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Anthony Martin Vandersteen

CV, August 2019

Medical Practice:

2010 - 2014	Consultant Clinical Genetics, North West Thames Regional Genetics Service (0.6), Ehlers-Danlos Syndrome UK National Diagnostic Service (0.4), North West London Hospitals NHS Trust, Harrow, Middlesex, UK.
2014 - Present	Staff Physician, Medical Genetics, IWK Health Centre, Halifax, Nova Scotia, Assistant Professor, Dept Pediatrics, Dalhousie University. Queen Elizabeth II Health Sciences Centre, Halifax, Nova Scotia.

Specialist Certification in Internal Medicine:

2005 - 2015	Membership of Royal College of Physicians of London
2015 – Present	**Fellow of Royal College of Physicians of London**

Specialist Certification in Clinical Genetics:

2010 - Present	UK (GMC) Specialist Registration, Clinical Genetics.
2014 – 2016	Associate member Canadian College of Medical Genetics
2016 – Present	**Fellow of Royal College of Physicians and Surgeons of Canada (passed specialty exam July 2016)**
2016 – Present	Fellow of Canadian College of Medical Genetics

Licensure:

2002 - 2003	General Medical Council UK (GMC) Provisional Registration.
2003 - Present	UK (GMC) Full Registration.
2008	Medical Council of New Zealand Temporary Registration.
2010 - Present	UK (GMC) Specialist Registration, Clinical Genetics.
2014 - 2016	College of Physicians and Surgeons of Nova Scotia (CPSNS) Defined license.
2016 – Present	**CPSNS Full license (Medical Genetics).**
2016 - Present	College of Physicians of Prince Edward Island, visiting physician.
2014 - Present	College of Physicians of New Brunswick, telehealth license.
2018 - Present	College of Physicians of New Brunswick, visiting physician.

USMLE:

2002-2003	Passed steps 1, 2, clinical skills exam, ECFMG certification 2004.

Education:

1988 - 1991	MA (Hons), Natural Sciences, Cambridge University, Jesus College, II.2.
1991 - 1995	Doctor of Philosophy, Chemistry, King's College, London University. "Studies in the Chemical synthesis of oligo- and poly-ribonucleotides." Supervisor: Professor Colin Reese FRS. (Daniell Professor).
1997 - 2001	Bachelor of Medicine. University of Southampton, UK.

Graduate Training:

2002 - 2003	Internship: General and Respiratory Medicine, Southampton University Hospital. General Surgery, Salisbury District Hospital.
2003-2005	Residency: Internal Medicine (Emergency, Hematology, General medicine, Elderly Care, Intensive Care, Expedition Medicine, Oncology, Clinical Genetics), Wessex, Eastbourne and London Hospitals.
2006 - 2010	Clinical Genetics Specialist Training, London Deanery, UK. Guy's Hospital (2006-2007), Great Ormond Street Hospitals (2008-2010), Wellington Hospital, New Zealand (6 month locum 2008).

Academic Appointments:

1995 -1997	Post-doctoral Research Fellowship: The Scripps Research Institute, San Diego. The synthesis of small molecule and peptide libraries using combinatorial chemistry to investigate small molecule enzyme mimics. Development of novel Poly ethylene glycol (PEG) supported reagents for chemical synthesis. Assays of antibodies for catalysis. Supervisor: Professor KD Janda (Callaway Professor).
Jul 1997 - Aug 1997	Post-doctoral research: Hammersmith Hospital MRC Lab; Amyloid Peptide Synthesis in the core laboratories. Prof. Ted Tuddenham FMedSci.
Jul 1999 - Aug 1999	Post-doctoral research Fellow Tenovus; Southampton, *In-vitro* and murine studies of Iodine^{131} labelled anti-CD20 and anti-CD40 antibodies for lymphoma treatment. Professor Tim Illidge.

Nov 2004 - Dec 2004	Molecular Genetics Research Fellow/Honorary Contract, Southampton University Hospitals. Genotyping the Hertfordshire cohort for ghrelin polymorphisms, Prof. Ian Day.
2010 - 2014	Honorary Senior Lecturer, Human Genetics, Imperial College London University, Human Genetics faculty.
2014 - 2019	Assistant Professor, Department of Pediatrics, Faculty of Medicine, Dalhousie University.
July 2019 - Present	**Associate Professor, Department of Pediatrics, Faculty of Medicine, Dalhousie University.**
Jan 2019 – July 2019	Senior Research Fellow, Institute for Molecular Medicine, University of Edinburgh, (0.2 FTE), Exome Sequencing for Ehlers-Danlos Syndromes.

Honors and Awards:

1987	School chemistry prize (King's College School Wimbledon).
2000	David Millar memorial prize, Medical School, Competitive Essay: "Human Cloning".
	Zeneca prize in Pharmacology and Therapeutics.
2008	Distinction: Human Genetics degree module (Biol345) Macquarie University, Australia. This is mandatory training for Australasian trainees in medical genetics.
2009	Bethan Nancy Lecture Series, Great Ormond Street Hospital,
	First prize: "Mutations, Connotations, Implications".

Other Employment

Aug 1991	Cambridge Cyrenians, residential care worker for ex-homeless.
Jul - Aug 1989	Arthur Andersen & Co. Accountancy/ Tax internship. 2 months
Sep 1987 - Mar 1988	Industrial Society award scheme (INDEX) Shell Exploration.

