PROFESSOR PETER BEIGHTON
Abridged Curriculum Vitae

Date of Birth:	28th June 1934, Bolton, England
Nationality:	British
Medical Qualification, 1957:	St Mary's Hospital Medical School, University of London
1957 - 1960	Internships, London
1960 - 1962	National Service, Captain R.A.M.C., Parachute Regt, UK, United Nations, Congo
1962 - 1963	London School of Hygiene & Tropical Medicine
1963 - 1965	WHO International Medical Expedition to Easter Island, South Pacific
1965 - 1968	Residency in Internal Medicine, St. Thomas's Hospital, London
1968 - 1969	Research Fellow in Medical Genetics, Johns Hopkins Hospital, Baltimore, USA
1970 - 1972	Senior Research Associate, Dept of Orthopaedic Surgery, Univ. of the Witwatersrand
1982 - 1998	Director: MRC Unit for Medical Genetics, University of Cape Town
1972 - 1999	Professor of Human Genetics, University of Cape Town
2000 -	Emeritus Professor of Human Genetics, University of Cape Town
2000 -	Senior Research Associate, Faculty of Dentistry, University of the Western Cape
2007 -	Professor Extraordinarius, Faculty of Dentistry, University of the Western Cape
2011-	Senior Scholar, University of Cape Town

Professional Qualifications:

MB.BS; MRCS.LRCP	1957	PhD (Wits)	1974
DRCOG	1959	FRCP (Ed)	1975
DCH	1962	FRCP (London)	1978
DTM & H	1963	FRCP (Rheumatology) (UK)	1998
MRCP (Ed)	1964	M.Phil (Lancaster)	1999
MRCP (London)	1966	(Social History)	
MD (London)	1968		

Academic Honours and Awards:

1967 Fulbright Research Fellowship, University of Maryland, USA.
1975 British Orthopaedic Association Robert Jones Gold Medal
1977 Smith and Nephew Literary Award (jointly)
1978 S A Orthopaedic Association President's Medallion
1979 Smith and Nephew Literary Award
1979 Oppenheimer Fellowship, University of Cambridge
1986 Smith and Nephew Literary Award (jointly)
1994 Fellowship of the University of Cape Town
1997 S A Medical Research Council Silver Medal
1999 Ehlers-Danlos Foundation, USA.; International research award.
2002 Order of Mapungubwe, bronze; awarded for outstanding achievements in medical genetics.

Membership of Learned Societies:

Royal Society of South Africa (Fellow; past Vice-President)
Royal Geographic Society (Fellow)
South African Human Genetics Society (past Chairman)
International Skeletal Society (Founder Member)
Clinical Genetics Society, UK
European Human Genetics Society

Doctoral Students

Fourteen of Professor Beighton's postgraduate students have been awarded Doctorates; nine of these persons have gained Full or Associate Professorial status.

Publications:

Books : 20 Chapters : 34 Medical articles : 400+

BOOKS AND MONOGRAPHS

20. Hypermobility of Joints (4th ed)
P Beighton, R Grahame and H Bird
Springer-Verlag, Heidelberg, 2012.

19. Gamut Index of Skeletal Dysplasias (3rd Ed)
K. Kozlowski and P Beighton
Springer, Heidelberg, 2001.

18. Hypermobility of Joints (3rd ed)
P Beighton, R Grahame and H Bird
Springer-Verlag, Heidelberg, 1999.

17. Blackpool Division, St John Ambulance Brigade; The Early Years
P Beighton
Creda, Cape Town, 1998.

16. Genetic Counseling in the Dawn of the 21st Century
CS Bartsocas and P Beighton (Editors)
HTA Medical Publications, Athens, 1998.

15. The Person Behind the Syndrome
P Beighton and G Beighton
Springer-Verlag, Heidelberg, 1997.

14. Gamut Index of Skeletal Dysplasias (2nd ed)
K. Kozlowski and P Beighton
Springer, Heidelberg, 1995.

13. Dysmorphology and Genetics of Cardiovascular Disorders
Bartsocas CS and Beighton P (Editors)
HTA Medical Publications, Athens, 1994.

