CURRICULUM VITAE

Professor Anand Kumar SAGGAR

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Qualifications

Doctorate of Medicine	2009
FRCP	2004
CCST (Clinical Genetics)	2000
MRCP (UK)	1986
MB BS (University of London)	1982
St. Bartholomew's Medical College Hospital	
General Medical Council (UK)	2574251
Irish Medical Council (Republic of Ireland)	430109

Professional Memberships

British Society of Human Genetics
Clinical Genetics Society (Honorary General Secretary)
The Genetical Society
American Society of human Genetics
Renal Association (UK)
International Society of Nephrology
British Hypertension Society
Royal Society of Medicine (Former President of the Medical Genetics Section)
Society of Apothecaries

Languages

English, Swedish, Hindi, Punjabi, limited French and Spanish

CV SUMMARY

Professor Saggar has recently been appointed as a Visiting Consultant at the Crumlin Hospital, Genetics Department in Dublin and Cork University Hospital, Cork. He is also director of the clinic, 'Inherited Disorders of Connective Tissue' and founder/lead of the 'International Gene Clinic'. These clinics are the largest UK private genetic clinics for genetic conditions and have an international reputation of good standing. The clinics offer a complete clinical and laboratory diagnostic service for patients and families with all forms of inherited conditions.

Professor Saggar recently retired as an NHS Consultant in the South West Thames Regional Genetics Department at St George's Hospital/Honorary Senior Lecturer within St Georges University of London Medical School but still retains an honorary NHS contract. He was the Senior Consultant within the NHS multidisciplinary team responsible for providing services for a population of approximately 2.7 million across the region.

Professor Saggar continues to take part in regional clinics in both general adult and paediatric dysmorphology alongside specialist clinics for patients with connective tissue diseases, kidney disorders, tuberous sclerosis, cancer family histories and other speciality areas. He was responsible for leading the Marfan/Aortopathy syndrome (connective tissue diseases) clinics at St George's and the Brompton/ Harefield Hospitals. Professor Saggar has a broad and comprehensive record of experience in general medicine, namely respiratory, diabetes/endocrinology, cardiology, intensive care, nephrology and neurology.

Professor Saggar regularly attends and contributes to the weekly Genetics Unit Journal Club and is an attending member of the British Society of Human Genetics, Clinical Genetics Society and the London Dysmorphology Club. He also contributes to international genetic meetings.

Professor Saggar has a long-standing interest in ethnic minority health and equity of access. In 2003 he helped design and complete a collaborative, multi-centre pilot survey examining ethnic access to 3 UK genetic centres.

He was Honorary General Secretary to the Clinical Genetics Society (CGS) for 4 years and was also on the Board of GenePool, a formative early part of the Department of Health's 'national electronic Library of Health' initiative to provide an information service and web portal for genetics information and incorporation of genetics knowledge into clinical practice. Professor Saggar also sits on numerous other advisory committees including Past President of the Medical Genetics Section of the Royal Society of Medicine, London, lead for International Development for the CGS and was formerly the Chair of the Medical Advisory Committee at a private hospital.

Professor Saggar is a founder trustee and medical advisor to the Polycystic Kidney Disease Charity (UK) and also medical advisor and former Chair to SPECS, an umbrella organization for local groups and societies for people with impaired vision.

Professor Saggar lectures locally, nationally and internationally. He is actively committed to research and is currently collaborating in several major national and international research projects. He has over 40 publications and continues to publish actively, both peer-reviewed original papers and book chapters.

PRESENT APPOINTMENT

March 2021 to date	**CONSULTANT IN CLINICAL GENETICS** CRUMLIN CHILDREN'S HOSPITAL, DUBLIN
Sept 2000 to Nov 2021	**CONSULTANT PHYSICIAN AND CLINICAL GENETICIST** **HONORARY SENIOR LECTURER IN MEDICINE** ST GEORGE'S HOSPITAL MEDICAL SCHOOL, LONDON

PREVIOUS POSTS

Oct 1996	Jul 2000	**SENIOR REGISTRAR IN CLINICAL GENETICS AND HONORARY LECTURER IN MEDICINE** ST GEORGE'S HOSPITAL MEDICAL SCHOOL
Oct 1993	Sept 1996	**WILLIAMS FELLOW OF THE UNIVERSITY OF LONDON** ST GEORGE'S HOSPITAL MEDICAL SCHOOL
May 1991	Sept 1993	**CLINICAL RESEARCH FELLOW** (Honorary Registrar) ST GEORGE'S MEDICAL SCHOOL, BLOOD PRESSURE UNIT,
Nov 1990	Mar 1991	**REGISTRAR IN GENERAL MEDICINE** ST. ANDREW'S HOSPITAL, LONDON
April 1988	Sept 1990	**RESEARCH FELLOW IN RENAL MEDICINE** KAROLINSKA INSTITUTE, STOCKHOLM
Feb 1987	Jan 1988	**REGISTRAR TO THE LIVER UNIT**

KING'S COLLEGE HOSPITAL, LONDON

Mar 1986 Feb 1986 **REGISTRAR WITH SPECIAL INTERESTS**
CHELMSFORD HOSPITALS, ESSEX

PUBLICATIONS

1. Kaplanis J, Samocha KE, Wiel L, Zhang Z, Arvai KJ, Eberhardt RY, Gallone G, Lelieveld SH, Martin HC, McRae JF, Short PJ, Torene RI, de Boer E, Danecek P, Gardner EJ, Huang N, Lord J, Martincorena I, Pfundt R, Reijnders MRF, Yeung A, Yntema HG; **Deciphering Developmental Disorders Study**, Vissers LELM, Juusola J, Wright CF, Brunner HG, Firth HV, FitzPatrick DR, Barrett JC, Hurles ME, Gilissen C, Retterer K. Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature. 2020 Oct;586(7831):757-762.

2. Wai HA, Lord J, Lyon M, Gunning A, Kelly H, Cibin P, Seaby EG, Spiers-Fitzgerald K, Lye J, Ellard S, Thomas NS, Bunyan DJ, Douglas AGL, Baralle D; **Splicing and disease working group.** Blood RNA analysis can increase clinical diagnostic rate and resolve variants of uncertain significance. Genet Med. 2020 Jun;22(6):1005-1014. Erratum in: Genet Med. 2020 Apr 1;

3. Perrino PA, Talbot L, Kirkland R, Hill A, Rendall AR, Mountford HS, Taylor J; WGS500 Consortium, Buscarello AN, Lahiri N, **Saggar A**, Fitch RH, Newbury DF. Multi-level evidence of an allelic hierarchy of USH2A variants in hearing, auditory processing and speech/language outcomes. Commun Biol. 2020 Apr 20;3(1):180.