RESEARCH

Publications:

1. **Vandersteen A. Studies in the Chemical Synthesis of Oligo-and Poly-ribonucleotides 1995**. PhD Thesis, University of London.

2. **Vandersteen A** and Janda K, **An investigation of two linear penta-peptides claimed to be serine protease mimics**. *J Am Chem Soc* **1996** (118) 8787-8789.

3. **Vandersteen A,** Han H, and Janda K, **Liquid phase Combinatorial synthesis, In search of serine protease enzyme mimics**. *Mol Divers* **1996** (2) 89-96.

4. Wentworth P, **Vandersteen A** and Janda K, **Polyethylene glycol (PEG) as a reagent support: The preparation and utility of PEG-tri-arylphosphine conjugate in liquid phase organic synthesis.** *J Chem Soc Chem Comm* **1997** 759-760.

5. Berg T, **Vandersteen A** and Janda K, **High-throughput synthesis and direct screening for the discovery of novel hydrolytic metal complexes** *Bioorg Med Chem Lett* **1998** 1221-1224.

6. Lloyd W, Reese C, Song Q, **Vandersteen A**, Visintin C and Zhang P-Z. *J. Chem Soc. Perkin Trans. 1* **2000** 165-176.
Thesis publication.

7. Illidge T, Honeychurch J, **Vandersteen A**, & Cragg M. **Radio-immunotherapy in the π-bcl-1 B-cell lymphoma model: efficacy depends more than on targeted irradiation alone** *Cancer Biother Radiopharm* **2000** (15) 581-591

8. Walter E, Gibbins N, **Vandersteen A,** Kinton L, Wark P, Jonas M. **Hyperkalaemic Ascending Paralysis**. *J R Soc Med* **2004** (97) 330-331.

9. **Vandersteen A**, Moore D, Donaghue C, MacFarlane N, Josifova D**. Genetic Diagnosis in Clinical Psychiatry: A Case report of a woman with a 47,XXX Karyotype and Fragile X syndrome** *Eur. J. Psychiatry*. **2009** (23) 31-36.

10. **Vandersteen A**, Turnbull J, Jan W, Simpson J, Lucas S, Anderson D, Lin J-P, Stratakis C, Pichert G , Lim M. **Cutaneous signs are important in the diagnosis of the rare neoplasia syndrome Carney complex**. *Eur. J. Pediatr* **2009** (168)11 1401**.**

11. **Vandersteen A** and Hennekam R, **Two Male Siblings with Mental Retardation, Dysmorphic Facial Features, Unusual Skull Shape, Premature Balding, Small Genitalia and Subluxed Patellae.** *Eur. J. Med. Genet.* **2010** (53) 314-317.

11 Rohrbach M, **Vandersteen A**, Yiş U, Serdaroglu G, Ataman E, Chopra M, Garcia S, Jones K, Kariminejad A, Kraenzlin M, Marcelis C, Baumgartner M, Giunta C. **Phenotypic variability of the kyphoscoliotic type of Ehlers-Danlos syndrome (EDS VIA): clinical, molecular and biochemical delineation.** *Orphanet J Rare Dis* **2011 (**6) 46.

12 **Vandersteen A** and Dixon J. **Adams-Oliver Syndrome, a family with dominant inheritance and a severe phenotype.** *Clin Dysmorph* **2011** 20(4) 210-3.

13 Lestner J, Chong K, Offiiah A, Kefas J, **Vandersteen A. Unusual Neuroradiological Features in Schinzel-Giedion Syndrome: A Novel Case**. *Clin Dysmorph* **2012** 21(3) 152-4.

14 Reinstein E, Frentz S, Morgan T, García-Miñaúr S, Leventer R, McGillivray G, Pariani M, **Vandersteen A,** Pope M, Holder-Espinasse M, Scott R, Thompson EM, Robertson T, Coppin B, Siegel R, Bret Zurita M, Rodríguez JI, Morales C, Rodrigues Y, Arcas J, Saggar A, Horton M, Zackai E, Graham JM, Rimoin D, Robertson S. **Vascular and connective tissue anomalies**

associated with X-linked periventricular heterotopia due to mutations in Filamin A. *Eur J Hum Genet.* **2012** 21(5)494-502.

15 **Vandersteen A,** Kenny J, Khan NL, Male A. **Marfan syndrome presenting with headache and coincidental ophthalmic artery aneurysm**. *BMJ Case Rep* **2013** Mar 15;2013

16 **Vandersteen A**, Lund A, Ferguson D, Sawle P, Pollitt R, Holder S, Wakeling E, Moat N, Pope M. **Four patients with Sillence type I Osteogenesis Imperfecta and mild bone fragility, complicated by left ventricular cardiac valvular disease and cardiac tissue fragility caused by type I collagen mutations.** *Am J Med Genet*, **2013** 164A(2):386-91

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18 Jones K, Choong A, Canham N, Renton S, Pollitt R, Nesbitt M, Kopcke D, Islam L, Buckley J, Ghali N, **Vandersteen A. A combined vascular surgical and clinical genetics approach to diffuse aneurysmal disease.** *Ann R Coll Surg Engl.* **2015** 7(5):e73-6