12. McKusick's Heritable Disorders of Connective Tissue (5th ed)
P Beighton (Editor)
C.V. Mosby, St. Louis, 1993.

11. Hypermobility of Joints (2nd ed)
P Beighton, R Grahame and H Bird
Springer-Verlag, Heidelberg, 1989.

10. Inherited Disorders of the Skeleton (2nd ed)
P Beighton
Churchill Livingstone, Edinburgh, 1988.

9. The Man Behind the Syndrome
P Beighton and G Beighton
Springer-Verlag, Heidelberg, 1986.

8. Gamut Index of Skeletal Dysplasias
K Kozlowski and P Beighton
Springer-Verlag, Heidelberg, 1984.

7. **Hypermobility of Joints**
P Beighton, R Grahame and H Bird
Springer-Verlag, Heidelberg, 1983.

6. **Genetics and Otology**
P Beighton and S Sellars
Churchill Livingstone, Edinburgh, 1982.

5. **Orthopaedic Problems in Inherited Skeletal Disorders**
F Horan and P Beighton
Springer-Verlag, Heidelberg, 1982.

4. **Sclerosing Bone Dysplasias**
P Beighton and B Cremin
Springer-Verlag, Heidelberg, 1980.

3. **Bone Dysplasias of Infancy**
B Cremin and P Beighton
Springer-Verlag, Heidelberg, 1978.

2. **Inherited Disorders of the Skeleton**
P Beighton
Churchill Livingstone, Edinburgh, 1978.

1. **The Ehlers-Danlos Syndrome**
P Beighton
William Heinemann, London, 1970.

CHAPTERS

34. **Giants in Mythology and Medicine.**
In: Festschrift, Professor C Bartsocas, Athens
ZHTA Medical Publications, Athens, Greece 2012

33. **Heritable Disorders of Connective Tissue in South Africa.**
In: Genomics and Health in the Developing World,
Ed. Dhavendra Kumar
Oxford University Press 2012, 482-486

32 **Heritable Disorders of the Skeleton in South Africa.**
In: Genomics and Health in the Developing World,
Ed. Dhavendra Kumar
Oxford University Press 2012, 475-481

31. **Hereditary Deafness in Southern Africa.**
P Beighton and S Sellars
In: Genomics and Health in the Developing World,
Ed. Dhavendra Kumar
Oxford University Press 2012, 466-474

30. **Genetic Disorders in Minority Groups in South Africa.**
In: Genomics and Health in the Developing World,
Ed. Dhavendra Kumar
Oxford University Press 2012, pp450-465

29. **Genetic Epidemiology of South Atlantic Islands.**
In: Genomics and Health in the Developing World,
Ed. Dhavendra Kumar
Oxford University Press 2012, pp444-449

28. **Genetic Deafness and Blindness.**
In: Fifty Years of Human Genetics, Festschrift and Liber Amicorum, Dr George R Fraser.
Eds. Oliver Mayo, Carolyn Leach
Wakefield Press, Australia, 2007, pp133-141

27. **Hereditary Noninflammatory Arthropathies**
In: Principles and Practice of Medical Genetics, Volume II, Fifth Edition, eds. David L Rimoin, J. Michael Connor, Reed E Pyeritz, Bruce R Korf
Churchill Livingstone, Elsevier, Philadelphia 2007, pp3865-3871

26. **The Natural History of Osteogenesis Imperfecta**
P Beighton and L Stephen
In: OI – Concensus Conference, ed. K Storhaug,
TAKO-Centre, Oslo, 2002 pp13-20.

25. **Hereditary Noninflammatory Arthropathies**
In: Principles and Practice of Medical Genetics, Volume II, Fourth Edition, eds. David L Rimoin, J. Michael Connor, Reed E Pyeritz
Churchill Livingstone, Edinburgh, 2001

24. **Ehlersov-Danlosov Syndróm**
In: Klinická Reumatológia, eds. J Rovenský, KPA Kolektív
Vydavatel'stvo Osveta, Slovak Republick 2000

23. **Hereditary Noninflammatory Arthropathies**
In: Principles and Practice of Medical Genetics, Volume II, Third Edition, eds. David L Rimoin, J. Michael Connor, Reed E Pyeritz
Churchill Livingstone, Edinburgh, 1997

22. **Auditory Dysfunction in Genetic Disorders of the Skeleton**
In: Genetics and Hearing Impairment
Ed: A Martin, A Read, D Stephens
Whurr, London, 1996

