

Prof. Imad Mohammad Dweikat

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EDUCATION

2004-2005	Metabolic training/The Hospital for Sick Children, Toronto, Canada
1994-1997	Pediatric Residency at Makassed Hospital
1992-1994	Pediatric Residency at Augusta Victoria Hospital
1990-1991	Internship at Jordan University Hospital
1984-1990	Bachelor of Medicine at Medical School Baghdad University, Baghdad-Iraq

QUALIFICATIONS

2024	Professor of Metabolic Diseases
2016	Associate Professor
2004-2005	Training in the laboratory diagnosis of Metabolic Diseases
1999	Palestinian Board of Pediatrics
1997	Jordanian Board of Pediatrics
1990	MBBS – Baghdad University

MEMBERSHIPS

Member of the Society for Inherited Metabolic Disorders (SIMD)

Member of the Society for Study of Inborn Errors of Metabolism (SSIEM)

Member of the Jordanian Medical Association Member of the Pediatric

Society - Palestine

PROFESSIONAL EXPERIENCE

Professor of Metabolic Diseases	March 2024
February, 2023 – Present	Academic staff at AAUP
September, 2020 – February, 2023	Dean, Faculty of Medicine, Arab American University- Palestine
September, 2020 – Present	Member of the Palestinian Medical Council
March, 2020-September, 2020	Head of Pediatric Department & Metabolic Consultant at Palestine Medical Complex
2019- September, 2020	Head of scientific committee and member of higher scientific committee at Palestine Medical Council
2017-2020	Associate Professor at Al-Quds University
2016-2017	Dean of Faculty of Medicine and Health Sciences & Associate Professor at An-Najah National University
1997-2013	Pediatric Consultant at Makassed Hospital
2004-2013	Metabolic disease Consultant at Makassed Hospital
2004-2020	Head of the metabolic unit at Makassed Hospital
2013-2017	Pediatric and Metabolic Consultant at An-Najah National University Hospital
2013-2014	Dean of Faculty of Medicine and Health Sciences at An-Najah National
2014-2017	Pediatric Advanced Life Support Provider of the American Heart Association
2007-2013	Tutor of Pediatric Advanced Life Support in Palestine
1999-2013	Assistant Professor of Pediatrics at Al-Quds University

RESEARCH & PUBLICATIONS

1. Imad M. Dweikat, Hamza A. Abdul-Hafez, Alaa Zayed, Moien Kanaan, Hanin Kasem, Rami Bzour (2025). Molecular characterization, clinical phenotype, and neurological outcome of twelve Palestinian children with beta-ketothiolase deficiency: report of two novel variants in the ACAT1 gene. BMC Medical Genomics. Published on July 1, 2025. This original research describes the clinical phenotype, genotype, and outcome of this rare ketone body utilization disorder. Two novel variants in the ACAT1 gene were reported, expanding the genotype of this rare disease.
2. Imad Dweikat, Moien Kanaan, Hanin Kassem, Huthaifa H. Ahmad (2025). Genotype and Clinical Phenotype of Monocarboxylate Transporter 1 Deficiency in Three Palestinian Children: Report of Two Novel Variants in the *SLC16A1* Gene. American Journal of Medical Genetics Part A. Published online on June 4, 2025. This article describes the genotype and clinical phenotype of three Palestinian children with MCT1 deficiency from two unrelated families. The clinical phenotypes and biochemical abnormalities were similar across all three patients, characterized by acute recurrent vomiting, severe dehydration, metabolic acidosis, and hyperuricemia. Two novel variants were reported, expanding the genotype of this rare disease.
3. Tamer Hodrob, Alaaeddin Abusalameh, Ibrahim Ismail, Imad Dweikat, Sarah Abu Rmeilah, Mutaz Sultan, Bassam Abu Libdeh, Abd-Al-Salam Abu Libdeh, Shaher Shweiki, Nadirah Damseh (2024). Genetic, Clinical, and Biochemical Characterization of a Large Cohort of Palestinian Patients with Fanconi-Bickel Syndrome. Clinical Genetics. Accepted for publication on November 4, 2024. *This cohort presents clinical, biochemical, and genetic data from 20 FBS individuals in Palestine. It identifies five pathogenic variants, including three novel mutations not previously reported in the literature.*
4. Reham Khalaf-Nazzal, Imad Dweikat, Nishanka Ubeyratna, James Fasham, Maysa Alawneh, Joseph Leslie, Mosab Maree, Adam Gunning, Deyala Z. Zayed, Nikol Voutsina, Lucy McGavin, Reem Sawafta, Martina Owens, Wisam Baker, Peter Turnpenny, Fida' Al-Hijawi, Emma L. Baple, Andrew H. Crosby, Lettie E. Rawlins (2024). TECPR2-related hereditary sensory and autonomic neuropathy in two siblings from Palestine. American Journal of Medical Genetics A. Accepted for publication on February 2024. *This study identifies two Palestinian siblings presenting with acute encephalopathy and severe life-threatening multi-organ dysfunction/ failure manifesting with liver injury and kidney failure. Transient myopathy has not previously been reported in association with TECPR2-HSAN*
5. James Fasham, Antje K. Huebner, Lutz Liebmann, Reham Khalaf-Nazzal, Reza Maroofian, Nderim Kryeziu, Saskia B. Wortmann, Joseph S. Leslie, Nishanka Ubeyratna, Grazia M.S. Mancini, Marjon van Slegtenhorst, Martina Wilke, Tobias B. Haack, Hanan Shamseldin, Joseph G. Gleeson, Mohamed Almuhaizea,

