

CURRICULUM VITAE

Donald Basel MD

**Chief, Professor
Department of Pediatrics
Division of Genetics**

OFFICE ADDRESS:

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EDUCATION:

01/1987 - 11/1989 BSc, University of Witwatersrand, South Africa
01/1990 - 11/1994 MBBCh, University of Witwatersrand, South Africa
11/12/2006 Educational Commission for Foreign Medical Graduates

POSTGRADUATE TRAINING AND FELLOWSHIP APPOINTMENTS:

01/1995 - 12/1995 Internship, Tintswalo Hospital, Maphumulanga, South African
01/1996 - 06/1996 Senior House Officer, Accident & Emergency Medicine, Worthing Hospital, Worthing, West Sussex, UK
01/1998 - 06/1998 Senior House Officer, Pediatrics, Red Cross War Memorial Children's Hospital, Cape Town, South Africa
07/1998 - 12/2000 Registrar, Department of Human Genetics, University of Cape Town, South Africa
01/2001 - 07/2007 Research Fellow, Department of Human Genetics and Developmental Biology, (Managed Lab 2003-2007), University of Connecticut Health Center, Farmington, CT
07/2007 - 06/2009 Fellow, Medical Genetics, Oregon Health & Sciences University, Portland, OR
07/2009 - 06/2010 Resident for ABMG, Oregon Health & Sciences University, Portland, OR
07/2012 International Skeletal Dysplasia Workshop, (Selected as one of 10 candidates from 150 international applicants), Lusanne, Switzerland

FACULTY APPOINTMENTS:

07/2010 - 06/2015 Assistant Professor, Department of Pediatrics, Medical Genetics, Medical College of Wisconsin, Milwaukee, WI
07/2015 - Present Associate Professor, Department of Pediatrics, Genetics, Medical College of Wisconsin, Milwaukee, WI

ADMINISTRATIVE APPOINTMENTS:

07/2015 - 06/2016 Interim Section Chief, Department of Pediatrics, Genetics, Medical College of Wisconsin, Milwaukee, WI
07/2016 - Present Section Chief, Department of Pediatrics, Genetics, Medical College of Wisconsin, Milwaukee, WI
07/2017 - Present Associate Director, Human and Molecular Genetics Center, Rare and Undiagnosed Disease Program, Medical Director, Milwaukee, WI

EDUCATIONAL ADMINISTRATIVE APPOINTMENTS:

07/01/2010 - 08/31/2015 Associate Program Director, Pediatrics, Genetics, Medical College of Wisconsin, Medical Genetics Residency Program, Milwaukee, WI
2012 - 2015 Program Chair, Pediatrics, Clinical Competency Committee, Medical College of Wisconsin,

Medical Genetics Residency Program, Milwaukee, WI
 2012 - Present Program Chair, Pediatrics, Program Evaluation Committee, Medical College of Wisconsin, Medical Genetics Residency Program, Milwaukee, WI
 2014 - Present Member, Pediatrics, Program Evaluation Committee, Medical College of Wisconsin, Medical Genetics Residency Program, Milwaukee, WI
 2014 - Present Member, Pediatrics, Clinical Competency Committee, Medical College of Wisconsin, Medical Genetics Residency Program, Milwaukee, WI
 09/01/2015 - Present Program Director, Pediatrics, Genetics, Medical College of Wisconsin, Medical Genetics Residency Program, Milwaukee, WI 53226

HOSPITAL AND CLINICAL ADMINISTRATIVE APPOINTMENTS:

1991 - 1994 Student Rural Health Clinic, (administrative/funding and implementation of primary health care services at a rural health clinic)
 2010 - Present Co-Director, Children's Hospital of Wisconsin, Neurofibromatosis and RASopathy Center, Milwaukee, WI
 2010 - 2011 Member, (core group that redefined practice model), Medical College of Wisconsin, Fetal Concerns Steering Committee, Milwaukee, WI
 2012 - Present Clinical Practice Committee, Children's Hospital of Wisconsin, Genetics Center, (review current clinical practices in division and recommend changes in accordance with Best Practices), Milwaukee, WI
 2013 - Present Faculty Council, Children's Specialty Group, Medical College of Wisconsin, Board Member, Milwaukee, WI
 07/2016 - Present Medical Director, Genetics Center, Children's Hospital of Wisconsin, Milwaukee, WI