21. **Disorders of Collagen and Elastin**
In: Inherited Skin Disorders, ed. J Harper
Butterworth-Heinemann, Oxford, 1996

20. **Sclerosteosis**
In: Congenital Malformation Syndromes, eds. D Donnai & RM Winter.
Chapman & Hall Medical, London, 1995

19. **Dyggve-Melchoir-Clausen syndrome**
In: Congenital Malformation Syndromes, eds. D Donnai & RM Winter.
Chapman & Hall Medical, London, 1995

18. **Osteoglophonic dysplasia**
In: Congenital Malformation Syndromes, eds. D Donnai & RM Winter.
Chapman & Hall Medical, London, 1995

17. **Gigantism**
In: Dysmorphology and Genetics of Cardiovascular Disorders, eds. C.S. Bartsocas, P. Beighton.
HTA Medical Publications, Athens, 1994

16. **Skeletons in the Tower of Fools**
In: Dysmorphology and Genetics of Cardiovascular Disorders, eds. C.S. Bartsocas, P. Beighton.
HTA Medical Publications, Athens, 1994

15. **Other Heritable and Generalized Disorders**
In: McKusick's Heritable Disorders of Connective Tissue, 5th edition, ed. P. Beighton.
C.V. Mosby, St. Louis, 1992

14. **Fibrodysplasia Ossificans Progressiva**
In: McKusick's Heritable Disorders of Connective Tissue, 5th edition, ed. P. Beighton.
C.V. Mosby, St. Louis, 1992

13. **Alkaptonuria** (with P. Berman & S. Srsen)
In: McKusick's Heritable Disorders of Connective Tissue, 5th edition, ed. P. Beighton.
C.V. Mosby, St. Louis, 1992

12. **Ehlers-Danlos Syndrome**
In: McKusick's Heritable Disorders of Connective Tissue, 5th edition, ed. P. Beighton.
C.V. Mosby, St. Louis, 1992

11. **Ehlers-Danlos Syndrome**
In: Recent Advances in Paediatrics 11, ed. T.J. David.
Churchill Livingstone, London, 1992

10. **Hereditary Deafness**
In: Principles and Practice of Medical Genetics,(2nd Ed), eds. E.H. Emery, D.L. Rimoin.
Churchill Livingstone, Edinburgh, 1990

9. **The Diagnosis of Genetic Disease**
In: South African Medical Research, ed. A.J. Brink.
Owen Burgess, Pinetown, 1988

8. **Hereditary Deafness**
In: Principles and Practice of Medical Genetics,eds. E.H. Emery, D.L. Rimoin.
Churchill Livingstone, Edinburgh, 1983

7. **Sclerosing Bone Dysplasias**
In: Progress in Clinical and Biological ResearchVol 104, ed. C.J. Papadatos, C.S. Bartsocas.
Alan R Liss, Inc., New York, 1982

6. **Genetic Diseases and Congenital Malformations**
In: Clinical Medicine in Developing Africa,ed. G.D. Campbell.
David Philip Ltd., Cape Town, 1982

5. **The Arthritides in the Negro Peoples of Southern Africa**
In: Western Diseases, eds. D.R. Burkitt, H.C. Trowell.
E. Arnold, London, 1981 (jointly with L Solomon)

4. **Geographic Variation in Joint Disorders**
In: Recent Advances in Rheumatology, ed. W. Carson Dick
Churchill Livingstone, Edinburgh, 1981

3. **Medical Genetics in Orthopaedic Surgery**
In: Scientific Foundations of Orthopaedics and the Surgery of Trauma, eds. R. Owen & J.W. Goodfellow.
Heinemann, London, 1980

2. **Genetic Mechanisms in Carcinoma**
In: Carcinoma of the Oesophagus, ed. W. Silber.
A.A. Balkema, Cape Town, 1978

1 **Disproportionate Dwarfism in the Newborn**
In: Modern Trends in Human Genetics 2, ed. A.E.H. Emery
Butterworths, London and Boston, 1975

PUBLICATIONS

419. **Vorster A, Beighton P, Ramesar R**
Spondyloepimetaphyseal Dysplasia with Joint Laxity (SEMDJL) Beighton type: Mutation analysis in 10 affected South African families.
Clin Genet 2014 Apr 26.doi: 10.1111/oge.12413