Imad Dweikat, Bassam Abu-Libdeh, Muhannad Daana, Maha S. Zaki, Matthew N Wakeling, Lucy McGavin, Peter D. Turnpenny, Fowzan S. Alkuraya, Henry Houlden, Peter Schlattmann, Kai Kaila, Andrew H. Crosby, Emma L. Baple, Christian A. Hübner (2023). SLC4A10 mutation causes a neurological disorder associated with impaired GABAergic transmission. BRAIN journal. Accepted for publication on June, 6 2023.

This original article defines a novel characteristic neurodevelopmental disorder associated with biallelic pathogenic variants in SLC4A10 and highlight the importance of further analyses of the consequences of SLC4A10 loss-of-function for brain development, synaptic transmission and network properties

6. **Imad Dweikat**, Reham Khalaf-Nazzal. Clinical heterogeneity of Hyperornithinemia-Hyperammonemia-Homocitrullinuria syndrome in thirteen Palestinian patients and report of a novel variant in the *SLC25A15* gene (2022). Frontiers in Genetics. Published on November 24, 2022.

I am the first and corresponding author of This original article which describes the heterogenous phenotype of HHH syndrome varying from severe neonatal presentation with encephalopathy and coagulopathy to adult-onset neurological manifestations. We identified a novel mutation in SLC25A15

7. Fajr M A Sarhan, Afnan W.M. Jobran, Ali Fayyad, Zaid Ghanim, **Imad Dweikat**, Shireen Elewie, Ala Mustafa Habboub (2022). Late onset hyperornithinemia, hyperammonemia, and homocitrullinuria syndrome, presenting as recurrent metabolic encephalopathy. Annals of Medicine and Surgery. Published online on November 8, 2022.

8. R Khalaf-Nazzal, **I Dweikat**, M Maree, M Alawneh, M Barahmeh, RT Doulani, M Qarareyah, M Qadi, A Dudin (2022). Prevalent MLC1 mutation causing autosomal recessive Megalencephalic Leukoencephalopathy in consanguineous Palestinian Families. Brain and Development Journal. Published online on April 16, 2022.

9. **I Dweikat**, B Abu-Libdeh, I Abu Libdeh, M Asshab, H Zitawi (2022). L-2-Hydroxyglutaric aciduria in two Palestinian siblings with a novel mutation in the *L2HGDH* gene. Journal of the Arab American University. Accepted for Publication on March 2, 2022.

I am the first and corresponding author of this article which describes a novel genotype and a distinct phenotype in two siblings with L-2-Hydroxyglutaric aciduria.

10. M A. Sharoor, F M. Lasorsa, V Porcelli, **I Dweikat**, M Antonietta De Noia, M Gur, G Agostino, A Shaag, T Rinaldi, G Gasparre, F Guerra, A Castegna, S Todisco, B Abu-Libdeh, O Elpeleg, L Palmieri (2022). PNC2 (*SLC25A36*) Deficiency Associated with the Hyperinsulinism/Hyperammonemia Syndrome. The Journal of Clinical Endocrinology & Metabolism.