4. Tye C, Mcewen FS, Liang H, Underwood L, Woodhouse E, Barker ED, Sheerin F, Yates JRW, Bolton PF; **Tuberous Sclerosis 2000 Study Group**. Long-term cognitive outcomes in tuberous sclerosis complex. Dev Med Child Neurol. 2020 Mar;62(3):322-329.

5. Rio-Machin A, Vulliamy T, Hug N, Walne A, Tawana K, Cardoso S, Ellison A, Pontikos N, Wang J, Tummala H, Al Seraihi AFH, Alnajar J, Bewicke-Copley F, Armes H, Barnett M, Bloor A, Bödör 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 EM, Protheroe RE, Preudhomme C, Pujol-Moix N, Renneville A, Russell N, **Saggar A**, Sciuccati G, Taussig D, Toze CL, Uyttebroeck A, Vandenberghe P, Schlegelberger B, Ripperger T, Steinemann D, Wu J, Mason J, Page P, Akiki S, Reay K, Cavenagh JD, Plagnol V, Caceres JF, Fitzgibbon J, Dokal I. The complex genetic landscape of familial MDS and AML reveals pathogenic germline variants. Nat Commun. 2020 Feb 25;11(1):1044.

6. Aitken S, Firth HV, McRae J, Halachev M, Kini U, Parker MJ, Lees MM, Lachlan K, Sarkar A, Joss S, Splitt M, McKee S, Németh AH, Scott RH, Wright CF, Marsh JA, Hurles ME, FitzPatrick DR; **DDD Study**. Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. Am J Hum Genet. 2019 Nov 7;105(5):933-946.

7. Tye C, Mcewen FS, Liang H, Underwood L, Woodhouse E, Barker ED, Sheerin F, Yates JRW, Bolton PF; **Tuberous Sclerosis 2000 Study Group**. Long-term cognitive outcomes in tuberous sclerosis complex. Dev Med Child Neurol. 2019 Sep 19. doi: 10.1111/dmcn.14356.

8. Ceroni F, Aguilera-Garcia D, Chassaing N, Bax DA, Blanco-Kelly F, Ramos P, Tarilonte M, Villaverde C, da Silva LRJ, Ballesta-Martínez MJ, Sanchez-Soler MJ, Holt RJ, Cooper-Charles L, Bruty J, Wallis Y, McMullan D, Hoffman J, Bunyan D, Stewart A, Stewart H, Lachlan K; DDD Study, Fryer A, McKay V, Roume J, Dureau P, **Saggar A**, Griffiths M, Calvas P, Ayuso C, Corton M, Ragge NK. New GJA8 variants and phenotypes highlight its critical role in a broad spectrum of eye anomalies. Hum Genet. 2019 Sep;138(8-9):1027-1042.

9. O'Donnell-Luria AH, Pais LS, Faundes V, Wood JC, Sveden A, Luria V, Abou Jamra R, Accogli A, Amburgey K, Anderlid BM, Azzarello-Burri S, Basinger AA, Bianchini C, Bird LM, Buchert R, Carre W, Ceulemans S, Charles P, Cox H, Culliton L, Currò A; **Deciphering Developmental Disorders (DDD) Study**, Demurger F, Dowling JJ, Duban-Bedu B, Dubourg C, Eiset SE, Escobar LF, Ferrarini A, Haack TB, Hashim M, Heide S, Helbig KL, Helbig I, Heredia R, Héron D, Isidor B, Jonasson AR, Joset P, Keren B, Kok F, Kroes HY, Lavillaureix A, Lu X, Maas SM, Maegawa GHB, Marcelis CLM, Mark PR, Masruha MR, McLaughlin HM, McWalter K, Melchinger EU, Mercimek-Andrews S, Nava C, Pendziwiat M, Person R, Ramelli GP, Ramos LLP, Rauch A, Reavey C, Renieri A, Rieß A, Sanchez-Valle A, Sattar S, Saunders C, Schwarz N, Smol T, Srour M, Steindl K, Syrbe S, Taylor JC, Telegrafi A, Thiffault I, Trauner DA, van der Linden H Jr, van Koningsbruggen S, Villard L, Vogel I, Vogt J, Weber YG, Wentzensen IM, Widjaja E, Zak J, Baxter S, Banka S, Rodan LH. Heterozygous Variants in KMT2E Cause a Spectrum of Neurodevelopmental Disorders and Epilepsy. Am J Hum Genet. 2019 Jun 6;104(6):1210-1222.

10. Gorman KM, Meyer E, Grozeva D, Spinelli E, McTague A, Sanchis-Juan A, Carss KJ, Bryant E, Reich A, Schneider AL, Pressler RM, Simpson MA, Debelle GD, Wassmer E, Morton J, Sieciechowicz D, Jan-Kamsteeg E, Paciorkowski AR, King MD, Cross JH, Poduri A, Mefford HC, Scheffer IE, Haack TB, McCullagh G; **Deciphering Developmental Disorders Study**; UK10K Consortium; NIHR BioResource, Millichap JJ, Carvill GL, Clayton-Smith J, Maher ER, Raymond FL,

Kurian MA. Bi-allelic Loss-of-Function CACNA1B Mutations in Progressive Epilepsy-Dyskinesia. Am J Hum Genet. 2019 May 2;104(5):948-956.

11. Loveday C, Tatton-Brown K, Clarke M, Westwood I, Renwick A, Ramsay E, Nemeth A, Campbell J, Joss S, Gardner M, Zachariou A, Elliott A, Ruark E, van Montfort R; **Childhood Overgrowth Collaboration**, Rahman N. Mutations in the PP2A regulatory subunit B family genes PPP2R5B, PPP2R5C and PPP2R5D cause human overgrowth. Hum Mol Genet. 2015 Sep 1;24(17):4775-9. Epub 2015 May 13. Erratum in: Hum Mol Genet. 2019 May 1;28(9):1578.