418. **Roberts T, Stephen LXG, Chetty M, Fieggen K, Beighton P**
Trichorhinophalangeal syndrome; premature tooth eruption, oral manifestations and management protocol.
SADJ 69(4):158-161, 2014

417. **Roberts TS, Chetty M, Stephen L, Urban M, Fieggen K, Beighton P**
Rubinstein-Taybi syndrome: Dental manifestations and management.
S Afr J CH 8(1):28-30, 2014

416. **Bertie JD, Thompson D, Beighton P**
The Torg-Winchester form of hereditary osteolysis: orthopaedic manifestations and management.
SA Orthopaedic J 12(2):23-27, 2013

415. **Muller L, Wainwright H, Beighton P**
Ultrasonic diagnosis of perinatal lethal hypophosphatasia.
Ultrasound 21:132-136, 2013

414. **Roberts T, Stephen LXG, Beighton P**
Cleido-cranial dysplasia: a review of the dental, historical, and practical implications with an overview of the South African experience.
Oral Surg Oral Med Oral Pathol Oral Radiol Endod 115(1):46-55, 2013

413. **Dandara C, Scott C, Urban M, Fieggen K, Arendse R, Beighton P**
Confirmation of the recurrent ACVR1 617G>A mutation in South Africans with fibrodysplasia ossificans progressiva.
S Afr Med J 102(7):631-633,2012 *(IF 2.042)*

412. **Beighton P, Fieggen K, Wonkam A, Greenberg J, Ramesar R**
The University of Cape Town's contribution to medical genetics in Africa - from the past into the future.
S Afr Med J 102(6):446-448,2012 *(IF 2.042)*

411. **Beighton P, Hamersma H, Brunkow ME**
SOST-Related Sclerosing Bone Dysplasias in: GeneReviews at GeneTests: Medical Genetics Information Resource [database online]. January 2012
Available at: http://www.genetests.org

410. **Wessels A, Wainwright H, Beighton P**
Atelosteogenesis type I. Autopsy findings.
Pediatr Dev Pathol 14:496-500, 2011 *(IF 1.034)*

409. **Winship WS, Beighton P**
Genetic disorders in the Indian Community of South Africa.
S Afr Med J 101:481-484, 2011 *(IF 1.676)*

408. **Bertie JD, Thompson D, Beighton P**
Cleidocranial dysplasia presenting as familial coxa vara in a South African family.
SA Orthopaedic Journal 10(2):34-38, 2011

407. **Roberts T, Stephen LXG, Scott C, Urban M, Sudi S, Beighton P**
Fibrodysplasia ossificans progressive (FOP) in South Africa: dental implications in 5 cases.
Oral Surg Oral Med Oral Pathol Oral Radiol Endod 112:11-18, 2011 *(IF 1.997)*

406. **Kelley BP, Symoens S, Bonafe L, Baldridge D, Malfait F, Elcioglu N, Napierala D, Krakow D, Beighton P, Superti-Furga A, Depaepe A, Lee B**
Mutations in the FKBP10 cause recessive osteogenesis imperfecta and Bruck syndrome.
J Bone Miner Res 26(3):666-672, 2011 *(IF. 6.043)*

405. **Scott C, Urban M, Arendse R, Dandara C, Beighton P**
Fibrodysplasia Ossificans Progressiva in South Africa. Difficulties in management in a developing country.
J Clin Rheumatol 17:37-41, 2011

404. **Wainwright H, Beighton P**
Warfarin Embryopathy; foetal manifestations.
Virchows Archiv 457(6):735-739, 2010 *(IF 2.082)*

403. **Wainwright H, Beighton P**
Dyssegmental Dysplasia with Bone Entrapment and Visceral Manifestations.
Pediatr Dev Pathol 13(1):46-49, 2010 (*IF 1.156*)

402. **Borck G, Beighton P, Wilhelm C, Kohlhase J, Kubisch C**
Arterial rupture in non-classic Ehlers-Danlos syndrome with COL5A1 mutation.
Am J Med Genet Part A 152A:2090-2093, 2010 (*IF 2.404*)