HOSPITAL STAFF PRIVILEGES:

1996 - 1997 Associate Physician, London Wall, London, UK, Dr. Dorothy Kelly & Associates (4/7 days/week)
 1996 - 1997 Primary Care Physician, Harley Street, London, UK, Medcall UK (1 day/week)
 09/2010 - Present Children's Hospital of Wisconsin, 9000 W. Wisconsin Avenue, Milwaukee, WI 53226
 12/13/2010 - Present Froedtert Memorial Lutheran Hospital, 9200 W. Wisconsin Ave, Milwaukee, WI 53226
 01/2012 - Present Children's Hospital of Wisconsin, Fox Valley, 130 Second St, Neenah, WI 54956
 04/2012 - 01/2013 Aurora Sinai Medical Center, Aurora West Allis Medical Center, Aurora Health Care, Milwaukee, WI

SPECIALTY BOARDS AND CERTIFICATION:

<u>Board Certified</u>	<u>Issue Date</u>	<u>Expiration</u>
Clinical Genetics	09/01/2011	12/31/2021

<u>Licensure</u>	<u>Number</u>	<u>Issue Date</u>	<u>Expiration</u>
Wisconsin License	54954	09/2010	10/2021

AWARDS AND HONORS:

2014 - 2015 Teaching Award: High 5 Winner, Medical College of Wisconsin, Department of Pediatrics, Milwaukee, WI
 2016 - 2017 Expert Teacher, Medical College of Wisconsin, Department of Pediatrics, Milwaukee, WI
 2016 - 2019 Expert Clinician, Medical College of Wisconsin, Department of Pediatrics, Milwaukee, WI
 2017 Best Doctors in America
 2018 BEST Teacher: Pediatrics Residency, Medical College of Wisconsin, Department of Pediatrics, Milwaukee, WI
 2019 Expert Teacher, Medical College of Wisconsin, Department of Pediatrics
 2019 Best Doctors in America

MEMBERSHIPS IN HONORARY AND PROFESSIONAL SOCIETIES:

1999 - 2000 Western Cape Genetics Ethics Committee (Member)

1999 - 2000 Cape Regional Spina Bifida Society (Medical Advisor)
07/2007 - Present American College of Medical Genetics (Member)
07/2009 - Present American Pediatric Association (Member)
2011 - Present Association of Professors of Human and Medical Genetics (Member)
07/2012 - Present International Skeletal Dysplasia Society (Member)

EDITORSHIPS/EDITORIAL BOARDS/JOURNAL REVIEWS:

Journal Review

2003 - 2007 Human Molecular Genetics
2004 - 2007 American Journal of Medical Genetics
2009 - Present European Journal of Medical Genetics
2009 - Present Genetics in Medicine
2011 - Present GeneReviews
2012 - Present Cochrane Reviews
2012 - Present Journal of the American College of Nutrition

Ad-Hoc Reviewer

2016 Grant Review, Advancing a Healthier Wisconsin
2017 Grant Review, Clinical and Translational Science Institute (CTSI)

LOCAL/REGIONAL APPOINTED LEADERSHIP AND COMMITTEE POSITIONS:

2013 - Present Member, Wisconsin Heterotaxy Interest Group, Medical College of Wisconsin
07/01/2014 - 06/30/2017 Representative, Faculty Council, Medical College of Wisconsin
04/01/2015 - Present Member, Genetics Advisory Committee, State of Wisconsin
05/2016 - 07/2017 Member, HMGC Search Committee, Medical College of Wisconsin

NATIONAL ELECTED/APPOINTED LEADERSHIP AND COMMITTEE POSITIONS:

06/2016 - Present Council member, Association of Professors of Human and Medical Genetics

RESEARCH GRANTS/AWARDS/CONTRACTS/PROJECTS:

Active

Non-Peer Review

Title:	PKUDOS-PKU Demographics, Outcomes and Safety Registry
Source:	BioMarin Pharmaceuticals
Role:	Principal Investigator
PI:	Donald Basel, MD
Dates:	09/2008 - 09/2023
Title:	Long-term Extension of a Phase 2, Open-Label Dose-Finding Study to Evaluate the Safety, Efficacy and Tolerability of Multiple Subcutaneous Doses of rAvPAL-PEG in Subjects with PKU, Pal-003
Source:	BioMarin Pharmaceutical
Role:	Sub-Investigator
PI:	William J. Rhead, MD, PhD
Dates:	01/2010 - 01/2019
Title:	A Four Part, Phase 3, Randomized, Double-Blind, Placebo-Controlled, Four-Arm Discontinuation Study to Evaluate the Efficacy and Safety of Subcutaneous Injections of BMN 165 Self-Administered by Adults with Phenylketonuria, PRISM 302

Source:	BioMarin Pharmaceuticals
Role:	Sub-Investigator
PI:	William J. Rhead, MD, PhD
Dates:	06/2013 - 12/2018
Title:	A Phase 3B Open-Label study to Evaluate the Effect of Kuvan on Neurocognitive Function, Maintenance of blood Phenylalanine concentrations, safety and population pharmacokinetics in Young Children with Phenylketonuria, PKU-015
Source:	BioMarin Pharmaceuticals
Role:	Sub-Investigator
PI:	William J. Rhead, MD, PhD
Dates:	12/2014 - 12/2017
Title:	A Multicenter, Multinational Clinical Assessment Study for Pediatric Patients with Achondroplasia
Source:	BioMarin Pharmaceuticals
Role:	Principal Investigator
PI:	Donald Basel, MD
Dates:	04/2016 - 04/2024
Title:	A Phase 2, Multicenter, Multinational, Open-Label, Dose-Escalation Study to Evaluate the Safety and Efficacy of ALXN1101 in Pediatric Patients with Molybdenum Co-Factor Deficiency (MOCD) Type A Currently Treated with rcPMP, ALXN201
Source:	Alexion Pharmaceuticals
Role:	Principal Investigator
PI:	Donald Basel, MD
Dates:	06/2016 - 06/2020
Title:	A Phase 2/3, Multicenter, Multinational Open-Label Study to Evaluate the Efficacy and Safety of ALXN1101 in Neonates with Molybdenum CoFactor Deficiency (MoCD) Type A, ALXN 202
Source:	Alexion Pharmaceuticals
Role:	Principal Investigator
PI:	Donald Basel, MD
Dates:	06/2016 - 06/2020
Title:	Morquio, A Registry Study
Source:	BioMarin Pharmaceuticals
Role:	Principal Investigator
PI:	Donald Basel, MD
Dates:	06/2016 - 09/2024
Title:	Active, non-industry: DNA testing for Identifying Underdiagnosed Genetics Disorders
Role:	Principal Investigator

PI: Donald Basel, MD
Dates: 06/2016 - 06/2018

Prior

Peer Review

Title: Investigation into the role of FBN in Spontaneous Pneumothorax
Role: Co-Investigator
Title: Use of Multimetric hammerhead Ribozymes in a Murine Model of OI
Title: Use of Hammerhead Ribozymes in a Cellular Model of OI
Role: Co-Investigator
Title: Investigation into the Molecular Mechanisms of Split Hand Foot Malformation Syndrome
Source: Co-Investigator

Non-Peer Review

Title: Skeletal Dysplasia Database
Source: Medical College of Wisconsin
Role: Principal Investigator
PI: Donald Basel, MD
Dates: 07/01/2010 - 06/30/2013
Direct Funds: \$20,000

Title: Study to Compare the Efficacy and Safety of Oral AT1001 and Enzyme Replacement Therapy in Patients with Fabry Disease
Source: Amicus Therapeutics
Role: Co-Investigator
PI: David Dimmock, MD
Dates: 12/2010 - 09/2015

Title: Open, Prospective, History-controlled, Multicenter Study to Evaluate the Safety and Efficacy of Infusion of Liver Cell Suspension (HHLivC) in Children with Urea Cycle Disorders
Source: Cytonet GmbH & Co
Role: Co-Investigator
PI: David Dimmock, MD
Dates: 12/2010 - 05/2015