12. Snijders Blok L, Rousseau J, Twist J, Ehresmann S, Takaku M, Venselaar H, Rodan LH, Nowak CB, Douglas J, Swoboda KJ, Steeves MA, Sahai I, Stumpel CTRM, Stegmann APA, Wheeler P, Willing M, Fiala E, Kochhar A, Gibson WT, Cohen ASA, Agbahovbe R, Innes AM, Au PYB, Rankin J, Anderson IJ, Skinner SA, Louie RJ, Warren HE, Afenjar A, Keren B, Nava C, Buratti J, Isapof A, Rodriguez D, Lewandowski R, Propst J, van Essen T, Choi M, Lee S, Chae JH, Price S, Schnur RE, Douglas G, Wentzensen IM, Zweier C, Reis A, Bialer MG, Moore C, Koopmans M, Brilstra EH, Monroe GR, van Gassen KLI, van Binsbergen E, Newbury-Ecob R, Bownass L, Bader I, Mayr JA, Wortmann SB, Jakielski KJ, Strand EA, Kloth K, Bierhals T; **DDD study**, Roberts JD, Petrovich RM, Machida S, Kurumizaka H, Lelieveld S, Pfundt R, Jansen S, Deriziotis P, Faivre L, Thevenon J, Assoum M, Shriberg L, Kleefstra T, Brunner HG, Wade PA, Fisher SE, Campeau PM. CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nat Commun. 2018 Nov 5;9(1):4619. Erratum in: Nat Commun. 2019 Feb 15;10(1):883. Erratum in: Nat Commun. 2019 May 2;10(1):2079.

13. Wright, C., et al, **DDD study**. Clinically-relevant postzygotic mosaicism in parents and children with developmental disorders in trio exome sequencing data. Nature Communications volume 10, Article number: 2985 (2019)

14. Snijders Blok L, Rousseau J, Twist J, Ehresmann S, Takaku M, Venselaar H, Rodan LH, Nowak CB, Douglas J, Swoboda KJ, Steeves MA, Sahai I, Stumpel CTRM, Stegmann APA, Wheeler P, Willing M, Fiala E, Kochhar A, Gibson WT, Cohen ASA, Agbahovbe R, Innes AM, Au PYB, Rankin J, Anderson IJ, Skinner SA, Louie RJ, Warren HE, Afenjar A, Keren B, Nava C, Buratti J, Isapof A, Rodriguez D, Lewandowski R, Propst J, van Essen T, Choi M, Lee S, Chae JH, Price S, Schnur RE, Douglas G, Wentzensen IM, Zweier C, Reis A, Bialer MG, Moore C, Koopmans M, Brilstra EH, Monroe GR, van Gassen KLI, van Binsbergen E, Newbury-Ecob R, Bownass L, Bader I, Mayr JA, Wortmann SB, Jakielski KJ, Strand EA, Kloth K, Bierhals T; **DDD study**, Roberts JD, Petrovich RM, Machida S, Kurumizaka H, Lelieveld S, Pfundt R, Jansen S, Deriziotis P, Faivre L, Thevenon J, Assoum M, Shriberg L, Kleefstra T, Brunner HG, Wade PA, Fisher SE, Campeau PM. CHD3 helicase domain mutations cause a neurodevelopmental syndrome with macrocephaly and impaired speech and language. Nat Commun. 2018 Nov 5;9(1):4619.

15. Faundes V, Newman WG, Bernardini L, Canham N, Clayton-Smith J, Dallapiccola B, Davies SJ, Demos MK, Goldman A, Gill H, Horton R, Kerr B, Kumar D, Lehman A, McKee S, Morton J, Parker MJ, Rankin J, Robertson L, Temple IK; Clinical Assessment of the Utility of Sequencing and Evaluation as a Service (CAUSES) Study; **Deciphering Developmental Disorders (DDD) Study**, Banka S. Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. Am J Hum Genet. 2018 Jan 4;102(1):175-187.

16. Alcantara D, Timms AE, Gripp K, Baker L, Park K, Collins S, Cheng C, Stewart F, Mehta SG, **Saggar A,** Sztriha L, Zombor M, Caluseriu O, Mesterman R, Van Allen MI, Jacquinet A, Ygberg S, Bernstein JA, Wenger AM, Guturu H, Bejerano G, Gomez- Ospina N, Lehman A, Alfei E, Pantaleoni C, Conti V, Guerrini R, Moog U, Graham JM Jr, Hevner R, Dobyns WB, O'Driscoll M, Mirzaa GM. Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain. 2017 Oct 1;140(10):2610-2622.

17. Sadleir LG, Mountier EI, Gill D, Davis S, Joshi C, DeVile C, Kurian MA; **DDD Study,** Mandelstam S, Wirrell E, Nickels KC, Murali HR, Carvill G, Myers CT, Mefford HC, Scheffer IE. Not all SCN1A epileptic encephalopathies are Dravet syndrome: Early profound Thr226Met phenotype. Neurology. 2017 Sep 5;89(10):1035-1042.

18. Wan Y., Simpson M., Aragon-Martin J., Osborn P., Regalado E., Guo D., Boileau C., Jondeau G., Benarroch L., Isekame Y., Bharj J., Sneddon J., Fisher E., Dean J., Tome Esteban MT, **Saggar A**., Milewicz D., Jahangiri M., Behr E., Smith A., Child A. A mutation in the LMOD1 actin-binding domain segregating with disease in a large British family with thoracic aortic aneurysms and dissections. bioRxiv 153452; 2017. doi: https://doi.org/10.1101/ 153452

19. Salpietro, V., Lin, W., Vedove, A.D., Storbeck, M., Liu, Y., Efthymiou, S., Manole, A., Wiethoff, S., Ye, Q., **Saggar, A**., McElreavey, K., Krishnakumar, S.S., Pitt, M., Bello, O.D., Rothman, J.E., Basel-Vanagaite, L., Hubshman, M.W., Aharoni, S., Manzur, A.Y., Wirth, B., Houlden, H. (2017) 'Homozygous mutations in VAMP1 cause a presynaptic congenital myasthenic syndrome', Annals of Neurology. John Wiley and Sons Inc., 81(4), pp. 597–603.

20. Sithambaram S, Bishop N, Shankar L, Offiah A, Pollitt R, Balasubramanian M, **Saggar A**, Arundel P. Osteogenesis imperfecta type VI presenting as suspected physical abuse -- a report of two cases. Bone Abstracts (2017) 6 P042

21. Christodoulou L, Krishnaiah A, Spyridou C, Salpietro V, Hannan S, **Saggar A**, Mankad K, Deep A, Kinali M. Kenny Caffey syndrome with severe respiratory and gastrointestinal involvement: expanding the clinical phenotype. Quant Imaging Med Surg. 2015 Jun;5(3):476-9.

22. **Deciphering Developmental Disorders Study**. Prevalence and architecture of de novo mutations in developmental disorders. Nature. 2017 Feb 23;542(7642):433-438.

23. Kharbanda M, Pilz DT, Tomkins S, Chandler K, **Saggar A**, Fryer A, McKay V, Louro P, Smith JC, Burn J, Kini U, De Burca A, FitzPatrick DR, Kinning E; DDD Study. Clinical features associated with CTNNB1 de novo loss of function mutations in ten individuals. Eur J Med Genet. 2017 Feb;60(2):130-135.