401. **Arendse RE. Gcelu A, Scott C, Beighton P, Kalla AA**
Do the radiographic features of joint destruction in tophaceous gout imply a different pathophysiology to that of rheumatoid and psoriatic arthritis?
Clin Rheumatol 29(10):1181-1183, 2010 (*IF 1.668*)

400. **Wainwright H, Beighton P**
Lethal epiphyseal stippling in the foetus and neonate; pathological implications.
Virchows Archiv 456:301-308, 2010 *(IF 2.082)*

399. **Roberts T, Stephen LXG, Fieggen K, Beighton P**
Wolf-Hirschhorn syndrome: orofacial manifestations and management.
J Clin Pediatr Dent 34(2):173-176, 2009

398. **Arendse R, Brink P, Beighton P**
Hereditary bone dysplasia with pathological fractures and nodal osteoarthropathy.
Skel Radiol 38:1197-1203, 2009 *(IF 1.568*

397. **Tapley E, Beighton P**
Calcification of breast tissue in the Ehlers-Danlos syndrome.
The Breast Journal 15(5):537-539, 2009 *(IF 1.610)*

396. **Wicomb G, Beighton P**
Familial Hyperdontia in the Deciduous Dentition.
J Int Dent Med Res 2(1):1-5, 2009

395. **Fakir E, Roberts T, Stephen LXG, Beighton P**
Klippel-Trenaunay-Weber syndrome; oro-dental manifestations and management.
Oral Surg Oral Med Oral Pathol Oral Radiol Endod 107(6):754-758,2009 *(IF 1.997)*

394. **Wainwright H, Beighton P**
Multiple malformations; a possible Sonic hedgehog phenotype?
Virchows Archiv 454(3):345-347, 2009 (*IF 2.082*)

393. **Wu H, Wainwright HH, Beighton P**
Tetraphocomelia with the Waardenburg syndrome.
Clin Dysmorphol 18(2):112-115, 2009 (*IF 0.521*)

392. **Wainwright H, Beighton P**
Osteogenesis imperfecta type IIa with pseudoanencaphaly.
Clin Dysmorphol 17(4):271-272, 2008 *(0.521)*

391. **Wainwright H, Beighton P**
Achondrogenesis type II with cutaneous hamartomata.
Clin Dysmorphol 17(3):207-209, 2008 *(0.521)*

390. **Winship W, Beighton P**
Dyssegmental Dysplasia
Clin Dysmorphol 17(2):95-98, 2008 *(0.521)*

389. **Wainwright H, Beighton P**
Visceral manifestations of hypochondrogenesis.
Virchows Arch 453:203-207, 2008 *(IF 2,082)*

388. **Stephen L, Sellars S, Beighton P**
Dentistry in profound childhood deafness in South Africa.
Int Dent SA 2(4):66-72, 2007

387. **Child A, Comeglio P, Arno G, Beighton P**
Marfan syndrome in South Africa – a molecular genetic approach to diagnosis.
S Afr Med J 97(9):845-847, 2007

386. **Beighton P, Kozlowski K**
Spondylometaphyseal dysplasia Sutcliffe type – case report.
Pol J Radiol 72(1):76-78, 2007

385. **Wainwright H, Beighton P**
Osteogenesis imperfecta and holoprosencephaly.
Clin Dysmorphol 16 (3):189-191, 2007

384. **Beighton P, Mennen U, Golele SS, Urban M**
Orthopaedic implications of heritable osteolysis in South Africa.
SA Orthopaedic Journal 6(2):26-32, 2007

383. **Hundleby CJB, Beighton P**
Duplication of the Nipples and Areolae.
Clin Dysmorphol 16(2):115-116, 2007

382. **Beighton P, Hamersma H, Brunkow M**
SOST-Related sclerosing bone dysplasias. In: GeneReviews at GeneTests: Medical Genetics Information Resource [database online].
Available at http://www.genetests.org, update February 2007.