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23 George S, **Vandersteen A,** Nigar E, Ferguson D, Topham E, Pope M. **Two Ehlers Danlos syndrome type VIII patients with unexpected hoarseness** *Clin Exp Dermatol.* **2016** (7):771-4

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

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38 Zawerton A, Mignot C, Sigafoos A, Blackbur PR, Haseeb A, McWalter K, Ichikawa S, Nava C, Keren B, Charles P, Marey I, Tabet A-C, Perrin L, Hartmann A, Lesca G, Schluth-Bolard, C, Monin P, Dupuis-Girod S, Guillen Sacoto MJ, Schnur RE, Zhu Z, Poisson A, El Chehadeh,S, Alembik Y, Bruel AL, Lehalle D, Nambot S, Moutton S, Odent S, Jaillard S, Dubourg, C, Hilhorst-Hofstee Y, Barbaro-Dieber, T, Ortega, L, Bhoj EJ, Masser-Frye, D, Bird L, Lindstrom K, Ramsey KM, Narayanan V, Fassi E,, Willing M, Cole T, Salter CG Akilapa R, **Vandersteen A**, Canham N, Rump P, Gerkes E, Wassink-Ruiter JSK, Bijlsma, Hoffer JV,, Vargas M, Wojcik A, Cherik F, Francannet C, Rosenfeld JA, Machol K, Scott DA, Bacino CA, Wang X, Clark GD, Bertoli M, Zwolinski S, Thomas R, Akay E, Chang RC, Bressi R, Sanchez Russo R, Srour M, Russell L, Goyette AM, Dupuis L, Mendoza-Londono R, Karimov C, Joseph M, Nizon M, Cogné B, Kuechler A, Piton A, Deciphering Developmental Disorder Study, Klee EW, , Lefebvre V, Clark KJ, Depienne C, **Widening of the genetic and clinical spectrum of Lamb-Shaffer syndrome, a neurodevelopmental disorder due to *SOX5* haploinsufficiency** *Genet Med* **2019***, paper accepted*

Publications in Preparation:

Hubmacher D, Dickinson J, **Vandersteen A**, Rideout A, Callewaert B **Weill-Marchesani syndrome caused by *ADAMTS17* missense variants, clinical, functional, molecular investigations**, *Matrix Biology.*

Morlino S, Micale L, Ritelli M, Rohrbach M, Zoppi M, **Vandersteen A**, Mackay S, Agolini E, Cocciadiferro D, Sasaki E, Madeo A, Ferraris A, Reardon W, Di Rocco M, Novelli A, Grammatico P, Malfait F, Mazza T, Hakim A, Giunta C, Colombi M, and Castori M. ***COL1*-related overlap disorder: an additional Ehlers-Danlos syndrome form due to *COL1A1-COL1A2* variants and comprising the osteogenesis imperfecta/Ehlers-Danlos syndrome overlap**.

Book Chapters:

1. **Dentistry** in *Surgery and Healing in the Developing World*, Foskett D and **Vandersteen A,** 2005 Ed. Glenn W. Geelhoed, Landes Biosciences

2. **Combinatorics, Peptide Mimetics and Combizymes**. Han H., **Vandersteen A.,** Janda K in: *Peptides: Frontiers of Peptide Science* (**Proceedings of the 15th American Peptide Symposium)** eds. 1997 , J.P. Tam , T. Pravin, P. Kaumaya, Kluwer/Escom, Nashville, TN**.**

3. **Approach to Dysmorphic Child. Vandersteen A,** in *Clinical Cases in Paediatrics: Trainee Handbook,* 2014 JB Publishing, UK.

Letters to the Editor:

1. **Vandersteen A. Infertility defined.** *The Times 3rd Nov 2017.*

2. **Vandersteen A. Life as a new consultant in a specialist Service**. *British Society of Human Genetics Newsletter Nov. 2011.*

3. **Vandersteen A. Invisible Managers.** *Health Service Journal* 2005 (115) 5978.

4. Hancock A, **Vandersteen A. *Who'd be a surgeon?*** *BMA News* 14th Mar 2009.

5. **Vandersteen A. Staffordshire Hospital is not an isolated case.** *The Times 24th Mar 2009.*

Public Education Materials:

1. **Vandersteen A**, Bartlett M, Cummings C, Pope FM **Guidelines for the management of Joint Hypermobility Syndrome, UK National EDS service patient/clinician information (2012).**

2. **Sobey G**, Bartlett M, Cummings C, **Vandersteen A,** Pope FM **Guidelines for the management of classical Ehlers Danlos Syndrome,UK National EDS service patient/clinician information (2013).**

3. Bartlett M, Cummings C, **Vandersteen A,** Pope FM **Guidelines for the management of vascular Ehlers Danlos Syndrome, UK National EDS service patient/clinician information (2013).**

4. Advisor to ***Coronation Street*** Script writers on EDS storyline 2013.

Publications as Collaborator:

Childhood Overgrowth Study, PIs, Naz Rahman, Kate Tatton-Brown, Royal Marsden Hosp.

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.* **2011** *(12):1127-33.*

Breast and Ovarian Cancer Susceptibility collaboration (BOCS), PI, Naz Rahman Royal Marsden Hosp.

