Name	Professor Inderjeet Dokal FMedSci
Current Position & Affiliation	Emeritus Professor and Honorary Consultant in Haematology, Centre for Genomics and Child Health, Blizard Institute, Queen Mary University of London and Barts Health NHS Trust
Country	England, United Kingdom
Major Field	Inherited bone marrow failure particularly those due to defective telomere maintenance

Educational Background: Undergraduate and postgraduate qualifications

MBChB	1983	University of Leicester
MRCP	1986	Royal College of Physicians (London)
MRCPath	1993	Royal College of Pathologists (London)
MD	1994	University of Leicester
MRCPCH	1997	Royal College of Paediatrics and Child Health (London)
FRCPCH	1998	Royal College of Paediatrics and Child Health (London)
FRCP	1999	Royal College of Physicians (London)
FRCPath	2001	Royal College of Pathologists (London)

Professional Experience

Inderjeet Dokal graduated in Medicine from the University of Leicester in 1983. He moved to Hammersmith Hospital in 1984 where he received his post-graduate clinical and research training. He was appointed Consultant in Paediatric Haematology in 1995 and was conferred the title of Professor of Haematology at Imperial College London in 2003. In 2006 he was recruited to the Chair of Child Health at Barts and The London School of Medicine and Dentistry, Queen Mary University of London. He is also Honorary Consultant in Haematology (Barts Health) and was Centre Lead for Genomics and Child Health (2006-2024). For over four decades Inderjeet Dokal has had a clinical and research interest in the inherited bone marrow failure syndromes. His group has made seminal advances in the bone marrow failure (BMF) field including the identification and characterization of BMF due to defective telomere maintenance. He was elected Fellow of The Academy of Medical Sciences in 2010.

Other Experience and Professional Memberships

Medical Research Council (MRC) Training Fellowship, 1988

Elected to 1942 Club, 2000

Awarded Personal Chair (Professor of Haematology) at Imperial College London, 2003

Recruited to Chair of Child Health at Barts and The London/QMUL, 2006

Centre Lead, Barts and The London/QMUL, 2006-2024

Chair of Paediatric East London Cancer Network, 2007-2011

Director of Paediatric Clinical Academic Group (Barts Health), 2008-2010

Member of MRC Leukaemia Data Monitoring and Ethics Committee (LDMEC), 2007-2010

Chair of MRC Leukaemia Data Monitoring and Ethics Committee (LDMEC), 2010-2014

Elected Fellow of The Academy of Medical Sciences (FMedSci), 2010-present
Member of Sectional Committee 4 of The Academy of Medical sciences, 2011-2013
Member and then Chair of Scientific Committee for Bone Marrow Failure (ASH), 2015-2018
Member of European School of Hematology (ESH), 2016-2018
Member of Scientific Advisory Board for Action Medical Research, 2016-2020

Main Scientific Publications

Inderjeet Dokal is author/co-author of 178 peer reviewed publications. Listed below are 20 of his significant publications:

1 Dokal I, Bungey J, Williamson P, Oscier D, Hows JM, and Luzzatto L. Dyskeratosis congenita fibroblasts are abnormal and have unbalanced chromosomal rearrangements. *Blood* 1992; 80: 3090-3096.

2 Heiss NS, Knight SW, Vulliamy TJ, Klauck SM, Wiemann S, Mason PJ, Poustka A, Dokal I. X-linked dyskeratosis congenita is caused by mutations in a highly conserved gene with putative nucleolar functions. *Nature Genetics* 1998; 19: 32-38.

3 Vulliamy T, Marrone A, Goldman F, Dearlove A, Bessler M, Mason PJ, Dokal I. The RNA component of telomerase is mutated in autosomal dominant dyskeratosis congenita. *Nature* 2001; 413: 432-435.

4 Vulliamy T, Marrone A, Dokal I, Mason PJ. Association between aplastic anaemia and mutations in telomerase RNA. *Lancet* 2002; 359: 2168-2170.

5 Vulliamy T, Marrone A, Szydlo R, Walne A, Mason PJ, Dokal I. Disease anticipation is associated with progressive telomere shortening in families with dyskeratosis congenita due to mutations in *TERC*. *Nature Genetics* 2004; 36: 447-449.

6 Vulliamy TJ, Marrone A, Knight SW, Walne A, Mason PJ, Dokal I. Mutations in dyskeratosis congenita: their impact on telomere length and the diversity of clinical presentation. *Blood* 2006; 107: 2680-2685.

7 Walne AJ, Vulliamy T, Marrone A, Beswick R, Kirwan M, Masunari, Y, Al-Qurashi F, Aljurf M, Dokal I. Genetic Heterogeneity in autosomal recessive dyskeratosis congenita with one subtype due to mutations in the telomerase-associated protein NOP10. *Human Molecular Genetics* 2007; 16: 1619-1629.

8. Marrone A, Walne A, Tamary H, Masunari Y, Kirwan M, Beswick R, Vulliamy T, D okal I. Telomerase reverse transcriptase homozygous mutations in autosomal rec essive dyskeratosis congenita and Hoyeraal-Hreidarsson syndrome. *Blood* 2007; 110: 4198-2205.