24. Pavlidou E, Ramachandran V, Govender V, Wilson C, Das R, Vlachou V, Pavlou E, **Saggar A**, Mankad K, Kinali M. A novel PLP1 mutation associated with optic nerve enlargement in two siblings with Pelizaeus-Merzbacher disease: A new MRI finding. Brain Dev. 2017 Mar;39(3):271-274.

25. Pavlidou E, Salpietro V, Phadke R, Hargreaves IP, Batten L, McElreavy K, Pitt M, Mankad K, Wilson C, Cutrupi MC, Ruggieri M, McCormick D, **Saggar A,** Kinali M. Pontocerebellar hypoplasia type 2D and optic nerve atrophy further expand the spectrum associated with selenoprotein biosynthesis deficiency. Eur J Paediatr Neurol. 2016 May;20(3):483-8.

26. Saunders EJ, Dadaev T, Leongamornlert DA, Al Olama AA, Benlloch S, Giles GG, Wiklund F, Gronberg H, Haiman CA, Schleutker J, Nordestgaard BG, Travis RC, Neal D, Pasayan N, Khaw KT, Stanford JL, Blot WJ, Thibodeau SN, Maier C, Kibel AS, Cybulski C, Cannon-Albright L, Brenner H, Park JY, Kaneva R, Batra J, Teixeira MR, Pandha H, Govindasami K, Muir K; **UK Genetic Prostate Cancer Study Collaborators**; UK ProtecT Study Collaborators; PRACTICAL Consortium, Easton DF, Eeles RA, Kote-Jarai Z. Gene and pathway level analyses of germline DNA-repair gene variants and prostate cancer susceptibility using the iCOGS-genotyping array. Br J Cancer. 2016 Apr 12;114(8):945-52.

27. Steel D, Salpietro V, Phadke R, Pitt M, Gentile G, Massoud A, Batten L, Bashamboo A, Mcelreavey K, **Saggar A,** Kinali M. Whole exome sequencing reveals a MLL de novo mutation associated with mild developmental delay and without 'hairy elbows': expanding the phenotype of Wiedemann-Steiner syndrome. J Genet. 2015 Dec;94(4):755-8.

28. Mircsof D, Langouët M, Rio M, Moutton S, Siquier-Pernet K, Bole-Feysot C, Cagnard N, Nitschke P, Gaspar L, Žnidarič M, Alibeu O, Fritz AK, Wolfer DP, Schröter A, Bosshard G, Rudin M, Koester C, Crestani F, Seebeck P, Boddaert N, Prescott K; **DDD Study**, Hines R, Moss SJ, Fritschy JM, Munnich A, Amiel J, Brown SA, Tyagarajan SK, Colleaux L. Mutations in NONO lead to syndromic intellectual disability and inhibitory synaptic defects. Nat Neurosci. 2015 Dec;18(12):1731-6.

29. Al Olama, AA., Dadaev T, Hazelett DJ, Li Q, Leongamornlert D, Saunders EJ, Stephens S, Cieza-Borrella C, Whitmore I, Benlloch Garcia S, Giles GG, Southey MC, Fitzgerald L, Gronberg H, Wiklund F, Aly M, Henderson BE, Schumacher F, Haiman CA, Schleutker J, Wahlfors T, Tammela TL, Nordestgaard BG, Key TJ, Travis RC, Neal DE, Donovan JL, Hamdy FC, Pharoah P, Pashayan N, Khaw KT, Stanford JL, Thibodeau SN, Mcdonnell SK, Schaid DJ, Maier C, Vogel W, Luedeke M, Herkommer K, Kibel AS, Cybulski C, Wokołorczyk D, Kluzniak W, Cannon-Albright L, Brenner H, Butterbach K, Arndt V, Park JY, Sellers T, Lin HY, Slavov C, Kaneva R, Mitev V, Batra J, Clements JA, Spurdle A, Teixeira MR, Paulo P, Maia S, Pandha H, Michael A, Kierzek A, Govindasami K, Guy M, Lophatonanon A, Muir K, Viñuela A, Brown AA; PRACTICAL Consortium; COGS-CRUK GWAS-ELLIPSE (Part of GAME-ON) Initiative; Australian Prostate Cancer BioResource; **UK Genetic Prostate Cancer Study Collaborators;** UK ProtecT Study Collaborators, Freedman M, Conti DV, Easton D, Coetzee GA, Eeles RA, Kote-Jarai Z. Multiple novel prostate cancer susceptibility signals identified by fine-mapping of known risk loci among Europeans. Hum Mol Genet. 2015 Oct 1;24(19):5589-602.

30. Salpietro V, Phadke R, **Saggar A**, Hargreaves IP, Yates R, Fokoloros C, Mankad K, Hertecant J, Ruggieri M, McCormick D, Kinali M. Zellweger syndrome and secondary mitochondrial myopathy. Eur J Pediatr. 2015 Apr;174(4):557-63.

31. Wright CF, Fitzgerald TW, Jones WD, Clayton S, McRae JF, van Kogelenberg M, King DA, Ambridge K, Barrett DM, Bayzetinova T, Bevan AP, Bragin E, Chatzimichali EA, Gribble S, Jones P, Krishnappa N, Mason LE, Miller R, Morley KI, Parthiban V, Prigmore E, Rajan D, Sifrim A, Swaminathan GJ, Tivey AR, Middleton A, Parker M, Carter NP, Barrett JC, Hurles ME, FitzPatrick DR, Firth HV; **DDD study**. Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data. Lancet. 2015 Apr 4;385(9975):1305-14.

32. **Deciphering Developmental Disorders Study**. Large-scale discovery of novel genetic causes of developmental disorders. Nature. Mar 12;519(7542):223-8. 2015.