I am a co-author of this clinical research article which reports for the first time a mutation in PNC2/SLC25A36 leading to HI/HA and provides functional evidence of the molecular mechanism responsible for this phenotype.

11. **I Dweikat**, O Thaher, A Abosleem, A Zeer, A Abo Mokh (2021). Niemann-Pick disease type C in Palestine: Genotype and phenotype of sixteen patients and report of a novel mutation in the *NPC1* gene. *BMC Medical Genomics* 14:228
I am the first and the corresponding author of this original article which describes the genotype and the genotype-phenotype correlation, and the outcome of 16 Palestinian patients with NPC. We reported a novel mutation in the NPC1 gene, further expanding the genotype of this rare neurovisceral disorder.
12. **I Dweikat**, N Qawasmi, A Najeeb, M Radwan (2021). Phenotype, genotype, and outcome of 25 Palestinian patients with hereditary tyrosinemia type 1. *Metabolism Open* 9 100083.
I am the first and the corresponding author of this original article which reports the phenotype-genotype correlation of 25 patients with hereditary tyrosinemia type 1 and describes the outcome in response to orfadin (NTBC) treatment.
13. F Alqarajeh, SA Abukhalaf, JO Omorodion, **IM Dweikat** (2020). Genotype and clinical phenotype in four patients with glutathione synthetase deficiency. *Meta Gene*, 2020.
I am the corresponding author of this original article which describes the genotype-phenotype of this rare autosomal recessive disorder which has been reported the literature in roughly 80 patients. Currently, there is little known about genotype-phenotype correlations in GSSD
14. Yeshaya Langer, Adi Aran, Suleyman Gulsuner, Bassam Abu Libdeh, Paul Renbaum, Dario Brunetti, Pedro-Filipe Teixeira, Tom Walsh, Sharon Zeligson, Roberta Ruotolo, Rachel Beeri, **Imad Dweikat**, Maher Shahrour, Ariella Weinberg-Shukron, Fouad Zahdeh, Enrico Baruffini, Elzbieta Glaser, Mary-Claire King, Ephrat Levy-Lahad, Massimo Zeviani, Reeval Segel (2018). Mitochondrial PITRM1 peptidase loss-of-function in childhood cerebellar atrophy. This article describes two brothers from a consanguineous Palestinian family presented with progressive spinocerebellar ataxia, mental retardation and psychotic episodes. Serial brain imaging showed severe progressive cerebellar atrophy. Whole exome sequencing revealed a novel mutation: pitrilysin metallopeptidase 1 (*PITRM1*) c.2795C>T, p.T931M, homozygous in the affected children and resulting in 95% reduction in *PITRM1* protein.
15. W Khraim, B Abu-Libdeh, S Ayesh, **I Dweikat** (2017). Clinical Heterogeneity of Glycine Encephalopathy in three Palestinian Siblings: A novel mutation in the Glycine Decarboxylase (GLDC) Gene. *Brain and Development*, 39 (7), 601-605.
I am the corresponding author of this original article, which describes the clinical, biochemical, and molecular characteristics of three siblings who have distinct clinical phenotypes with a novel mutation in the GLDC gene. This mutation further expands the genetic spectrum of Glycine Encephalopathy (Non-ketotic Hyperglycinemia) and adds to the evidence of clinical heterogeneity of the disease even in siblings with identical mutations.
16. KN Reham, GM Paula, **M D Imad**, O Iker, M Dominik, CM Félix (2017). Phenotypic and genetic Heterogeneity linked to the Magnesium Transport

Mediator CNNM2. Front. Hum. Neurosci. doi:
10.3389/conf.fnhum.2017.222.00035

I am one of the authors of this article and research project which describes heterogenous clinical phenotypes related to CNNM2 gene mutations and protein functions. In the first family, we report for the first time the involvement of CNNM2 in retinal photoreceptor development and function. CNNM4 is already described to be involved in retinal development. We also report for the first time the presence of a neurophenotype independent of magnesium status related to the CNNM2 protein mutation. It was presented as Conference Abstract: 2nd International Conference on Educational Neuroscience