9 Vulliamy T, Beswick R, Kirwan M, Marrone A, Digweed M, Walne A, Dokal I. Mutations in the telomerase component NHP2 cause the premature ageing syndrome dyskeratosis congenita. *Proceedings of the National Academy of Sciences (USA)* 2008; 105: 8073-8078.

10 Kirwan M, Walne AJ, Plagnol V, Velangi M, Ho A, Hossain U, Vulliamy T, Dokal I. Exome sequencing identifies autosomal dominant mutations in the signal recognition particle 72kDa component associated with familial aplasia and myelodysplasia. *American Journal of Human Genetics* 2012, 90: 888-892.

11 Walne AJ, Vulliamy T, Kirwan M, Plagnol V, Dokal I. Constitutional mutations in *RTEL1* cause severe dyskeratosis congenita. *American Journal of Human Genetics* 2013; 92 (3): 448-453.

12 Tummala H, Kirwan M, Walne AJ, Hossain U, Jackson N, Pondarre C, Plagnol V, Vulliamy T, Dokal I. *ERCC6L2* mutations link a distinct bone marrow failure syndrome to DNA repair and mitochondrial function. *American Journal of Human Genetics* 2014; 94: 246-256.

13 Tummala H, Walne A, Collopy L, Cardoso S, de la Fuente J, Lawson S, Powell J, Cooper C, Foster A, Mohammed S, Plagnol V, Vulliamy T, Dokal I. Poly(A)-specific ribonuclease deficiency impacts telomere biology causing dyskeratosis congenita. *Journal of Clinical Investigation* 2015; 125: 2151-2160.

14 Collopy LC, Walne AJ, Cardoso S, de La Fuente J, Mohamed M, Toriello H, Tamary H, Ling AJYV, Lloyd T, Kassam R, Tummala H, Vulliamy TJ, Dokal I. Triallelic and epigenetic-likeinheritance in human disorders of telomerase. *Blood* 2015; 126: 176-184.

15 Tummala H, Walne AJ, Williams M, Bockett N, Collopy L, Cardoso S, Ellison A, Wynn R, Leblanc T, Fitzgibbon J, Kelsell DP, van Heel DA, Payne E, Plagnol V, Dokal I, Vulliamy T. *DNAJC21* mutations link a cancer prone bone marrow failure syndrome to corruption in 60S ribosome subunit maturation. *American Journal of Human Genetics* 2016; 99: 115-124.

16 Rio-Machin A, Vulliamy T, Hug N, Walne A, Tawana K, Cardoso S, Ellison A, Pontikos N, Wang J, Hemanth Tummala H, Al Seraihi AFH, Alnajar J, Copley F, Armes H, Barnett M, Bloor A, Bodor C, Bowen D, Fenaux P, Green A, Hallahan A, Hjorth-Hansen H, Hossain U, Killick S, Lawson S, Layton M, Male AM, Marsh J, Mehta P, Mous R, Nomdedéu JF, Owen C, Pavlu J, Payne E, Protheroe R, Preudhomme C, Pujol-Moix N, Renneville A, Russell N, Saggar A, Sciuccati G, Taussig D, Toze C, Uyttebroeck A, Vandenberghe P, Schlegelberger B, Ripperger T, Steinemann D, Wu J, Mason J, Page P, Akiki SE, Reay K, Cavenagh JD, Plagnol V, Caceres JF, Fitzgibbon J, Dokal I. The complex genetic landscapes of familial MDS and AML reveals pathogenic germline variants. *Nature Communications* 2020;11(1):1044. doi: 10.1038/s41467-020-14829-5.

17 Tummala H, Walne AJ, Bewicke-Copley F, Ellison A, Pontikos N, Bridger MG, Rio-Machin A, Sidhu JK, Wang J, Hasle H, Fitzgibbon J, Vulliamy T, Dokal I. A frameshift variant in specificity protein 1 triggers superactivation of Sp1-mediated transcription in familial bone marrow failure. *Proceedings of the National Academy of Sciences (USA)* 2020; 117(29):17151-17155.

18 Walne AJ, Vulliamy T, Bewicke-Copley F, Wang J, Alnajar J, Bridger MG, Ma B, Tummala H, Dokal I. Genome wide whole blood transcriptome profiling across inherited bone marrow failure subtypes. *Blood Advances* 2021; 5(23): 5360-5371.

19 Tummala H, Walne A, Buccafusca R, Alnajar J, Szabo A, Robinson P, McConkie-Rosell A, Wilson M, Crowley S, Kinsler V, Ewins AM, Madapura PM, Patel M, Pontikos N, Codd V, Vulliamy T, Dokal I. Germline thymidylate synthase deficiency impacts nucleotide metabolism and causes dyskeratosis congenita. *American Journal of Human Genetics* 2022; 109:1472-1483.

20 Tummala H, Walne AJ, Badat M, Patel M, Walne AM, Alnajar J, Chow CC, Albursan I, Frost JM, Ballard D, Killick S, Szitányi P, Kelly A, Raghavan M, Powell C, Raymakers R, Todd T, Mantadakis E, Polychronopoulou S, Pontikos N, Liao T, Pradeepa MM, Hossain U, Vulliamy T, Dokal I. The evolving genetic landscape of telomere biology disorder dyskeratosis congenita. *EMBO Molecular Medicine* 2024; 16(10): 2560-2582.