Title: A Phase 2, Open-Label Study to Evaluate the Safety, Tolerability and Efficacy of 4 Subcutaneous Dose Levels of rAvPAL-PEG Administered Daily in Subjects with Phenylketonuria, Pal-004
Source: Biomarin Pharmaceuticals
Role: Co-Investigator
PI: David Dimmock, MD

Dates:	03/2011 - 03/2014
Title:	A Phase-4, Open-Label, Prospective Study in Patients with Pompe Disease to Evaluate the Efficacy and Safety of Alglucosidase Alfa Produced at the 4000 L Scale
Source:	Genzyme
Role:	Co-Investigator
PI:	David Dimmock, MD
Dates:	03/2012 - 03/2015
Title:	A Retrospective, Observational, Non-interventional Data Collection Study for Patients with Molybdenum Cofactor Deficiency Who Have Been Previously Treated with Cyclic Pyranopterin Monophosphate (cPMP) FP 5234
Source:	PPD Development
Role:	Co-Investigator
PI:	Gunter Scharer, MD
Dates:	05/2013 - 06/2015
Title:	A Cross-Sectional Study of Renal Function in Treatment-Naive, Young Male Patients with Fabry Disease
Source:	Genzyme
Role:	Co-Investigator
PI:	David Dimmock, MD
Dates:	06/2013 - 02/2016
Title:	A Phase 3, Open-Label, Randomized, Multicenter Study to Assess the Safety and Tolerability of an Induction, Titration and Maintenance Dose Regimen with BMN 165
Source:	Biomarin Pharmaceuticals
Role:	Co-Investigator
PI:	David Dimmock, MD
Dates:	07/2013 - 06/2016
Title:	Reference Values for Organic Acids from Term Newborns
Role:	Co-Investigator
PI:	David Dimmock, MD
Title:	Advancing Genomic Sequence to a Clinical Laboratory Assay
Role:	Co-Investigator
PI:	David Dimmock, MD

INVITED LECTURES/WORKSHOPS/PRESENTATIONS:

Local

Review of lethal congenital skeletal dysplasias, Fetal Concerns meeting, Froedtert Hospital, Milwaukee, WI, 07/10/2010

The value of Computerized Tomography in prenatal diagnosis of skeletal dysplasia, Fetal Concerns Meeting,

Froedtert Hospital, Milwaukee, WI, 07/11/2012
 NF1 and plexiform neurofibromatosis, Tumor Board, Children's Hospital of Wisconsin, Milwaukee, WI, 07/19/2012
 Case discussion on campomelic dysplasia and fetal imaging, Fetal Concerns Meeting, Froedtert Hospital, Milwaukee, WI, 08/29/2012
 Mechanisms of dysmorphogenesis, Pediatric Noon Conference, Children's Hospital of Wisconsin, Milwaukee, WI, 09/24/2012
 Mechanisms of dysmorphogenesis, Pediatric Noon Conference, Children's Hospital of Wisconsin, Milwaukee, WI, 09/24/2012
 Dwarfs and other celestial dysplasias, Genetics Exchange, Children's Hospital of Wisconsin, Milwaukee, WI, 10/05/2012
 To scan or not to scan, Children's Tumor Foundation, Children's Hospital of Wisconsin, Milwaukee, WI, 11/03/2012
 Notch Signaling and Biological Clocks, Fetal Concerns Lecture Series, Froedtert Hospital, Milwaukee, WI, 02/06/2013
 A brief look at free-cell DNA technology, The Fetal Concerns Center of WI Weekly Case Conference, Children's Hospital of Wisconsin, Milwaukee, WI, 06/05/2013
 Genetics and Low Grade Gliomas, Tumor Board, Children's Hospital of Wisconsin, Milwaukee, WI, 07/11/2013
 Geddes, G.C., BASEL, D.G., Yield of genetic testing in congenital heart disease requiring surgical intervention in the first year of life, Resident Day, Medical College of Wisconsin, Milwaukee, WI, 09/18/2013
 Hypermobility and Connective Tissue Disorders, Children's Hospital of Wisconsin, Physical and Occupational Therapy Department, Milwaukee, WI, 01/17/2014
 Many Faces of EDS, Rheumatology Conference, Medical College of Wisconsin, Milwaukee, WI, 05/22/2014
 Syndromes and Malformations, Noon Conference, Medical College of Wisconsin, Milwaukee, WI, 03/30/2015
 Genetics to Put You to Sleep, Pediatric Anesthesiology, Medical College of Wisconsin, 05/11/2015
 Dysmorphology-Clues from the Skin, Dermatology Fellows, Medical College of Wisconsin, Milwaukee, WI, 01/22/2016
 Genetics and Genomics: Clinical Relevance in APN Practice & Scope for Career Development, Advanced Practice Providers Retreat, Waukesha, WI, 10/05/2016
 Making Sense of Skeletal Dysplasia, Fellow Lecture, Medical College of Wisconsin, Milwaukee, WI, 01/25/2017
 Syndromes and Malformations, Pediatric Noon Conference, Medical College of Wisconsin, 02/24/2017
 When to Refer to Genetics, Autonomic Workshop, Children's Hospital of Wisconsin, 06/02/2017
 Syndromes and Microdeletions, Maternal Fetal Medicine Noon Conference, Medical College of Wisconsin, 08/16/2017