33. Stuart HM, Roberts NA, Hilton EN, McKenzie EA, Daly SB, Hadfield KD, Rahal JS, Gardiner NJ, Tanley SW, Lewis MA, Sites E, Angle B, Alves C, LourenÃ§o T, Rodrigues M, Calado A, Amado M, Guerreiro N, Serras I, Beetz C, Varga RE, Silay MS, Darlow JM, Dobson MG, Barton DE, Hunziker M, Puri P, Feather SA, Goodship JA, Goodship TH, Lambert HJ, Cordell HJ; UK VUR Study Group, **Saggar A**, Kinali M; 4C Study Group, Lorenz C, Moeller K, Schaefer F, Bayazit AK, Weber S, Newman WG, Woolf AS. Urinary Tract Effects of HPSE2 Mutations. J Am Soc Nephrol. Apr;26(4):797-804. 2015

34. McGovern AP, Jones S, van Vlymen J, **Saggar AK**, Sandford R, de Lusignan S. Identification of people with autosomal dominant polycystic kidney disease using routine data: a cross sectional study. BMC Nephrol. Nov 20;15:182. 2014

35. Martin CA, Ahmad I, Klingseisen A, Hussain MS, Bicknell LS, Leitch A, Naeumnberg G, Toliat MR, Murray JE, Hunt D, Khan F, Ali Z, Tinschert S, Ding J, Keith C, Harley ME, Heyn P, Mueller R, Hoffmann I, Daire VC, Dollfus H, Dupuis L, Bashamboo A, McElreavey K, Kariminejad A, Mendoza-Londono R, Moore AT, **Saggar A**, Schlechter C, Weleber R, Thiele H, Altmeuller J, Haehne W, Hurles ME, Noegel AA, Baig SM, Neurnberg P, Jackson AP. Mutations in PLK4, encoding a master regulator of centriole biogenesis, cause microcephaly, growth failure and retinopathy. Nat Genet. Dec;46(12):1283-92. 2014

36. Humphrey A, MacLean C, Ploubidis GB, Granader Y, Clifford M, Haslop M, Neville BG, Yates JR, Bolton PF; **Tuberous Sclerosis 2000 Study Group**. Intellectual development before and after the onset of infantile spasms: a controlled prospective longitudinal study in tuberous sclerosis. Epilepsia. Jan;55(1):108-16. 2014

37. Poole, RL., Docherty, LE., Al Sayegh, A., Caliebe, A., Turner, C., Baple, E., Wakeling, E., Harrison, L., Lehmann, A., Temple, IK., Mackay, DJ., International Clinical Imprinting Consortium., **Saggar, A.,** et al. Targeted methylation testing of a patient cohort broadens the epigenetic and clinical description or imprinting disorders. J Med Genet, 2013.

38. Tatton-Brown K, Murray A, Hanks S, Douglas J, Armstrong R, Banka S, Bird LM, Clericuzio CL, Cormier-Daire V, Cushing T, Flinter F, Jacquemont ML, Joss S, Kinning E, Lynch SA, Magee A, McConnell V, Medeira A, Ozono K, Patton M, Rankin J, Shears D, Simon M, Splitt M, Strenger V, Stuurman K, Taylor C, Titheradge H, Van Maldergem L, Temple IK, Cole T, Seal S; **Childhood Overgrowth Consortium**, Rahman N. Weaver syndrome and EZH2 mutations: Clarifying the clinical phenotype. Am J Med Genet A. Dec;161A(12):2972-80. 2013

39. Loup O, Daubeney PE, **Saggar A**, Rubens M, Naqvi N, Ghez O. Severe arterial tortuosity in an asymptomatic infant with coarctation. Circ Cardiovasc Imaging. May 1;6(3):487-90. 2013

40. Reinstein E, Frentz S, Morgan T, Garcia S, Leventer RJ, McGillivray G, Pariani M, van der Steen A, Pope M, Holder-Espinasse M, Scott R, Thompson EM, Robertson T, Coppin B, Siegel R, Bret Zurita M, Rodriques JI, Morales C, Rodrigues Y, Arcas J, **Saggar A**, Horton M, Zackai E, Graham JM, Rimoin DL, Robertson SP. Vascular and connective tissue anomalies associated with X-linked periventricular heterotopia due to mutations in Filamin A. Eur J Hum Genet. May;21(5):494-502. 2013

41. Riviere, JB., Mirzaa, GM., O'Roak, BJ., Beddaoui, M., Alcantara, D., Conway, RL., St-Onge, J., Schwartzentruber, JA., Gripp, KW., Nikkel, SM., Worthylake, T., Sullivan, CT., Ward, TR., Butler, HE., Kramer, NA., Albrecht, B., Armour, CM., Armstrong, L., Caluseriu, O., Cytrynbaum, C., Drolet, BA., Innes, AM., Lauzon, JL., Lin AE., Mancini, GM., Meschino, WS., Reggin, JD., **Saggar, AK.**, et al. De novo germline and postzygotic mutations in AKT3, PIK3R2 and PIK3CA cause a spectrum of related Megalencephaly syndromes. Nat Genet, 2012.

42. Jones WD, Dafou D, McEntagart M, Woollard WJ, Elmslie FV, Holder-Espinasse M, Irving M, **Saggar AK**, Smithson S, Trembath RC, Deshpande C, Simpson MA. De novo mutations in MLL cause Wiedemann-Steiner syndrome. Am J Hum Genet. Aug 10;91(2):358-64. 2012

43. Injarie AM, Narang A, Idrees Z, **Saggar AK**, Nischal KK. Ocular treatment of children with Stuve-Wiedemann syndrome. Cornea. Mar;31(3):269-72. 2012

44. Tatton-Brown K, Hanks S, Ruark E, Zachariou A, Duarte Sdel V, Ramsay E, Snape K, Murray A, Perdeaux ER, Seal S, Loveday C, Banka S, Clericuzio C, Flinter F, Magee A, McConnell V, Patton M, Raith W, Rankin J, Splitt M, Strenger V, Taylor C, Wheeler P, Temple KI, Cole T; **Childhood Overgrowth Collaboration**, Douglas J, Rahman N. Germline mutations in the oncogene EZH2 cause Weaver syndrome and increased human height. Oncotarget. Dec;2(12):1127-33. 2011

45. Jones, W., Saggar, A.K., Howe, F., Rich, P., Al-Memar, A. (2011) 'PO.03 A novel OPA1 mutation is associated with a multiple sclerosis-like illness in two sisters', Journal of Neurology, Neurosurgery & Psychiatry. BMJ, 82(9), pp. e4–e4.