Ruark E, Snape K, Humburg P, Loveday C, Bajrami I, Brough R, Rodrigues DN, Renwick A, Seal S, Ramsay E, Duarte Sdel V, Rivas MA, Warren-Perry M, Zachariou A, Campion-Flora A, Hanks S, Murray A, Ansari Pour N, Douglas J, Gregory L, Rimmer A, Walker NM, Yang TP, Adlard JW, Barwell J, Berg J, Brady AF, Brewer C, Brice G, Chapman C, Cook J, Davidson R, Donaldson A, Douglas F, Eccles D, Evans DG, Greenhalgh L, Henderson A, Izatt L, Kumar A, Lalloo F, Miedzybrodzka Z, Morrison PJ, Paterson J, Porteous M, Rogers MT, Shanley S, Walker L, Gore M, Houlston R, Brown MA, Caufield MJ, Deloukas P, McCarthy MI, Todd JA. **Breast and Ovarian Cancer Susceptibility Collaboration**; Wellcome Trust Case Control Consortium, Turnbull C, Reis-Filho JS, Ashworth A, Antoniou AC, Lord CJ, Donnelly P, Rahman N. **Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer.** *Nature* **2013** 493(7432):406-10., **Large-scale genotyping identifies 41 new loci associated with breast cancer risk.** *Nat Genet* **2013** 45*(4):353-61,*

Genetics of Learning Difficulties Consortium (GOLD), PI Lucy Raymond, Cambridge Univ.

Grozeva D, Carss K, Spasic-Boskovic O, Tejada MI, Gecz J, Shaw M, Corbett M, Haan E, Thompson E, Friend K, Hussain Z, Hackett A, Field M, Renieri A, Stevenson R, Schwartz C, Floyd JA, Bentham J, Cosgrove C, Keavney B, Bhattacharya S; Italian X-linked Mental Retardation Project; **UK10K Consortium; GOLD Consortium**, Hurles M, Raymond FL. **Targeted Next-Generation Sequencing Analysis of 1,000 Individuals with Intellectual Disability.** *Hum Mutat* **2015** (12):1197-204.

UK10K consortium,Rare genetic variants in Health and Disease: PI Durbin R, Incidental findings group chair Plagnol V.

Huang J, Howie B, McCarthy S, Memari Y, Walter K, Min JL, Danecek P, Malerba G, Trabetti E, Zheng HF**; UK10K Consortium**, Gambaro G, Richards JB, Durbin R, Timpson NJ, Marchini J, Soranzo N. **Improved imputation of low-frequency and rare variants using the UK10K haplotype reference panel.** *Nat Commun*. **2015** 6:8111.

Hendricks AE, Bochukova EG, Marenne G, Keogh JM, Atanassova N, Bounds R, Wheeler E, Mistry V, Henning E, Körner A, Muddyman D, McCarthy S, Hinney A, Hebebrand J, Scott RA, Langenberg C, Wareham NJ, Surendran P, Howson JM, Butterworth AS, Danesh J, Nordestgaard BG, Nielsen SF, Afzal S, Papadia S, Ashford S, Garg S, Millhauser GL, Palomino RI, Kwasniewska A, Tachmazidou I, O'Rahilly S, Zeggini E, Barroso I, Farooqi IS; Understanding Society Scientific Group; EPIC-CVD Consortium**; UK10K Consortium**. **Rare Variant Analysis of Human and Rodent Obesity Genes in Individuals with Severe Childhood Obesity.** *Sci Rep.* **2017** 7(1):4394

Deciphering Developmental Disorders 13,600 patients recruited, 71 publications

Wright CF, Fitzgerald T, Jones W, Clayton S, McRae J, van Kogelenberg M, King D, Ambridge K, Barrett D, 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 G, Tivey A, Middleton A, Parker M, Carter N, Barrett J, Hurles M, FitzPatrick D, Firth H, **DDD study. Genetic diagnosis of developmental disorders in the DDD study: a scalable analysis of genome-wide research data.** *Lancet.* **2015** 385(9975):1305-14.

71 publications as collaborator: https://www.ddduk.org/publications.html.

BRCA Testing to Treatment, Community of Practice, Society of Gynecological oncology of Canada

McCuaig JM, Stockley TL, Shaw P, Fung-Kee-Fung M, Altman AD, Bentley J, Bernardini MQ, Cormier B, Hirte H, Kieser K, MacMillan A, Meschino WS, Panabaker K, Perrier R, Provencher D, Schrader KA, Serfas K, Tomiak E, Wong N, Young SS, Gotlieb WH, Hoskins P, Kim RH. **BRCA TtoT Community of Practice. Evolution of genetic assessment for BRCA-associated gynaecologic malignancies: a Canadian multisociety roadmap.** *J Med Genet*. **2018** (9):571-577

UK NIHR BioResource–Rare Diseases

Wei W, Tuna S, Keogh MJ, Smith KR, Aitman TJ, Beales PL, Bennett DL, Gale DP, Bitner-Glindzicz MAK, Black GC, Brennan P, Elliott P, Flinter FA, Floto RA, Houlden H, Irving M, Koziell A, Maher ER, Markus HS, Morrell NW, Newman WG, Roberts I, Sayer JA, Smith KGC, Taylor JC, Watkins H, Webster AR, Wilkie AOM, Williamson C; **NIHR BioResource–Rare Diseases**; 100,000 Genomes Project–Rare Diseases Pilot, Ashford S, Penkett CJ, Stirrups KE, Rendon A, Ouwehand WH, Bradley JR, Raymond FL, Caulfield M, Turro E, Chinnery PF. **Germline selection shapes human mitochondrial DNA diversity.** *Science*. **2019** May 24;364(6442).