17. A Abu-Libdeh, ID Wexler, I Dweikat, D Zangen, B Abu-Libdeh (2017). A Novel mutation in AVPR2 Gene in a Palestinian Family with Nephrogenic Diabetes Insipidus. Journal of Child Science 7 (01), e1-e3
I am one of the authors of this case report which describes a two-month old infant and older brother with nephrogenic diabetes insipidus with a novel missense mutation in AVPR2 gene.
18. AM Khalaf-Nazzal, Imad Dweikat, Arwa Maqboul (2016). Whole exome sequencing reveals complex inheritance patterns and identifies two gene mutations implicated in the development of Autism and Intellectual Disability in a consanguineous Palestinian family. Frontiers. neuroscience
I am the corresponding author of this study, we applied whole exome sequencing (WES) followed by segregation analysis and phenotype-genotype correlation to study genetic changes in three siblings of a highly consanguineous Palestinian family. They presented with neurodevelopmental phenotype evident during childhood. Our results confirm that CNNM2, which was previously implicated in dominant isolated hypomagnesemia is now causing a variable neurodevelopmental phenotype including Autistic spectrum disorder intellectual disability when inherited in an autosomal recessive manner.
19. IM Dweikat, IS Alawneh, SF Bahar, MI Sultan (2016). Fanconi-Bickel syndrome in two Palestinian children: Marked phenotypic variability with identical mutation. BMC research notes 9 (1), 1-5.
I am the first and the corresponding author of this case report which describes two Palestinian children from two unrelated families with identical mutation in GLUT2 gene but showed marked phenotypic variability further expanding the clinical spectrum of FBS.
20. K Ayeshe, I Dweikat, T Ciecieroga, M Sultan (2016). Capsule Endoscopy detects Meckel's Diverticulum in a Child with Recurrent Gastrointestinal Bleeding: Case Report and Review of the Literature. Journal of Gastric Disorders and Therapy 2 (2). *I am one of co-authors of this case report which describes a 7-year old boy with recurrent gastrointestinal bleeding and had normal upper and lower endoscopy. Identification of Meckel's diverticulum was achieved by wireless capsule endoscopy.*

21. **I Dweikat**, I Alawneh, M Ashab. Late-infantile Metachromatic Leukodystrophy in a Palestinian girl: Report of a novel mutation in ARSA gene (2016). Journal of Inherited Metabolic Diseases 39 (39 (Suppl 1):S35–S284 S271).
I am the first and the corresponding author of this report which describes a 30-month old Palestinian girl with typical phenotype of Metachromatic Leukodystrophy and has novel mutation in ARSA gene further expanding the genetic spectrum of the disorder.
22. **I Dweikat**, E Naser, N Damsa (2016). A distinct Phenotype of Mevalonic Acidemia with absence of pathogenic mutations of Mevalonate Kinase Gene. An-Najah university journal of research-A (Natural sciences) An - Najah Univ. J. Res. (N. Sc.) Vol. 30(1), 2016 .
I am the first and the corresponding author of this clinical report which describes two brothers with mevalonic aciduria and distinct phenotype with characteristic absence of pathogenic mutations in the mevalonate kinase gene.
23. **IM Dweikat**, S Abdelrazeq, S Ayesh, T Jundi (2014). MEGDEL syndrome in a Child from Palestine: Report of a Novel Mutation in SERAC1 Gene. Journal of Child Neurology 30 (8), 1053-1056
I am the first and corresponding author of this brief communication which identifies a novel mutation in a child with the syndrome of 3-methylglutaconic aciduria, encephalopathy, sensorineural deafness and Leigh-like lesions on brain MRI. It was the first report from Palestine.
24. T Ciecieręga, **I Dweikat**, M Awar, M Shahrour, BA Libdeh, M Sultan (2014). Severe persistent unremitting dermatitis, chronic diarrhea and hypoalbuminemia in a child; Hartnup disease in setting of Celiac disease. BMC pediatrics 14 (1), 311
I am one of the coauthors of this clinical report which describes a 3-year old girl with chronic diarrhea, hypoalbuminemia and exfoliative diarrhea with the diagnosis of celiac disease but did not improve with gluten-free diet. Hartnup disease was confirmed by neutral aminoaciduria and she improved on Niacin.
25. **IM Dweikat**, N Damsah, R Khalaf (2014). A distinct phenotype of childhood leukodystrophy presenting as absence seizure. Journal of pediatric neurosciences (1), 63. *I am the first and corresponding author of this case report which describes a 4-year old female patient with diffuse white matter disease on brain magnetic resonance imaging but had seizure as the sole manifestation of the disease.*
26. **I Dweikat**, E Naser, N Damsah, BA Libdeh, I Bakri (2012). Ethylmalonic encephalopathy associated with crescentic glomerulonephritis. Metabolic brain disease 27 (4), 613-616
I am the first and corresponding author of this clinical report which describes an 8-month old girl with a rare organic acidemia called Ethylmalonic acidemia, petechial rash and encephalopathy that was complicated by crescentic glomerulonephritis. This was a novel association and represent severe phenotype of the disease.