Regional

The future of genetic testing in neurofibromatosis & clinical trials: the benefit for patients with NF1, NF1 Regional Family Symposium, 03/12/2011
 Review of genes and mechanisms in Stickler Syndrome, Annual Stickler Support Meeting, 07/09/2011
 Hypoglycemia and Cyclic Vomiting, 2011 State of Wisconsin Practical Genetics Conference for Health Care Providers, Marshfield, WI, 10/2011
 To EDS & Beyond the Matrix: An approach to hereditary disorders of the connective tissue, St. Agnes Hospital, Department of Pediatrics and Medical Staff CME meeting, Fond du lac, WI, 05/16/2013
 Connective Tissue Disorders, St. Agnes Hospital Medical Staff Conference, Fond du Lac, WI, 05/16/2013
 Many Faces of EDS, Genetics Exchange, Madison, WI, 06/06/2014
 The Many Changing Faces of EDS, Genetics Exchange, Madison, WI, 06/06/2014
 The connective tissue paradigm in NF1, Littlest Tumor Foundation, 2014
 Extra musculoskeletal disease in connective tissue disorders: Why do structural protein anomalies cause fibromyalgia,, Regional Genetic Conference, Madison, WI, 2014
 Evolving Genetic Service Delivery Models, Wisconsin Genetics Exchange 2016, Marshfield, WI, 09/09/2016
 Restructing of the Genetic Clinic Workflow, Oregon Health & Science University, Portland, OR, 12/02/2016
 Integrating Genomic Medicine into Clinical Practice, Best Practices, Children's Hospital of Wisconsin, Wisconsin Dells, WI, 03/11/2017

National

Dietz, De Paepe, et al, Consortium for Disorders of Connective Tissue, NIH Workshop, Bethesda, MD, 2005
BioMarin Best Practices in the Slowly Progressing MPS VI Patient, Biomarin Advisory Board, Washington, DC, 05/11/2012

Alexion Hypophosphatasia Advisory Board Meeting, Denver, Co, 05/30/2014

The Nelson Service - Lessons Learned from Undiagnosed and Rare Disease Program, 6th Annual Mayo Clinic Individualizing Medicine Conference, Rochester, MN, 10/09/2017 - 10/10/2017

International

Dysmorphology Examination, MCW/China Medical Association, Intensive Developmental and Behavioral Pediatrics Training for Chinese Pediatricians, Medical College of Wisconsin, Milwaukee, WI, 09/10/2012

An approach to Dysmorphology for Developmental Pediatricians, Intensive Developmental and Behavioral Pediatrics in Training for Chinese Pediatricians, Medical College of Wisconsin, Milwaukee, WI, 09/10/2012

MEDICAL COLLEGE TEACHING ACTIVITIES:

Medical Student Education

2010 Team Based Learning Pilot Group

2010 MS1 Facilitator in medical genetics component of TBL

2011 - Present Evaluator: Junior Medical Student Genetics Rotation

2012 Evaluator: M2 Physical Exam Workshop

01/24/2014 M3 Intercession on Professionalism

01/2014 - 12/2014 M1 Clinical Apprenticeship, Brianna McSorley (40-50 hours)