381. **Khumalo NP, Pillay K, Beighton P, Wainwright H, Walker B, Saxe N, Mayosi BM, Bateman ED**
Poikiloderma, tendon contractures, and pulmonary fibrosis: a new autosomal dominant syndrome?
Brit J Derm **155**:1057-1061, 2006

380. **Roberts TS, Stephen L, Beighton P**
Osteoglophonic dysplasia: dental and orthodontic implications.
Orthod Craniofacial Res **9**:153-156, 2006

379. **Farrow EG, Davis SI, Mooney SD, Beighton P, Mascarenhas L, Gutierrez YR, Pitukcheewanont P, White KE**
Extended mutational analysis of FGFR1 in osteoglophonic dysplasia (OD).
Am J Med Genet **140A(5)**:537-539, 2006

378. **Kozlowski K, Basel D, Beighton P**
Retrospective diagnosis of chondrodysplasia punctata.
Australas Radiol **50**:55-58, 2006

377. **Gardner JC, vanBezooijen RL, Mervis B, Hamdy NA, Lowik CW, Hamersma H, Beighton P, Papapoulos SE**
Bone mineral density in sclerosteosis; affected individuals and gene carriers.
J Clin Endocrinol Metab **90(12)**:6392-6395, 2005

376. **Mokete L, Robertson A, Viljoen D, Beighton P**
Bruck syndrome: congenital joint contractures with bone fragility.
J Orthop Sci **10(6)**:641-646, 2005

375. **Stephen LXG, Holmes H, Roberts T, Fieggen K, Beighton P**
Orthodontic Management of Achondroplasia in South Africa
S Afr Med J **95(8)**:588-589, 2005376

374. **Gleghorn L, Ramesar R, Beighton P, Wallis G**
A mutation in the variable repeat region of the aggrecan gene (AGC1) causes a form of spondyloepiphyseal dysplasia associated with severe, premature osteoarthritis.
Am J Hum Genet **77(3)**:484-490, 2005

373. **Roberts T, Stephen LXG, Naidoo T, Fieggen K, Beighton P**
Freeman-Sheldon Syndrome; orthodontic implications
J Clin Pediatr Dent **29(2)**:267-273, 2005374

372. **Kozlowski K, Basel D, Beighton P**
Chondrodysplasia punctata and maternal lupus erythematosus.
Clinical Genetics **66(6)**:545-549,2004

371. **Jeftha A, Stephen L, Morkel JA, BeightonP**
Crouzonodermoskeletal syndrome.
J Clin Pediatr Dent **28(2)**:173-176, 2004372

370. **Beighton P, Hamersma H, Brunkow M**
SOST-Related sclerosing bone dysplasias. In: GeneReviews: Genetic Disease Online Reviews at GeneTests-GeneClinics [database online].
Available at http://www.geneclinics.org, update September 2004.

369. **Wicomb GM, Stephen LXG, Beighton P**
Dental implications of tooth-nail dysplasia (Witkop syndrome): a report of an affected family and an approach to dental management.
J Clin Pediatr Dent **28(2)**:107-112, 2004

368. **Honey EM, van Rensburg M, Knoll DP, Mienie LJ, van der Werke L, Beighton P**
Spondyloenchondromatosis with D-2-hydroglutaric aciduria: a report of a second patient with this unusual combination.
Clin Dysmorph **12(2)**:95-99, 2003369

367. **Hamersma H, Gardner J, Beighton P**
The Natural History of Sclerosteosis.
Clin Genet **63**:191-196, 2003368.

366. **Stephen LXG, Beighton PH**
Oro-dental Manifestations of the Schwartz-Jampel Syndrome.
J Clin Pediatr Dent **27(1)**:67-70, 2002

365. **Beighton P, Kozlowski K**
Nowa, odrębna postać dysplazji kręgowo-nasadowo-prznasadowej.
Polish New Pediat **2**:63-66, 2002366.

364. **Eyre S, Roby P, Wolstencroft K, Spreckley K, Aspinwall R, Bayoumi R, Al-Gazali L, Ramesar R, Beighton P, Wallis G**
Identification of a locus for a form of spondyloepiphyseal dysplasia on chromosome 15q26.1: exclusion of aggrecan as a candidate gene.
J Med Genet **39(9)**:634-638, 2002365.

363. **Hall C, Beighton et al**
Nosology and classification of constitutional disorders of bone.
Am J Med Genet 113: 65-77, 2002.

362. **Kozlowski K, Godlonton J, Beighton P**
Chondrodysplasia punctata – retrospective diagnosis.
Hungarian Radiol 76(3): 118-122, 2002.

