# Curriculum Vitae

# C. L. MITCHELL

# BSc (Wits, 2000)

# BSc (Hons) (Wits, 2001)

# MSc (Med) (Wits, 2009)

# Diploma of Photography (Photography Institute, 2010)

June 2018

# CURRICULUM VITAE OF CLAIRE LYNNE MITCHELL

# PERSONAL DETAILS

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| --- | --- | --- | --- |
| Surname: | Mitchell | | |
| First names: | Claire Lynne | | |
| Date of birth / Age: | 18 January 1980 | | 38 years |
| Identification number: | 800118 0017 089 | | |
| Nationality: | South African | | |
| Gender: | Female | | |
| Marital status: | Married | | |
| Maiden surname: | Hetem | | |
| Residential address: | 2 Oregon  Honeydew Ridge Residential Estate  Emily Hobhouse Road  Wilgeheuwel, ext 17  Roodepoort  1735 | | |
| Postal address: | PO Box 5132  Weltevreden Park  1715 | | |
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| e-mail: | clairelynnemitchell@gmail.com | | |

# FORMAL EDUCATION / QUALIFICATIONS

**Tertiary Education**

**Diploma of Photography**

The Photography Institute

2010 (Part time study)

**MSc(Med) Human Genetics**

University of the Witwatersrand

2004-2009 (Part time study)

Project title: Improved mutation detection for Haemophilia A in South Africa

**BSc(Hons) Human Genetics**

University of the Witwatersrand

2001 (Full time study)

Project title: Investigation of the Trinucleotide Repeats of the Huntington Disease Gene in the South African population

**BSc**

University of the Witwatersrand

1998-2000 (Full time study)

Majors: Genetics and Advanced Biology

**High School**

Allen Glen High School (Allens Nek, Roodepoort)

1993-1997

Matriculated with 7 subjects

# PROFESSIONAL MEMBERSHIPS

Health Professional Council of South African (HPCSA)

Medical Scientist registration number: MW 0009199

2002-current

South African Society of Human Genetics (SASHG)

2001-2011

# CAREER HISTORY

**The King’s School West Rand**

Period of employment: January 2018 to present

Position held: Teacher

Functions performed:

* Educator in Grade 7 Mathematics and Grade 7 Natural Science
  + Teaching of classes
  + Setting class assessments, test and exams
  + Marking of all assessments and exams
  + Additional lessons

**Somatic Cell Genetics, Department of Haematology, National Health Laboratory Service**

Period of employment: June 2015 to December 2017

Position held: Medical Scientist

Functions performed:

* Diagnostic molecular testing and monitoring of response to treatment in patients with Chronic Myeloid Leukaemia
  + RNA extractions
  + Quantitative reverse transcriptase PCR (RQ-PCR)
  + Qualitative reverse transcriptase PCR (RT-PCR)
  + Mutation detection for resistance to treatment in CML
  + Interpretation of results
  + Writing patient reports
  + Liasing with doctors and laboratories
* Fluorescent *in situ* hybridisation (FISH) analysis on blood smears, cultured cells and tissue sections
  + Preparation of slides
  + Hybridisation of probes to slides
  + Analysis of patient results using different probes (break apart probes, translocation probes, deletion probes, amplification probes, inversion probes and centromeric probes)
  + Interpretation of results and writing reports
  + Liasing with clinicians regarding complicated cases
* Teaching and training of students, interns and registrars

**School of Health Science, Monash South Africa**

Period of employment: February 2014 to November 2014

Position held: Sessional lecturer

First semester unit: **Biological Basis of Health & Disease 1** (February – June)

Second semester unit: **Biological Basis of Health & Disease 2** (July – November)

Functions performed:

* Lecturing to 1st year students (85 students)
* Tutoring: 3 tutorial classes per week
* Marking: exams, tests and assessments
* Liaising with the Chief examiner in Australia regarding the unit
* Assisting with setting of test and exam papers for the unit

**School of IT, Monash South Africa**

Period of employment: June 2013 to November 2013

Position held: Temporary School Administrator

**Self employed**

Period of employment: May 2010 to 2013

Position held: Freelance Professional Photographer

**National Health Laboratory Service (NHLS)**

Period of employment: December 2001 to July 2007

Position held: Medical Scientist

Functions performed:

* Routine diagnostic testing
* Process, register and extract DNA from patient samples
* Perform the specific molecular genetic testing for over 30 single gene disorders, using numerous techniques
* Analyse the results obtained and write patient reports
* Process final patient reports and send to the referring doctor or laboratory
* Consult with doctors, nurses, private laboratories and patients (telephonically) regarding genetic testing and result enquiries
* Preparation of diagnostic laboratory reagents
* Special diagnostic testing
* Development of special/family-specific diagnostic tests in our laboratory for cases where a mutation has been identified in either our laboratory or another laboratory (eg: United States, United Kingdom, Europe or another South African laboratories) to aid with carrier detection and prenatal diagnosis for the family
* Teaching
* Actively involved with teaching and training of medical scientist interns
* Assistance with the BSc(Hons) course offered by the department, including
* Co-supervision of students for the year of their degree
* Assistance with the techniques course for all BSc(Hons) students
* Assist with other BSc(Hons) students in training of certain techniques and advice on project approach and optimisation
* Assistance with teaching and training of new staff members and other students
* Involved in tutorials and lectures for MMBCH and GEMP students
* Demonstration and discussion/explanation of techniques and tests to school and university students interested in a career in genetics/human genetics
* Safety and first aid officer (2003-2006)
* Research and development
* General research and development of new techniques and tests to be implemented into the diagnostic service, to continually improve the service offered by the department to the public.
* For example, research and development projects optimised, included, among others:
  + Duchenne/Beckers muscular dystrophy carrier detection
  + Determination, optimisation and implementation of the Huntington Disease type 2 (HDL2) mutation into routine diagnostic testing in the South African population
  + Optimisation of 4 additional mutations to improve the Ashkenazi carrier screen
  + Optimisation of the MLPA (Multiplex Ligation-Dependent Probe Amplification) assay for detection of deletions involved in Duchenne/Beckers Muscular Dystrophy
* Research projects for disorders of interest to the department, for example:
  + Rett syndrome
  + Determination of a HDL2 African founder mutation

# EXPERIENCE IN THE FOLLOWING MOLECULAR TECHNIQUES

* DNA extraction (from blood, CVS, amniotic fluid, paraffin embedded tissue, cultured cells and tissue samples)
* Polymerase chain reaction (PCR)
* Agarose gel electrophoresis
* Polyacrylamide electrophoresis
* Microsatellite analysis on the ABI377 automated DNA sequencer (Perkin Elmer)
* Restriction enzyme digests
* Southern Blotting
* dHPLC using the WAVE® machine (Transgenomic)
* Sanger sequencing using the ABI377 automated DNA sequencer and the 3130*xl* genetic analyser (Perkin Elmer)
* Multiplex Ligation-Dependent Probe Amplification
* RNA extraction from blood and bone marrow samples
* Qualitative reverse transcription PCR (RT-PCR)
* Quantitative reverse transcriptase PCR (RQ-PCR) using the ABI3700 light cycler
* Preparation of samples (blood smears, cultured cells and tissue sections) for FISH analysis
* Hybridisation of FISH probes to samples fixed onto slides
* Analysis of FISH probes (including break apart probes, translocation probes, deletion probes, amplification probes, inversion probes and centromeric probes)

# PUBLICATION RECORD

**Original articles**

A KRAUSE, **CL MITCHELL**, F ESSOP, S TAGER, J TEMLETT, G STEVANIN, CA ROSS, D RUDNICKI, RL MARGOLIS. (2015). Junctophilin 3 (*JPH3*) expansion mutations causing Huntington Disease Like 2 (HDL2) are common in South African patients with African ancestry and a Huntington Disease phenotype. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, **168B**:573–585.

T WAINSTEIN, R KERR, **CL MITCHELL**, S MADAREE, FB ESSOP, E VORSTER, R WAINWRIGHT, J POOLE, A KRAUSE (2013). Fanconi anaemia in black South African patients heterozygous for the FANCG c.637-643delTACCGCC founder mutation. *SAMJ* **103**(12):970-973.

MITCHELL C, **MITCHELL CL**, KRAUSE A (2007). New Factor IX linked marker alleles in African Haemophilia B patients. *Haemophilia* **13(5)**:642-644.