27. **IM Dweikat**, EN Naser, AIA Libdeh, OJ Naser, NNA Gharbieh, NF Maraqa. Propionic acidemia mimicking diabetic ketoacidosis (2011). *Brain and Development* 33 (5), 428-431.
I am the first and corresponding author of this clinical report which describes unusual presentation of Propionic acidemia in a 9-month old Palestinian boy resembling diabetic ketoacidosis.
28. **I Dweikat**, BA Libdeh, H Murrar, S Khalil, N Maraqa (2011). GM1 Gangliosidosis associated with neonatal-onset of diffuse ecchymoses and mongolian spots. *Indian journal of dermatology* 56 (1), 98
I am the first and corresponding author of this clinical report which describes a 7-month old girl with dermal melanocytosis or skin pigmentation as an early findings of GM1 Gangliosidosis and other lysosomal storage diseases .
29. C Viscomi, AB Burlina, **I Dweikat**, M Savoiaro, C Lamperti, T Hildebrandt (2010). Combined treatment with oral metronidazole and N-acetylcysteine is effective in ethylmalonic encephalopathy. *Nature medicine* 16 (8), 869-871.
I am one of the co-authors of this article which describes the clinical improvement of neurologic and gastrointestinal symptoms and signs in 4 children with a rare organic acidemia called Ethylmalonic aciduria (EPEMA) following the combined treatment with oral metronidazole and N-acetylcysteine.
Nature Medicine is a biomedical research journal devoted to publishing the latest and most exciting advances in biomedical research for scientists and physicians. The 2009 impact factor for this journal is 27.136
30. **I Dweikat**, M Sultan, N Maraqa, T Hindi, S Abu-Rmeileh, B Abu-Libdeh (2007). Tricho-Hepato-Enteric Syndrome: Report of a case of Intractable Diarrhea, Dysmorphic features, Hemochromatosis and Hair Anomaly. *American Journal of Medical Genetics Part A* 143 (6), 581-583.
I am the first and corresponding author of this clinical report which describes the rare syndromic iron storage disease.
31. **IM Dweikat**, I Naser, B Abu Libdeh, S Khalil, EA Naser (2007). Profound Biotinidase Deficiency Manifesting as Neonatal-onset Intractable Seizures and Diffuse Leukoencephalopathy. *Journal of Inherited Metabolic Disease* 30, 41-41
I am the first and corresponding author of this clinical report which describes severe white matter disease as early as first week of age caused by Biotinidase deficiency with dramatic improvement after treatment with Biotin. It was presented as a Poster in the annual symposium of the society of inborn errors of metabolism in Hamburg, 4-7 september, 2007.
32. SH Korman, C Jakobs, PS Darmin, A Gutman, MS van der Knaap, Ziva Ben-Neriah, **Imad Dweikat**, Isaiah D Wexler, Gajja S Salomons (2007). Glutaric aciduria type 1: clinical, biochemical and molecular findings in patients from Israel. *Eur J Paediatr Neurol*. 11(2):81-9. Epub 2006 Dec 26.

33. SH Korman, JJ Pitt, A Boneh, I Dweikat, M Zater, V Meiner, A Gutman (2006). A novel SLC25A20 splicing mutation in patients of different ethnic origin with neonatally lethal carnitine-acylcarnitine translocase (CACT) deficiency. *Mol Genet Metab.* 2006 Dec;89(4):332-8. Epub 2006 Aug 17.