46. Hachach-Haram N, Gerarchi P, Benyon SL, **Saggar A**, McLellan G, Kirkpatrick WN. Multidisciplinary surgical management of cherubism complicated by neurofibromatosis type 1. J Craniofac Surg. Nov;22(6):2318-22. 2011

47. Laycock-van Spyk S, Jim HP, Thomas L, Spurlock G, Fares L, Palmer-Smith S, Kini U, **Saggar A,** Patton M, Mautner V, Pilz DT, Upadhyaya M. Identification of five novel SPRED1 germline mutations in Legius syndrome. Clin Genet. Jul;80(1):93-6. 2011

48. Dunlop EA, Dodd KM, Land SC, Davies PA, Martins N, Stuart H, McKee S, Kingswood C, **Saggar A**, Corderio I, Medeira AM, Kingston H, Sampson JR, Davies DM, Tee AR. Determining the pathogenicity of patient-derived TSC2 mutations by functional characterization and clinical evidence. Eur J Hum Genet. Jul;19(7):789-95. 2011

49. Slade I, Bacchelli C, Davies H, Murray A, Abbaszadeh F, Hanks S, Barfoot R, Burke A, Chisholm J, Hewitt M, Jenkinson H, King D, Morland B, Pizer B, Prescott K, **Saggar A**, Side L, Traunecker H, Vaidya S, Ward P, Futreal PA, Vujanic G, Nicholson AG, Sebire N, Turnbull C, Priest JR, Pritchard-Jones K, Houlston R, Stiller C, Stratton MR, Douglas J, Rahman N. DICER1 syndrome: clarifying the diagnosis, clinical features and management implications of a pleiotropic tumour predisposition syndrome. J Med Genet. Apr;48(4):273-8. 2011

50. Aragon-Martin, J.A., D. Ahnood, D.G. Charteris, **A. Saggar**, et al. Role of ADAMTSL4 mutations in FBN1 mutation-negative ectopia lentis patients. Hum Mutat, 31(8) 2010

51. Matejas V, Hinkes B, Alkandari F, Al-Gazali L, Annexstad E, Aytac MB, Barrow M, Bláhová K, Bockenhauer D, Cheong HI, Maruniak-Chudek I, Cochat P, Dötsch J, Gajjar P, Hennekam RC, Janssen F, Kagan M, Kariminejad A, Kemper MJ, Koenig J, Kogan J, Kroes HY, Kuwertz-Bröking E, Lewanda AF, Medeira A, Muscheites J, Niaudet P, Pierson M, **Saggar A**, Seaver L, Suri M, Tsygin A, Wühl E, Zurowska A, Uebe S, Hildebrandt F, Antignac C, Zenker M. Mutations in the human laminin beta2 (LAMB2) gene and the associated phenotypic spectrum. Hum Mutat. 2010 Sep;31(9):992-1002.

52. Alazami, A.M., Shaheen, R., Alzahrani, F., Snape, K., Saggar, A., Brinkmann, B., Bavi, P., Al-Gazali, L.I., Alkuraya, F.S. (2009) 'FREM1 Mutations Cause Bifid Nose, Renal Agenesis, and Anorectal Malformations Syndrome', American Journal of Human Genetics. Cell Press, 85(3), pp. 414–418.

53. Coccia, M., Brooks, S.P., Webb, T.R., Christodoulou, K., Wozniak, I.O., Murday, V., Balicki, M., Yee, H.A., Wangensteen, T., Riise, R., **Saggar, A.K.**, Park, S.-M., Kanuga, N., Francis, P.J., Maher, E.R., Moore, A.T., Russell-Eggitt, I.M., Hardcastle, A.J. (2009) 'X-linked cataract and Nance-Horan syndrome are allelic disorders.', Human molecular genetics, 18(14), pp. 2643–55.

54. Eeles RA, Kote-Jarai Z, Al Olama AA, Giles GG, Guy M, Severi G, Muir K, Hopper JL, Henderson BE, Haiman CA, Schleutker J, Hamdy FC, Neal DE, Donovan JL, Stanford JL, Ostrander EA, Ingles SA, John EM, Thibodeau SN, Schaid D, Park JY, Spurdle A, Clements J, Dickinson JL, Maier C, Vogel W, Dörk T, Rebbeck TR, Cooney KA, Cannon-Albright L, Chappuis PO, Hutter P, Zeegers M, Kaneva R, Zhang HW, Lu YJ, Foulkes WD, English DR, Leongamornlert DA, Tymrakiewicz M, Morrison J, Ardern-Jones AT, Hall AL, O'Brien LT, Wilkinson RA, Saunders EJ, Page EC, Sawyer EJ, Edwards SM, Dearnaley DP, Horwich A, Huddart RA, Khoo VS, Parker CC, Van As N, Woodhouse CJ, Thompson A, Christmas T, Ogden C, Cooper CS, Southey MC, Lophatananon A, Liu JF, Kolonel LN, Le Marchand L, Wahlfors T, Tammela TL, Auvinen A, Lewis SJ, Cox A, FitzGerald LM, Koopmeiners JS, Karyadi DM, Kwon EM, Stern MC, Corral R, Joshi AD, Shahabi A, McDonnell SK, Sellers TA, Pow-Sang J, Chambers S, Aitken J, Gardiner RA, Batra J, Kedda MA, Lose F, Polanowski A, Patterson B, Serth J, Meyer A, Luedeke M, Stefflova K, Ray AM, Lange EM, Farnham J, Khan H, Slavov C, Mitkova A, Cao G; UK Genetic **Prostate Cancer Study Collaborators**/British Association of Urological Surgeons' Section of Oncology; UK ProtecT Study Collaborators; PRACTICAL Consortium, Easton DF. Identification of seven new prostate cancer susceptibility loci through a genome-wide association study. Nat Genet. 2009 Oct;41(10):1116-21.

55. Prasad, S., Sharma, S., Petros, A., Daubeney, P., Nischal, K., Connell, J., Rich, P., **Saggar-Malik, A**. (2008) 'Molybdenum Cofactor Deficiency in a Middle Eastern Child: Neuroradiology, Cardiomyopathy and Extension of Phenotype', in Conference: British Human Genetics Conference. Journal of Medical Genetics.

56. Griffith, E., Walker, S., Martin, C.A., Vagnarelli, P., Stiff, T., Vernay, B., Sanna, N. Al, **Saggar, A.**, Hamel, B., Earnshaw, W.C., Jeggo, P.A., Jackson, A.P., O'Driscoll, M. (2008) 'Mutations in pericentrin cause Seckel syndrome with defective ATR-dependent DNA damage signaling', Nature Genetics, 40(2), pp. 232–236.

57. Bredrup, C., Matejas, V., Barrow, M., Bláhová, K., Bockenhauer, D., Fowler, D.J., Gregson, R.M., Maruniak-Chudek, I., Medeira, A., Mendonça, E.L., Kagan, M., Koenig, J., Krastel, H., Kroes, H.Y., **Saggar, A.**, Sawyer, T., Schittkowski, M., Świetliński, J., Thompson, D., VanDeVoorde, R.G., Wittebol-Post, D., Woodruff, G., Zurowska, A., Hennekam, R.C., Zenker, M. (2008) 'Ophthalmological Aspects of Pierson Syndrome', American Journal of Ophthalmology. Elsevier Inc., 146(4), pp. 602–611.