07/01/2014 - Present Course Director, CEC Discovery M3-M4 Elective

2014 - Present M2 Neuropathology - Malformations and Developmental diseases (10 hours)

2014 M2 Elective Fair

03/09/2015 CNS Malformations Lecture

03/07/2016 CNS Malformations Lecture

10/27/2016 MCAD Small Discussion Group

03/02/2017 Malformations and Developmental Disorders

03/08/2017 Toxic and Metabolic Disorders of the CNS

Resident and Fellow Education

2010 - 2012 Noon Conference (varied subject content - 5 didactic sessions)

2011 Surf the Boards

09/29/2012 Ace the Boards

11/14/2012 Basic Science Lecture, Genetic Basis of Disorders with Orthopaedic Manifestations

09/30/2013 Ace the Boards

06/29/2016 Common Skeletal Malformations, Maternal Fetal Care Fellow Lecture

10/26/2016 Genetic Disorders with Orthopaedic Manifestations

11/03/2017 Moderate Lecture on Neurocutaneous Disorders

11/28/2017 Genetic Testing - Types of Tests

Graduate Student Education

11/01/2012 Clinical & Translational Science Institute, Developmental Genetics/Connective Tissue Disorders

EXTRAMURAL TEACHING:

Medical Student Education

1998 - 2000 University of Cape Town, South Africa, MS1-3 Medical genetics course

2001 - 2007 University of Connecticut (UConn), MS 1/2 and Dental Students: Small group teaching Genetics Course

2007 - 2010 Oregon Health and Sciences University, Portland, Oregon, MS1-2 Lecture and small group

facilitator
2007 - 2010 Oregon Health and Sciences University, Portland, OR, Small group facilitator
2007 - 2010 Oregon Health and Sciences University, Portland, OR, MS1-2 Lecture

Resident and Fellow Education

2007 - 2010 Oregon Health and Sciences University, Portland, Oregon, Noon seminar series on Genetics
2007 - 2010 OHSU and Shriner's, Portland, OR, Neurology, OB/GYN, Orthopedics

Continuing Medical Education

1998 - 2000 Southern Cape District, South Africa, Key organizer and educator for several 5 day conferences to educate Genetic nurse practitioners in basic clinical genetic skills and genetic counseling
2007 - 2010 Oregon Health and Sciences University, Grand Rounds, Pediatrics, Molecular and Medical Genetics

MCW STUDENTS, FACULTY, RESIDENTS AND CLINICAL/RESEARCH FELLOWS MENTORED:

Medical Students

Bryce Schuler, Medical College of Wisconsin, 07/2015 - Present Mentor

PROGRAMMATIC DEVELOPMENTS:

Clinical Programs

1998 - 2000 Western Cape District Medical Genetics Rural Health Program, Institutional & Outreach Genetics Screening Program (Key role in expanding existing services, helped to establish presymptomatic screening)
2000 - 2005 Dysmorphology Listserv (Developed in conjunction with Dr. Han Brunner)
2008 - 2009 Fetal Evaluation Program (Oregon Health and Sciences University - OHSU), Adult Connective Tissue Disorders clinic (Developed along with Dr. J. Sampson)
2010 - Present Co-Director, Neurofibromatosis and RASopathy Center
2010 - Present Hereditary Disorders of Connective Tissue Clinic
2010 - Present Fetal Concerns Program, Genetics consultant
2011 - Present Familial Cancer Service for children
2013 - Present Diagnostic Odyssey/Nelson Services - Rare Disease Consortium (core founding member with Drs. Kliegman, Bordini and Nocton)

COMMUNITY SERVICE ACTIVITIES:

1991 - 1994 Rural Health Services Development, University of Witwatersrand, South Africa
1995 Several health education initiatives for Nurse Practitioners in Gazankulu, South Africa

BIBLIOGRAPHY

Refereed Journal Publications/Original Papers

1. BASEL D., Sobey, G., Gardner, J., Beighton, P. The Gordon syndrome revisited. S Afr Med J. 2000 Sep;90(9):864-7.
2. BASEL, D., Goldblatt, J. Tibial aplasia--VACTERL association, a new syndrome? Clin Dysmorphol. 2000 Jul;9(3):205-8.
3. Ianakiev, P., Kilpatrick, M.W., Toudjarska, I., BASEL, D., Beighton, P., Tsipouras, P. Split-hand/split-foot malformation is caused by mutations in the p63 gene on 3q27. Am J Hum Genet. 2000 Jul;67(1):59-66 PMID: PMC1287102.
4. BASEL, D., Beighton, P., Kozlowski, K. Help! Unusual X-ray appearances of a congenital bone disease of unknown aetiology. Pediatr Radiol. 2001 Mar;31(3):212.