361. **Kozlowski K, Godlonton J, Gardner J, Beighton P**
Lethal non-rhizomelic dysplasia epiphysealis punctata.
Clin Dysmorph 11(3): 202-208, 2002.

360. **Beighton P, Hamersma H, Brunkow M**
SOST-Related sclerosing bone dysplasias. In: GeneReviews: Genetic Disease Online Reviews at GeneTests-GeneClinics [database online].
Available at http://www.geneclinics.org, June 2002.

359. **Staehling-Hampton K, Proll S, Paeper BW, Zhao L, Charmley P, Brown A, Gardner JC, Galas D, Schatzman RC, Beighton P, Papapoulos S, Hamersma H, Brunkow M**
A 52 kb deletion in the *SOST – MEOX1* intergenic region on 17q12-q21 is associated with van Buchem disease in the Dutch population.
Am J Med Genet 110: 144-152, 2002.

358. **Stephen LXG, Basel D, Beighton PH**
Developmental absence of the premolar teeth: dental management.
Int J Paed Dent **12**:216-219, 2002.

357. **Stephen LXG, Beighton P**
Dental management of severe dentinogenesis imperfecta in a mild form of osteogenesis imperfecta.
J Clin Pediatr Dent **26(2)**:131-136, 2002

356. **Stephen LXG, Hamersma H, Gardner J, Beighton P**
Dental and oral manifestations of sclerosteosis.
Int Dent J **51(4)**:7-10, 2001

356a **Gong Y, Slee RB, Fukai N, Rawadi G, Roman-Roman S, Reginato AM, Wang H, Cundy T, Glorieux FH, Lev D, Zacharin M, Oexle K, Marcelino J, Suwairi W, Heeger S, Sabatakos G, Apte S, Adkins WN, Allgrove J, Arslan-Kirchner M, Batch JA, Beighton P et al; Osteoporosis-Pseudoglioma Syndrome Collaborative Group.**
LDL receptor-related protein 5 (LRP5) affects bone accrual and eye development.
Cell. 2001 Nov 16;107(4):513-23.

355. **Basel D, Beighton P, Kozlowski K**
Unusual x-ray appearances of a congenital bone disease of unknown aetiology.
J Ped Radiol **31(3)**:212 only, 2001356.

354. **Stephen LXG, Behardien N, Beighton P**
Focal dermal hypoplasia: management of complex dental features.
J Clin Pediatr Dent **25(4)**:259-261, 2001

353. **Reichenberger E, Tiziani V, Watanabe S, Park L, Ueki Y, Santanna C, Baur ST, Shiang R, Grange DK, Beighton P, Gardner J, Hamersma H, Sellars S, Ramesar R, Lidral AC, Sommer A, Raposo do Amaral CM, Gorlin RJ, Mulliken JB, Olsen BR**
Autosomal dominant craniometaphyseal dysplasia is caused by mutations in the transmembrane protein ANK.
Am J Hum Genet **68(6)**:1321-1326, 2001

352. **Brunkow ME, Gardner J, Van Ness J, Paeper B, Kovacevich B, Proll S, Skonier J, Zhao L, Sabo P, Fu Y-H, Alisch R, Gillette L, Colbert T, Tacconi P, Galas D, Hamersma H, Beighton P, Mulligan J**
Bone dysplasia sclerosteosis results from loss of the SOST gene product, a novel cystine knot-containing protein.
Am J Hum Genet. **68**:577-589, 2001

351. **Nicole S, Davoine C-S, Topaloglu H, Cattolico L, Barral D, Beighton P, Ben Hamida C, Hammouda H, Cruaud C, White PS, Samson D, Urtizberea JA, Lehmann-Horn F, Weissenbach J, Hentati F, Fontaine B**
Perlecan, the major proteoglycan of basement membranes, is altered in Schwartz-Jampel syndrome (chondrodystrophic myotonia).
Nature Genetics **26(4)**:480-483, 2000352.

350. **Basel D, Sobey G, Gardner J, Beighton P**
The Gordon Syndrome Revisited
SAMJ **90**(9): 864-867, 2000351.

349. **Ianakiev P, Kilpatrick MW, Toudjarska I, Basel D, Beighton P, Tsipouras P.**
Split-hand/split-foot malformation is caused by mutations in the p63 gene on 3q27.
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