58. **Saggar, A.K.** and Bittles, A. H. (2008) 'Consanguinity and child health', Paediatrics and Child Health. Elsevier, 18(5), pp. 244–249.

59. Davies, D.M., Johnson, S.R., Tattersfield, A.E., Kingswood, J.C., Cox, J.A., McCartney, D.L., Doyle, T., Elmslie, F., **Saggar, A.**, De Vries, P.J., Sampson, J.R. (2008) 'Sirolimus therapy in tuberous sclerosis or sporadic lymphangioleiomyomatosis', New England Journal of Medicine. Massachussetts Medical Society, pp. 200–203.

60. Doulton TW, **Saggar-Malik AK**, He FJ, Carney C, Markandu ND, Sagnella GA, MacGregor GA. The effect of sodium and angiotensin-converting enzyme inhibition on the classic circulating renin-angiotensin system in autosomal-dominant polycystic kidney disease patients. J Hypertens. 2006 May; 24(5):939-45.0

61. Mehta P, **Saggar, AK**. Ethnicity, equity, and access to genetic services--the UK perspective. Ann Hum Biol. 2005; 32(2):204-10.

62. Patel U, Simpson E, Kingswood JC, **Saggar-Malik AK**. Tuberose sclerosis complex: analysis of growth rates aids differentiation of renal cell carcinoma from atypical or minimal-fat-containing angiomyolipoma. Clin Radiol. 2005 Jun;60(6):665-73; discussion 663-4.

63. Whittock NV, Izatt L, Mann A, Homfray T, Bennett C, Mansour S, Hurst J, Fryer A, **Saggar A**, Barwell JG, Ellard S, Clayton PT. Novel mutations in X-linked dominant chondrodysplasia punctata (CDPX2). The Soc for Investig Derm. 2003, October, 121 (4) 939 – 942.

64. Rossetti S, Chauveau D, Kubly V, Slezak JM, **Saggar-Malik AK**, Pei Y, Ong AC, Stewart F, Watson ML, Bergstralh EJ, Winearls, CG, Torres VE, Harris PC. Association of mutation position in polycystic kidney disease 1 (PKD1) gene and development of a vascular phenotype. Lancet 2003 June; 361 (9376) 2196-201.

65. Dong YB, Plange-Rhule J Owusu I, Micah F, Eastwood JB, Carter ND, **Saggar-Malik AK**, Cappuccio FP, Jeffery S. T594M mutation of the beta-subunit of the epithelial sodium channel in Ghanaian populations from Kumasi and London and a possible association with hypertension. Genet Test. 2002 Spring; 6(1):63-65.

66. Rosseti S, Chauveau D, Walker D **Saggar-Malik AK**, Winearls CG, Torres VE, Harris PC. A complete mutation screen of the ADPKD genes by DHPLC. Kidney Int. 2002 May;61(5):1588-1599.

67. Afzal AR, Florencio RN, Taylor R, Patton MA, **Saggar-Malik AK**, Jeffery S. Novel mutations in the duplicated region of the polycystic kidney disease 1 (PKD1) gene provides supporting evidence for gene conversion. Genet Test. 2000;4(4):365-370.

68. **Saggar-Malik AK**, Afzal AR, Swissman JS, Bland M, Sagnella GA, Eastwood JB, MacGregor GA, Jeffery S. Lack of association of ACE/angiotensinogen genotype with renal function in autosomal dominant polycystic kidney disease. Genet Test. 2000; 4(3):299-303.

69. Hateboer N, Torra R, Estivill E, Bogdanova N, Davies F, Lazarou L, v Dijk M, Breuning M, **Saggar-Malik A**, Jeffery S, San Millan J L, Martinez I, Walker R, Holmans P, Ravine D. Location of mutations within the PKD2 gene influences outcome. Kidney International, 2000. 57, 1444-1451

70. Jeffery S Crosby A Plange-Rhule, J., Amoah-Danquah J., Acheampong JW., Eastwood J., **Saggar-Malik AK**. Evidence from a Ghanian population of known African descent to support the proposition that haemochromatosis is a Caucasian disorder. Genetic testing 1999 3, 375-378

71. Jeffery S., **Saggar-Malik AK.**, Crosby A., Bland M., Eastwood J., Amoah-Danquah J., Acheampong JW., Plange-Rhule, J. A dominant relationship between the ACE-D allele and serum ACE levels in a Ghanaian population J.Med Genet 1999. 36 869-870.

72. Afzal. A. R., Hand, M., Ternes-Pereira, E., **Saggar-Malik, AK**., Taylor, R., Jeffery, S. Novel Mutations in the 3' region of the polycystic kidney disease 1 (PKD1) gene. Human Mutation. 1999. 105, 648-653.

73. Jeffery, S., Crosby, A., Plange-Rhule, J., Amoah-Danquah, J., Acheampong, J.W., Eastwood, J.B., **Saggar-Malik, A.K**. (1999) 'Evidence from a Ghanaian population of known African descent to support the proposition that hemochromatosis is a Caucasian disorder', Genetic Testing. Mary Ann Liebert Inc., 3(4), pp. 375–377.

74. Plange-Rhule, J, Phillips, R, Acheampong, JW, **Saggar-Malik, AK**, Eastwood. J, Cappuccio, FP. Hypertension and renal failure in Kumasi, Ghana. 1999. J Human Hypertension, 13, 37-40

75. Hateboer, N, v Dijk M, Bogdanova N, Coto E, Jeffery S, **Saggar-Malik AK**, San Milan J L, Torra R, Breuning M, Holmans P, Ravine D. Polycystic Kidney Disease types 1 and 2:a comparison of phenotypes. Lancet 9 January 1999

76. Jeffery S, Economides DL., **Saggar-Malik AK**, MacDermott KD, Apparent normalisation of fetal renal size in autosomal dominant polycystic kidney disease. Clinical Genetics 1998. 53,303-307.

