

## CURRICULUM VITAE

### Donald Basel MD

Chief, Professor  
Department of Pediatrics  
Division of Pediatrics 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: Principal Investigator  
PI: Donald Basel, MD  
Dates: 06/2016 - 06/2020

Title: Morquio, A Registry Study  
Source: BioMarin Pharmaceuticals  
Role & Effort: Principal Investigator  
PI: Donald Basel, MD  
Dates: 06/2016 - 09/2024

Title: Active, non-industry: DNA testing for Identifying Underdiagnosed Genetics Disorders  
Role & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: 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 & Effort: Co-Investigator  
 PI: David Dimmock, MD  
 Dates: 07/2013 - 06/2016

Title: Reference Values for Organic Acids from Term Newborns

Role & Effort: Co-Investigator  
 PI: David Dimmock, MD

Title: Advancing Genomic Sequence to a Clinical Laboratory Assay

Role & Effort: Co-Investigator  
 PI: David Dimmock, MD

**INVITED LECTURES/WORKSHOPS/PRESENTATIONS:**

**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

### **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 Hypohosphatasia 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

### **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

Restructuring 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

### **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

#### **MCW 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 Intersession 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

##### **Graduate Student Education**

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

##### **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

#### **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**

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

### **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., Tsiouras, 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., DePaepe, A., Kilpatrick, M.W., Tsiouras, P. Split hand foot malformation is associated with a reduced level of Dactylin gene expression. *Clin Genet.* 2003 Oct;64(4):350-4.
8. Kozłowski, 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., Tsiouras, 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. Kozłowski, 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., Tsiouras, 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., Tsiouras, 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 Fluït, 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., Gheorghie, 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., Tétreault, 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:

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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.
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