LECTURES & PRESENTATIONS

1. Branched-chain organic acidemias; A case series of 53 palestinian patients . This study was presented by me at The 33d UMEMPS Congress and The 13th Congress of Jordan Pediatric Society in Amman, Jordan on May, 2011
2. Participation in the 33d UMEMPS Congress & The 13th Congress of Jordan Pediatric Society in Amman-Jordan on May 2011 as oral presentation with following title: Branched-Chain organic acidemias; Case series of 53 Palestinian patients
3. Participation in the 18th Conference of the Union of Arab Pediatrician in Beirut on October 2011 as oral presentation with following title: A stepwise clinical approach to inborn errors of metabolism
4. Glycogen storage disease type III; Report of 24 cases.
This work was presented by me at the Fifth Scientific Conference organized by Al-Quds university which was held at Jericho on April 30th-May 1st 2009. This work was also presented by me at the 17th Conference of the Union of Arab Pediatricians in Amman, Jordan on May2009
5. When to think of a metabolic disorder.
This lecture was presented by me at the second conference of the medical association in Bethlehem on May, 2009
6. Propionic & Methylmalonic Acidemia Diagnosed at Makassed Hospital 2000-2005.
This work was presented by me at the First Palestinian Symposium on Clinical Genetics & Metabolic Disorders in Children in Jericho on April 2006
7. Epidemiology of Bacterial meningitis in Palestine; Report of 80 cases in the pre H. Influenza vaccine era and 55 cases in the post vaccine era.
This work was presented by me at the Pan Arab Conference for Childhood Diseases in Beirut, Lebanon on November, 2000.
8. Chronic Granulomatous Disease (CGD); Report of 10 cases.
This work was presented by me at the Pan Arab Conference for Childhood Diseases in Beirut, Lebanon on November, 2000 and it is being prepared for submission for an international journal for publication.

CONFERENCES

1. Successful completion of the Emergency Medicine Education and Development Course held by the John Hopkins University Center for International Emergency, Disaster and Refugee Studies and CARE International in West Bank/Gaza from January 8, 2003 to January 14, 2003.
2. Successful completion of the Teaching Pediatrics and Child Health course held by The Royal College of Pediatrics and Child health in Amman, Jordan in November 2006
3. Successful completion of Pediatric Life Support Course held by The Royal College of Pediatrics and Child health in Amman, Jordan in November 2006
4. Attendance at the 42nd annual symposium of the Society for The Study of Inborn Errors of Metabolism (SSIEM) in Paris, France on September, 2006
5. Participation in the annual symposium of the SSIEM in Hamburg, Germany on September, 2007 with the following title:
Profound Biotinidase Deficiency Manifesting as Neonatal-onset Intractable Seizures and Diffuse Leukoencephalopathy
6. I was the General Coordinator of the 5TH Pediatric Conference which was held in Jericho by the Pediatric Society-Palestine on March, 2008
7. Attendance of the 3d Congress of European Association of Paediatric Societies (EAPS) in Copenhagen/Denmark on October 2010
8. Participation in the 18th Conference of the Union of Arab Pediatrician in Beirut on October 2011 as oral presentation with following title: *A stepwise clinical approach to inborn errors of metabolism*
9. Attendance of the annual symposium of the Society for The Study of Inborn Errors of Metabolism (SSIEM) in Bermingham/United Kingdom on September 2012
10. Attendance of the annual symposium of the Society for The Study of Inborn Errors of Metabolism (SSIEM) in Innsbruck/Austria on September 2014
11. Participation in the Child Emotional and Behavioural Difficulties workshop as part of Continuous Medical Education of the Pediatric Society/ Palestine in Ramallah/Palestine on August 2014
12. Participation in the 5th International Palestinian Conference of the Medical Association in Bethlehem/ Palestine on November 2015 as oral presentation with the following title: *Metabolic Disorders in Adults*
13. Participation in the annual symposium of the Society for The Study of Inborn Errors of Metabolism (SSIEM) with abstract in Rome/Italy on September 2016

14. Attendance of the annual symposium of the Society for The Study of Inborn Errors of Metabolism (SSIEM) in Athens/ Greece on September 2018

REFERENCES

1. Bassam Y. Abu-Libdeh, MD
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Genetics Makassed Hospital
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2. Hatem Khammash, MD
Head of NICU
Makassed Hospital
Jerusalem
E. mail: Khamash60@yahoo.com
3. John Callahan, PhD
Head of Metabolic Laboratory
The Hospital for Sick Children
Toronto, Canada