5. Sifakis, S., BASEL, D., Ianakiev, P., Kilpatrick, M., Tsipouras, P. Distal limb malformations: underlying mechanisms and clinical associations. *Clin Genet.* 2001 Sep;60(3):165-72.
6. Stephen, L.X., BASEL, D., Beighton, P.H. Developmental absence of the premolar teeth: dental management. *Int J Paediatr Dent.* 2002 May;12(3):219-22.
7. BASEL, D., DePaape, A., Kilpatrick, M.W., Tsipouras, P. Split hand foot malformation is associated with a reduced level of Dactylin gene expression. *Clin Genet.* 2003 Oct;64(4):350-4.
8. Kozlowski, K., BASEL, D., Beighton, P. Chondrodysplasia punctata in siblings and maternal lupus erythematosus. *Clin Genet.* 2004 Dec;66(6):545-9.
9. BASEL, D., Kilpatrick, M.W., Tsipouras, P. Haplotype analysis enables the diagnosis of Marfan syndrome. *Conn Med.* 2004 Jun-Jul;68(6):363-6.
10. BASEL, D., Sklar, D., Viljoen, D. A newly recognized syndrome of cutis aplasia, lipomatous footpads, microcephaly, hypotelorism, and, variably, single maxillary central incisor, and holoprosencephaly. *Am J Med Genet A.* 2005 Aug 1;136A(4):354-6.
11. Kozlowski, K., BASEL, D., Beighton, P. Retrospective diagnosis of chondrodysplasia punctata. *Australas Radiol.* 2006 Feb;50(1):55-8.
12. Cordero, D.R., Goldberg, Y., BASEL, D., Kilpatrick, M.W., Klugman, S., Tsipouras, P., Gross, S. Prenatal sonographic diagnosis of Grebe syndrome. *J Ultrasound Med.* 2006 Jan;25(1):115-8; quiz 119-21.
13. BASEL, D., Kilpatrick, M.W., Tsipouras, P. The expanding panorama of split hand foot malformation. *Am J Med Genet A.* 2006 Jul 1;140(13):1359-65.
14. BASEL, D., Steiner, R.D. Osteogenesis imperfecta: recent findings shed new light on this once well-understood condition. *Genet Med.* 2009 Jun;11(6):375-85.
15. Chiu, Y.E., Dugan, S., BASEL, D., Siegel, D.H. Association of Piebaldism, multiple café-au-lait macules, and intertriginous freckling: clinical evidence of a common pathway between KIT and sprouty-related, ena/vasodilator-stimulated phosphoprotein homology-1 domain containing protein 1 (SPRED1). *Pediatr Dermatol.* 2013 May-Jun;30(3):379-82 PMID: PMC3967413.
16. Kwon, E.K., BASEL, D., Siegel, D., Martin, K.L. A review of next-generation genetic testing for the dermatologist. *Pediatr Dermatol.* 2013 Jul-Aug;30(4):401-8.
17. Klein-Tasman, B.P., Colon, A.M., Brei, N., van der Fluit, F., Casnar, C.L., Janke, K.M., BASEL, D., Siegel, D.H., Walker, J.A. Adaptive behavior in young children with neurofibromatosis type 1. *Int J Pediatr.* 2013;2013:690432 PMID: PMC3852810.
18. Fitzgerald, J., Holden, P., Wright, H., Wilmot, B., Hata, A., Steiner, R.D., BASEL, D. Phenotypic variability in individuals with Type V Osteogenesis Imperfecta with identical IFITM5 mutations. *Journal of Rare Disorders* 1:2, 37, 2013.
19. Fitzgerald, J., Holden, P., Wright, H., Wilmot, B., Hata, A., Steiner, R.D., Basel, D. Phenotypic variability in individuals with type V osteogenesis imperfecta with identical IFITM5 mutations. *J Rare Disord.* 2013 Dec;1(2):37-42. PMID: PMC5560441
20. Bellamkonda-Athmaram, V., Sulman, C.G., BASEL, D.G., Southern, J., Konduri, G.G., Basir, M.A. Alveolar capillary dysplasia with multiple congenital anomalies and bronchoscopic airway abnormalities. *J Perinatol.* 2014 Apr;34(4):326-8.
21. Brickler, M.M., BASEL, D.G., Gheorghe, G., Margolis, D.M., Kelly, M.E., Ehrhardt, M.J. Early therapy-related myeloid sarcoma and deletion of 9q22.32 to q31.1. *Pediatr Blood Cancer.* 2014 Sep;61(9):1701-3.
22. Aldinger, K.A., Mosca, S.J., Tetreault, M., Dempsey, J.C., Ishak, G.E., Hartley, T., Phelps, I.G., Lamont, R.E., O'Day, D.R., BASEL, D., Gripp, K.W., Baker, L., Stephan, M.J., Bernier, F.P., Boycott, K.M., Majewski, J., Parboosingh, J.S., Innes, A.M., Doherty, D. Mutations in LAMA1 cause cerebellar dysplasia and cysts with and without retinal dystrophy. *Am J Hum Genet.* 2014 Aug 7;95(2):227-34 PMID: PMC4129402.
23. Dwan, K., Phillipi, C.A., Steiner, R.D., BASEL, D. Bisphosphonate therapy for osteogenesis imperfecta. *Cochrane Database Syst Rev.* 2014(7):CD005088.
24. Alamillo, C.L., Powis, Z., Farwell, K., Shahmirzadi, L., Weltmer, E.C., Turocy, J., Lowe, T., Kobelka, C., Chen, E., BASEL, D., Ashkinadze, E., D'Augelli, L., Chao, E., Tang, S. Exome sequencing positively identified relevant alterations in more than half of cases with an indication of prenatal ultrasound anomalies. *Prenat Diagn.* 2015 Jul 5. doi: 10.1002/pd.4648. [Epub ahead of print] PMID: 26147564.
25. Acharya, K. BASEL, D., Segall, H., Sampath, V. Term Newborn with Unilateral Craniofacial Defects. DOI: 10.1542/neo.17-3-e184, *NeoReviews* 2016;17:e184.
26. Horan, F.T., Hall, D.N., Mannion, S., BASEL, D. Tributes. *S Afr Med J* 2016;106(6):10986 PMID:

27. Loomba, R.S., Geddes, G.C., BASEL, D., Benson, D.W., Leuthner, S.R., Hehir, D.A., Ghanayem, N., Shillingford, A.J. Bacteremia in Patients with Heterotaxy: A Review and Implications for Management. *Congenit Heart Dis.* 2016 Jul 18.
28. McCoy, G., Joyce, J., BASEL, D., Siegel, D.H. Pseudoarthrosis of the Ulna in Neurofibromatosis Type I. *J Pediatr.* 2016 Jul 22, PMID: 2745337
29. Chelimsky, G., Kovacic, K., Simpson, P., Nugent, M., BASEL, D., Banda, J., Chelimsky, T. Benign Joint Hypermobility Minimally Impacts Autonomic Abnormalities in Pediatric Subjects with Chronic Functional Pain Disorders. *J Pediatr.* 2016 Oct;177:49-52, PMID:27496265
30. Dwan, K., Phillipi, C.A., Steiner, R.D., BASEL, D. Bisphosphonate therapy for osteogenesis imperfecta. *Cochrane Database Syst Rev.* 2016 Oct 19;10:CD005088.
31. Kliegman, R.M., Bordini, B.J., BASEL, D., Nocton, J.J. How Doctors Think: Common Diagnostic Errors in Clinical Judgment-Lessons from an Undiagnosed and Rare Disease Program. *Pediatr Clin North Am.* 2017 Feb;64(1):1-15. PMID 27894438.
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