Abstracts and Presentations:

1. **Vandersteen A** &and Janda K. **1999. Polyethylene glycol supported Combinatorial Chemistry**," *platform presentation and practical demonstration, International Conference on Combinatorial Library Methods for Basic Research and Drug Discovery, Tuscon Arizona, USA.*

2. Honeychurch J, **Vandersteen A**, Johnson P, Glennie M, Illidge T. **2000**. **Timing and dose of chemotherapy are critical to successful outcome with radio-therapy in B-cell lymphomas.** *Poster*, *British Journal of Cancer, Research meeting, UK*.

3. **Vandersteen A**, Turnbull J, Jan W, Simpson J, Lucas S, Anderson D, Lin J-P, Stratakis C, Pichert G, Lim M. **2007**. **An unusual presentation of a rare syndrome: A case of Carney Complex presenting with a stroke**. *Platform presentation, British Paediatric Neurology Association, Edinburgh, UK.*

4. **Vandersteen A** and Hennekam R**. Two Male Siblings with Mental Retardation, Dysmorphic Facial Features, Unusual Skull Shape, Premature Balding, Small Genitalia and Subluxed Patellae. 2009.** *Poster*, *British Society of Human Genetics annual meeting*

5. **Vandersteen A**, Kenny J, Male A. **2011. Ophthalmic Artery Aneurysm as the presenting feature in a woman with Marfan Syndrome: unusual phenotype or incidental finding?** *Poster, British Society of Human Genetics annual meeting, Warwick UK.*

6. Sobey G**, Vandersteen A,** Ghali N, Pope FM. **2012**. **The clinical and Genetic Spectrum of Classical EDS.** *Poster, Nature Genetics/ J. Investigative Dermatology Miami*

7. Lemmon J, Nesbitt M, Pollitt R, Dalton A, Leary J, **Vandersteen A**, Pope M, Ghali N, Sobey G **. 2012. The UK National EDS Service***. Poster, Belgian Society of Human Genetics First Intl EDS symposium Ghent.*

8. Stembridge NS, Sawle P, Holden S, Ferguson D, **Vandersteen A**, Payne S, Nesbitt M, Pollitt R, Dalton A, Henderson A, Pope FMP, **2012**. **Mutations in the C-propeptide region of the COL3A1 gene cause both typical and atypical forms of the Vascular Ehlers-Danlos Syndrome** *Belgian Society of Human Genetics/ First Intl EDS symposium Ghent, Belgium.*

9. **Vandersteen A. Rare Mendelian Syndromes and Joint Hypermobility. 2013** *Platform presentation, British Society of Rheumatology annual meeting, Birmingham, UK.*

10. Pope M. Burrows N, Shugar A, Pollitt/Nesbitt, Sobey G, Ferguson D, **Vandersteen A. 2013. Some novel COL3A1 mutations cause unexpected clinical phenotypes** *Poster, European Society of Human Genetics annual meeting, Paris, France.*

11. Stembridge N, **Vandersteen A**, Ghali N, Ferguson D, Pope M. **2013**. **Dermal electron microscopy findings in the Ehlers-Danlos Syndromes.** *Poster, European Society of Human Genetics annual meeting, Paris, France.*

12. Stembridge N, **Vandersteen A,** MacDermott K, Pope M**. 2013. Vascular EDS** *Platform presentation and patient demonstration, Royal Society of Medicine (Dermatology section), London UK.*

13. Vandrovcova J, Weerakkody R, Biggs J, Norsworthy PJ, Neuwirth C, Game L, **Vandersteen A**, Pope M, Cheshire N, Aitman T, 2013. **Mutation detection in aortopathy and other vasculopathies complicating hereditary disorders of connective tissue by next generation sequencing.** *Poster, American Society of Human Genetics, Boston, USA*

14. Sangster M, **Vandersteen A**, Finley A. **2015. Hypermobility and Pain***, platform presentation Canadian Pain Society annual meeting, PEI, Canada.*

15. Lindert U, Baumann M, Zschocke J, Fauth C, Abdalla E, Kariminejad A, Lozic B, Pope M, **Vandersteen A**, Rohrbach M, Giunta C. **2016. Clinical Assessment and Molecular Characterization of Patients with FKBP14-Related Ehlers-Danlos Syndrome: Refinement of the Clinical *Presentation.*** *Poster, EDS International Consortium conference, New York, USA.*

16. Reinstein E & **Vandersteen A**. **2016 FLNA – EDS**, *Platform Presentation EDS International Consortium conference, New York, USA.*

17. **Vandersteen A**, Sangster M, Finley A, **2016**. **All my joints hurt and it's my parents' fault**, *Platform Presentation ILC Foundation annual conference, Toronto, Canada.*

