513-514.
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74. Kalaydjieva L, Hallmayer J, Chandler D, Savov A, Nikolova A, Angelicheva D, King R, Ishpekova B, Honeyman K, Calafell F, Shmarov A, Petrova J, Turnev I, Hristova A, Moskov M, Stancheva S, Petkova I, Bittles A, Georgieva V, Middleton L, Thomas PK (1996) Gene mapping in Gypsies identifies a novel demyelinating neuropathy on 8q24. *Nature Genet* 14: 214-216.
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