HAW T, WAINWRIGHT L, HALE M, POOLE J, GUNTER K, PATEL M, **MITCHELL CL**, ESSOP F, KRAUSE A. (In Press). The FANCG deletion mutation in black South African patients with acute myeloid leukaemia or aplastic anaemia.

**Presentations at Scientific Meetings (2001-2005)**

**MITCHELL CL**, MADAREE S, KRAUSE A (poster presentation). Fanconi Anaemia in black South African patients heterozygous for the 637-643del mutation. 11th Southern African Society of Human Genetics Congress, Golden Gate, 1-3 March 2007.

MUDAU MM, ESSOP F, **MITCHELL CL**, ROBINSON C, KUHN K, KRAUSE A, RAMSAY M (poster presentation). Assessment of DNA quality over a 20 year period following the salting out extraction procedure. 11th Southern African Society of Human Genetics Congress, Golden Gate, 1-3 March 2007.

ROBINSON CJ, SHARIR Y, ESSOP F, **MITCHELL CL**, KUHN K, KRAUSE A (poster presentation). Carrier screening at the NHLS for 9 diseases common in the Ashkenazi Jewish population. 11th Southern African Society of Human Genetics Congress, Golden Gate, 1-3 March 2007.

KRAUSE A, **HETEM C**, HOLMES SE, MARGOLIS RL (paper). HDL2 mutations are an important cause of Huntington’s Disease in patients with African ancestry. Journal of Neurology, Neurosurgery & Psychiatry, World congress on Huntington’s Disease. Manchester, October 2005

**HETEM CL**, KRAUSE A, HOLMES SE, MARGOLIS R (paper presentation). Huntington Disease type 2 (HDL2) haplotype analysis suggests that the mutation has a single African origin.Combined meeting of the South African Human Genetics Society and the African Human Genetics Society, Glenburn Lodge, Muldersdrift, 13-17 March 2005

MITCHELL C, **HETEM CL**, KRAUSE A. (poster). Assessment of 3 linked markers for Haemophilia B for carrier detection and prenatal diagnosis in South Africa. Combined meeting of the South African Human Genetics Society and the African Human Genetics Society, Glenburn Lodge, Muldersdrift, 13-17 March 2005

KRAUSE A, **HETEM CL**, HOLMES SE, MARGOLIS R. (poster) Genetic heterogeneity in South African patients with the Huntington Disease phenotypes. American Society of Human Genetics Congress, Toronto, Canada.

**HETEM CL**, ESSOP FB, KRAUSE A (poster) Rett syndrome in South Africa. University of the Witwatersrand, Faculty of Health Science, Research Day, 4 August 2004.

**HETEM CL**, ESSOP F, KRAUSE A (paper) Rett syndrome in South Africa. 10th Biennial Congress of the Southern African Society of Human Genetics, Riverside Hotel, Durban, 11-14 May 2003.

LUBBE SJ, VAN HOUGENHOUCK-TULLEKEN WG, **HETEM CL**, LABRUM R, KERR R, BROOKSBANK R, RAMSAY M (poster) Examining candidate genes for complex disorders: idiopathic cardiomyopathy and XX true hermaphroditism as examples. 9th Biennial Congress of the Southern African Society of Human Genetics Congress, Kruger Park, 12-15 August 2001.

**Higher degrees co-supervised and completed (2003 – 2008):**

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| Smita Madaree | 2005 | BSc (Hons) | FA-G in the Black Populations of South Africa: Searching for a second mutation |
| Cathrine Mitchell | 2004 | BSc (Hons) | Assessment of three polymorphic markers for carrier detection and prenatal diagnosis of Haemophilia B in South Africa |
| Neo Motaung | 2003 | BSc (Hons) | Screening for two mutations in the FANCA gene in South African black patients with Fanconi Anaemia |

# REFERENCES

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| --- | --- | --- | --- |
| 1 | Prof Amanda Krause | 011 489 9219 | amanda.krause@nhls.ac.za |
| 2 | Mrs Hannelie Bothma | 011 489 8596 | hannelie.bothma@nhls.ac.za |
| 3 | Dr Basia Diug | +61 439 560 809 | basia.diug@monash.edu |