18. Zschocke J, Kapferer-Seebacher I, Pepin M, Werner R, Aitman T, Nordgren A, Stoiber H, Thielens N, Gaboriaud C, Amberger A, Schossig A, Gruber R, Giunta C, Bamshad M, Björck E, Chen C, Chitayat D, Dorschner M, Schmitt-Egenolf M, Hale C, Hanna D, Hennies H, Heiss-Kisielewsky I, Lindstrand A, Lundberg P, Mitchell A, Nickerson D, Reinstein E, Rohrbach M, Romani N, Schmuth M, Silver R, Fulya T, **Vandersteen A**, Vandrovcova J, Weerakkody R, Yang M, the Consortium "Molecular Basis of Periodontal EDS", Pope M, Byers P. **2016**. **Periodontal Ehlers-Danlos syndrome is caused by mutations in *C1R* and *C1S*, which encodes subcomponents C1r and C1s of complement.** *Poster, American Society of human Genetics, Vancouver, Canada.*

19. Chakraborty P, Tingley K, Kowalski M, Lamoureux M, Potter B, Coyle D, Wilson K, Austin V, Brunel C, Buhas D, Chapman M, Chan A, Dyack S, Feigenbaum A, Geraghty M, Giezen A, Gillis J, Goobie S, Jain S, Kozenko M, Langley E, Little J, MacKenzie J, Maranda B, Mhanni A, Mitchell G, Mitchell J, Nagy L, Pender A, Potter M, Prasad C, Ratko S, Salvarinova R, Schulze A, Siriwardena K, Sondheimer N, Sparkes R, Stockler S, Trakadis Y, Turner L, Ueda K, Van Karnebeek C, Vallance H, **Vandersteen A**, Walia J, Wilson W, Yuskiv N, and Kronick J. **2016**. **Variation in diagnostic care and disease classification for PAH deficiency: Initial findings from the Canadian Inherited Metabolic Diseases Research Network (CIMDRN).** *Poster, Garrod Conference Montreal, Canada*

20. Murphy L, Tee J, Ramsey S, **Vandersteen A,** Issekutz T and Derfalvi B.**2017**. **Monogenic cause of lupus requiring personalized Treatment***, Poster, IWK Dept Pediatrics Trainee Research Day, Halifax, Canada.*

21. Rideout A, **Vandersteen A,** Parkash S, MacKay S, Dyack S. **2018**. **Case report of mistaken identity, a family with a COL1A1 variant masquerading as Loeys Dietz syndrome due to a TGFB3 mutation**. *Poster Tenth International Research Symposium on Marfan Syndrome, Amsterdam, Holland.*

22. Falkenham A, Issekutz T, **Vandersteen A**, Derfalvi B. **2018**, **Genetic panel testing in children with suspected primary immunodeficiencies and immune dysregulation: experiences in a pediatric tertiary care center** *Platform presentation, IWK Dept Pediatrics Trainee Research Day, Halifax, Canada and Poster, Canadian Society of Allergy and Clinical Immunology, Halifax, Canada.*

23. Duong J, Rideout A, Beis J, Parkash S, Schwarze U**, Vandersteen A. 2018 second patient with Classical Ehlers-Danlos Syndrome (cEDS) and congenital hip dislocation caused by the pathogenic variant COL1A1 c. 934C>T, p.Arg312Cys.** *Poster, European Society of Human Genetics annual meeting, Milan, Italy.*

24. Ghali N, Angwin C, Baker D, Brady A, Johnson D, Pope M, Sobey G, **Vandersteen A**, van Dijk F, **2018. Arterial complications in Classical Ehlers Danlos Syndrome** *Platform Presentation, International symposium on the Ehlers-Danlos Syndromes, Ghent, Belgium.*

25. Ghali N, Brady A, Baker D, Warburton R, Frank M, Legrand A, Hulmes D, Cervi D, von Klemperer C, Pope M, Burrows N, **Vandersteen A**, Kannu P, Robertson L, Lefroy H, Cilliers D, Whiteford M, Jacquemont ML, Germain D and van Dijk F**. 2018**. **Atypical *COL3A1* variants (glutamic acid to lysine) cause vascular Ehlers Danlos Syndro with a consistent phenotype of tissue fragility and skin hyperextensibility.** *Platform Presentation, International symposium on the Ehlers-Danlos Syndromes, Ghent, Belgium.*

26. Ghali N, Green C, Anwin C, Baker D, Bartlett M, Bowen J, Brady A, O'Connell V, Johnson D**, Vandersteen A**, Waburton R, Sobey G & Van Dijk F. **2018**. **A clinical description of 20 newly identified individuals with classical-like EDS with evidence of serious gastro-intestinal complications in two individuals.** *Platform Presentation, International symposium on the Ehlers-Danlos Syndromes, Ghent, Belgium.*

27. Angwin C, Ghali N, Brady A, **Vandersteen A**, Ferguson D, Wagner B, Van Dijk F**. 2018**. **Electron microscopy in the diagnosis of EDS: A descriptive study.** *Platform Presentation, International symposium on the Ehlers-Danlos Syndromes, Ghent, Belgium.*

28. Vandersteen A**, 2019.** *Patient presentation EDS Society annual conference: Rare EDS types, Nashville*

29. Pinard A, Cecchi AC, Rideout AL, Fiander M, Guo D, Parkash S, Walling S, Vandersteen A, Milewicz D ***De Novo* Rare Variants Clustered in a Limited Region of *RNF213* Lead to Early Onset and Aggressive Moyamoya Disease. 2019** Poster Presentation accepted, American Society of Human Genetics Conference, Houston.