77. Sagnella, GA, **Saggar-Malik, AK**, Buckley, MG, Markandu, ND, Eastwood, JB, MacGregor, GA. Association between atrial natriuretic peptide and cyclic GMP in hypertension and in chronic renal failure. Clinica Chemica Acta 275, 9-18, 1998

78. MacDermott KD, **Saggar-Malik AK**, Economides DL, Jeffery S. Prenatal diagnosis for autosomal dominant polycystic kidney disease (PKD1) presenting in utero and prognosis for very early onset disease J Med Genet 35 1 13-16, 1998

79. Weston, BS, Jeffery, S, Jeffrey I, Sharaf, FA, Carter N, **Saggar-Malik AK**, Price, RG. Polycystin expression during embryonic development of human kidney in adult tissues and ADPKD tissue. Histochem J 29, 847-856. 1997

80. **Saggar-Malik AK**, Markandu, ND, MacGregor, GA, Cappuccio FP. Case report: Moderate salt restriction for the management of hypertension and hypercalciuria. J Human Hypertens 10, 811-813. 1996

81. **Saggar-Malik AK**. Making the change. A guide to changing antihypertensive therapy in the elderly. (Editorial) Geriatrics, 10, 1996

82. S**aggar-Malik, A.K**. and Patton, M. A. (1997) 'Original contributions by medical students: Increased fees may discourage research', BMJ Clinical Research. BMJ, 314(7087).

83. Buckley MG, **Saggar-Malik AK**, Markandu ND, Sagnella GA, MacGregor GA. Effects of changes in dietary sodium intake on plasma brain natriuretic peptide levels in patients with autosomal dominant polycystic kidney disease. J Hum Hyperten, 10, 1996

84. Jeffery S, Morgan S, WarmingtonVJ, MacGregor GA, **Saggar-Malik AK**. A family with autosomal dominant polycystic kidney disease not linked to 4q21-23 J Med Genet 1995, 32 493-94

85. Bath PMW, **Saggar-Malik AK**, MacDougall I, Eastwood JB, MacGregor1995. Increased platelet volume in autosomal dominant polycystic kidney disease Platelets 1995 6 (6): 336-339

86. Jeffery, S, **Saggar-Malik AK**, Morgan, S, Eastwood, JB, Patton, M. Genetic analysis of 20 families with autosomal dominant polycystic kidney disease from the South West Thames Region. Clinical Genetics, 1995; 77, (6) 290-4

87. Mahieu, I., **Saggar-Malik,** A., Hollande, E., Carter, N. (1995) 'Localisation and characterisation of carbonic anhydrase isozymes (CA I, CA II, CA III and CA IV) in an umbilical vein endothelial cell line (EA-hy926)', Biochemical Society Transactions, 23(2).

88. Sagnella, G., **Saggar-Malik,** A., Markandu, N., Buckley, M., Eastwood, J., MacGregor, G. (1994) 'Anp, Cyclic Gmp and Urinary Sodium Excretion in Chronic Renal Failure', Clinical Science. Portland Press Ltd., 86(s30), pp. 29P-29P.

89. **Saggar-Malik, A.K.**, Cappuccio, F.P. (1994) 'Potassium Supplements and Potassium-Sparing Diuretics: A Review and Guide to Appropriate Use', Drugs, 46(6), pp. 986–1008.

90. Buckenham T, **Saggar-Malik AK**. Alcohol for parenchymal cysts. Australasian Radiology 1994 38: 85-

91. **Saggar-Malik, A.K.,** Jeffery, S. and Patton, M. A. (1994) 'Autosomal dominant polycystic kidney disease', BMJ Clinical Research, 308(6938), p. 1184.

92. **Saggar-Malik, A.K.,** Missouris, C.G., Gill, J.S., Singer, D.R.J., Markandu, N.D., Macgregor, G.A. (1995) 'Left ventricular mass in normotensive subjects with autosomal dominant polycystic kidney disease', BMJ Clinical Research, 309(6969), p. 1617.

93. Jeffery, S., **Saggar-Malik AK**, Morgan, S. and MacGregor, G. A. A family with autosomal dominant polycystic kidney disease not linked to chromosome 16p 13.3. Clinical Genetics, 44: 173-176 1994

94. **Saggar-Malik, A. K.**, Missouris, C.G., Gill, J., Markandu, N.D., Singer, D.R.J., Leech, G., Johnston, D., MacGregor, G. (1993) '181 Unexplained left ventricular hypertrophy in normotensive patients with autosomal dominant polycystic kidney disease', Journal of Hypertension. Ovid Technologies (Wolters Kluwer Health), 11(5), p. S477.

95. Stenvinkel P, **Saggar-Malik A.K**. Alvestrand A. Renal haemodynamics and tubular sodium handling following volume expansion with sodium chloride (NaCl) and glucose in healthy humans. Scand J Lab Invest 52: 837-846, 1992

96. Stenvinkel P, **Saggar-Malik AK**, Wahrenberg H, Diczfalusy U, Bolinder J, Alvestrand A Impaired intrarenal dopamine production following intravenous sodium chloride infusion in insulin-dependent (Type 1) diabetes mellitus. Diabetologia, 34, 114-118, 1991

97. Cundy TF Butler J, Pope RM, **Saggar-Malik AK**, Wheeler MJ, Williams R Amenorrhoea in Women with Non-alcoholic Chronic Liver Disease. Gut 32, 2 p 202-206. 1991

BOOK CHAPTERS

1. **Saggar A**. Inherited Renal diseases. In: Horizons in Medicine 17. Updates in major medical clinical advances. Ed Dorian Haskard. Royal College of Physicians. London, 2005
2. **Saggar Malik AK**, Somlo S. Autosomal dominant polycystic kidney disease. In: The Genetics of Renal Disease. Eds. Flinter F, Maher E, Saggar-Malik AK. Oxford University Press, Oxford.2003.
3. **Saggar Malik AK**. Cystic kidney disease. In: The Genetics of Renal Disease. Eds. Flinter F, Maher E, Saggar-Malik AK. Oxford University Press, Oxford. 2003.
4. **Saggar Malik AK**. Gene therapy in renal disease. In: The Genetics of Renal Disease. Eds. Flinter F, Maher E, Saggar-Malik AK. Oxford University Press, Oxford. 2003.
5. **Saggar Malik AK** and Jeffery S. Gene structure and regulation and laboratory techniques. In Inherited disorders of the kidney, Ed Morgan SA and Gruenfeld JP. Oxford University Press, Oxford. 1998
6. **Saggar-Malik, AK,** Pettit K, Autosomal dominant polycystic kidney disease: An information booklet for patients and their families. St George's Medical School, London. 1996.
7. **Saggar-Malik, AK.** Cardiovascular risk factors in the ethnic minorities. (Bilingual Booklet). Wandsworth Borough Council, London. 1993
8. **Saggar-Malik, AK.** Cappucino, FP. Potassium supplements and potassium sparing diuretics. A review and guide to appropriate use. Drugs, 46, (6), 986-1008. 1993

EDITED BOOKS

1 Flinter F, Maher E , **Saggar-Malik AK**. The Genetics of Renal Disease, Oxford monographs on medical genetics, 48. Oxford University Press, Oxford.2003.