Grants / Research Funding:

2011 - 2014 Medical Research Council/ Biomedical Research Centre Imperial College London, (£40,000), MRC PhD student fellowship (Dr. Weerakoddy), 2011- **Sequencing Technologies for Mendelian Disease**, Clinical P.I. Anthony Vandersteen, Nick Cheshire, Tim Aitman, Mike Pope. Laboratory P.I. Prof Tim Aitman (500 patients recruited).

2017 Biomarin pharmaceuticals, unrestricted education grant, IWK PKU clinic $25,000.

2019 IGMM, University of Edinburgh, senior research fellowship 0.2FTE, (10,000GBP). "Exomes and unsolved Ehlers-Danlos Syndromes". PI Prof. Tim Aitman

2019 Rare Disease Foundation and the BC Children's Hospital Foundation (BCCHF), "Identifying the Genetic cause for Familial Abdominal Aortic Aneurysm**" $5,000.00**

2019 Genome Canada, Genomic Applications Partnership Program**, NGS in clinical care,** (3,000,000 CAD application for provincial funding in process)**,** Brock J-A, Dyack S**, Vandersteen A,** Bedard K**.**

2019 Canadian Children's Hospital Foundations**, Personalized medicine initiative, Vandersteen A,** Brock J-A, application in process

Articles Reviewed:

2014 *Orphanet Journal of rare diseases* (1)
2015 *Human Mutation* (1)
American Journal of Medical Genetics part A (1)
2016 *International Journal of Molecular Sciences* (1)
British Medical Journal case Reports (1)
2018 *Matrix Biology (1)*
Medicina (1)

2019 *Genes (1)*
Sage Open Case reports (1)

Grant Review:

2015 Research Foundation, Flanders (Research Grant)
2017 Research Foundation, Flanders (Career Grant)
2018 Research Foundation, Flanders (Research Grant)
2018 - 2020 **Rare Diseases: Models & Mechanisms Network (Canadian National Committee)** monthly telehealth meetings (microgrants)
2019 PSI Foundation Ontario (Research grant)

Clinical/Genetics Research Consortia:

2013 - Present Ehlers-Danlos Syndrome (EDS) International Nosology and Research Consortium.

2018- Present Hypermobile EDS genetic evaluation study

2016 - Present Centre for Genomics Enhanced Medicine, Halifax NS.

2016 - Present Canadian Inherited Metabolic Disease Network (CIMDRN).

2017 - Present Epithelial Genetics Research Group, Halifax NS.

2016 – Present Acadian Breast Cancer Genetics, PI Lynette Penney.

2016 - Present Adams Oliver Syndrome, PI Milan Patel, BC

2015 - Present Periventricular Nodular Heterotopia, PI Stephen Robertson, NZ.

TEACHING

Invited Presentations/CME

1995 **Vandersteen A,** Reese CB. **Use of Henderson-Hasselbach equation in protecting group design**, Chemistry Research meeting, King's College London, UK.

1997 **Vandersteen A**, Han H, Janda K. **Small Molecule Catalysis and Combinatorial Chemistry,** Chemistry Research Meeting, Scripps Research Institute, San Diego USA.

2001 **Vandersteen A, Smoking Cessation**, Respiratory Rounds. Southampton General Hospital, UK.

2002 **Vandersteen A, Weekend Handover Audit Results,** Surgery Rounds, Salisbury Hospital, UK.

2002 **Vandersteen A,** Hulbert D. **Osteomyelitis in a 5 year old Girl**, Emergency Medicine rounds, Southampton General Hospital, UK.

2003 **Vandersteen A**, Hockey P. **Depression in a 78 year old Partially Sighted Lady After Pneumonia**, Grand Rounds, Lymington Hospital, UK.

2004 **Vandersteen A**, Day INM. **Grehlin Receptor Polymorphisms and Obesity in the Hertfordshire Cohort,** Genomics research meeting, Southampton General Hospital, UK.

2005 V**andersteen A, Toxic Shock Syndrome,** Grand Rounds, Eastbourne Hospital, UK.

2007 **Vandersteen A**, Pichert G, Lim M, **Carney Complex**, Ground Rounds, Guys Hospital, London, UK.

2008 **Vandersteen A**, **Macrocephaly,** Paediatric Grand Rounds, Wellington Hospital, New Zealand.

2008 **Vandersteen A, 15q Triplication and Autism,** Genetics/Lab Rounds, Guy's Hospital London, UK.

2010 **Vandersteen A, Mutations, Connotations, Implications**. Grand Rounds, Great Ormond Street Hospitals, London UK.

2011 **Vandersteen A, Genetic Testing and Hypermobility Syndromes**. East Herts Paediatrics Grand Round, Watford General Hospital.

2011 **Vandersteen A, Genetic Testing and Hypermobility Syndromes** Hypermobility Club, London,UK.

2013 **Vandersteen A**, **National EDS service; what is it for and what do we do**. NW Thames Regional Genetics Service Rounds, Harrow, UK.

2014 **Vandersteen A, Genetics and Joint Hypermobility Syndrome**, Rheumatology Rounds, IWK, Canada.

2016 **Vandersteen A** and Parkash S. **Marfan Syndromes and hereditary disorders of connective tissue**, IWK Paediatrics Grand rounds, Canada.

2017 **Vandersteen A**. **Ehlers Danlos Syndromes, lessons from a specialist service** Genome Atlantic/CGEM invited lecture, Halifax, Canada.

2019 Vandersteen A, Snow S, **Exome sequencing in Clinical Care**, IWK Paediatrics Grand rounds, Canada

National Level CME Courses:

2012 **Vandersteen A,** Bartlett M, Pope M, Sobey G**, Classical EDS Masterclass**, Sheffield children's Hospital.
A combined patient support and teaching day for dermatologists and medical geneticists (attendance 50).

2013 Turnpenny P, **Vandersteen A**, Pope M**, EDS Masterclass,** Royal Devon and Exeter Hospital, UK.
A combined patient support and teaching day for dermatologists and medical geneticists (attendance 50).

2013 **Vandersteen A, Genetics and Joint Hypermobility Syndrome**, Institute for Child Health/GOSH National Paediatric Rheumatology training course: 1 hour lecture.

Undergraduate Medical Teaching:

2007 MED2, medical genetics case based learning, King's College Hospital, UK.
2009 MED2 lecture**, Single Gene Disorders** University College Hospitals Medical School, UK.
2009 - 2010 MED2, medical genetics, cased based learning, University College Hospitals Medical School, UK.
2015 - 2016 MED1 human development (half unit), Dalhousie Case based learning tutor.
2017 MED2 integration (full unit), Dalhousie Case based learning tutor.
2018 MED2 integration (full unit), Dalhousie Case based learning tutor.
2018 MED2 lecture **Polygenic Inheritance**.
2018 Clinical Skills: Pediatric Neurology Cases and Examination.
2019 MED2 (full unit), Human Development
2019 MED1 Pediatric clinical skills teaching

Postgraduate Medical Teaching:

2002 **Management of Hip & Pelvic Fractures,** residents teaching**,** Emergency Medicine, Southampton General Hospital, UK.
2002 **Management of Status Epilepticus**, residents teaching**,** Emergency Medicine, Southampton General Hospital, UK.
2003 **Management of Constipation**, residents teaching, Geriatrics, Lymington Hospital, UK.
2005 **The cardiac cycle,** , residents teaching, anaesthetics, Eastbourne Hospitals, UK.
2005 **Foundation year pilot scheme**. First use of competency based training,

evaluation tools (CBD, miniCEX, DOPS) Eastbourne Hospital, UK.

2007 **Agenesis of the Corpus Callosum: antenatal and postnatal outcomes,** FATC rounds, Guy's Hospital, London UK.

2009 **Teaching Genetics course**, National Genetics Education Centre, Birmingham University Hospital, UK. **Mutations, Connotations, Implications**, Bethan Nancy SpR Lecture, Great Ormond Street Hospitals, London, UK, First Prize.

2010 - 2014 **Postgraduate Training supervision for trainees in medical genetics, general medicine (F2 residents) and dermatology (2 clinics per month).**

2013 – 2014 **Clinical Genetics**, Family Doctor/ GP Deanery Teaching, Hertfordshire (1/2 day).

2015 IWK Pediatric residents OSCE examiner.

2017 **Medical Genetics in adult medicine.** CDHA/ QEII Internal Medicine Residents' teaching (1 hour lecture).

2017 **Neurocutaneous syndromes**, IWK Pediatric residents' teaching, (1hr lecture).

2019 **Medical Genetics in adult medicine.** CDHA/ QEII Internal Medicine Residents' teaching (1 hour lecture).

Undergraduate Chemistry Teaching:

1994 - 5 Undergraduate practical class supervision

Undergraduate Genetics Teaching:

2016 **Medical Genetics**, Human Genetics BSc module, (2 x 1 hr lectures), Dalhousie.

Postgraduate Genetics Teaching:

2010 - 2014 **Cancer Genetics,** Human Genetics MSc lecture (1hr lecture), Imperial College London

2011 Hereditary Disorders of Connective Tissue, SpR Teaching (London Deanery)

Secondary Education Teaching Experience:

1990 Cambridge University Teaching Experience Program, 6 x ½ day science/math teaching assistant in local high school classes.

ADMINISTRATION

Committee Memberships/ Clinical Audit and Administration Service:

Jun 2016 - Present	MGTRAC (genetic testing committee) **Chairperson**, IWK/MMGS.
2014 - 2017	LSRI/CGEM Translational Medicine Lectures series **organization committee.**
2015 - Present	Educational rounds scheduling, MMGS.
2014 - Present	Cancer Genetics clinical committee, MMGS.
2014 - Present	Connective tissue genetic disorder committee, MMGS.
2012 - 2014	**Division head -** Ehlers-Danlos Syndrome National Diagnostic Service, London.
2012 - 2013	**Clinical Governance Lead Clinician** - NW Thames Regional Genetics Service.
2010 - 2014	**Genetics Management Group**, monthly management meetings, NW Thames Regional Genetics Service.
2009 - 2010	Great Ormond Street Hospital, Cancer Genetics clinical committee, MDT coordinator: ocular genetics, dysmorphology, skeletal dysplasia meetings.
2008 - 2009	Guy's and St. Thomas' NHS Foundation Trust, London UK. Cancer Genetics, neurogenetics clinical committee Resuscitation/safety officer, library coordinator.
2005	ICU Mortality and Morbidity meeting presentations x 2- Eastbourne Hospital.