EXHIBIT Y

EXPERT WITNESS STATEMENT

PREPARED BY PROFESSOR JONATHAN R SKINNER 31/03/2019

My name is Jonathan Robert Skinner

I have read the expert witness code of conduct provided to me and agree to be bound by it.

Current Position

I am a paediatric cardiologist and cardiac electrophysiologist, since 1998, working as a Senior Medical Officer, employee of Auckland District Health Board, within the Department of Paediatric and Congenital Cardiac Services at the Starship Children's Health, Auckland City Hospital, Park Road, Auckland, New Zealand. This report was prepared in my own time and does not necessarily reflect the views of my employer.

I am Honorary Professor in Paediatrics, Child and Youth Health at the University of Auckland.

Qualifications

My qualifications are MBCHB, DCH (RCP Lond), MRCP(UK), FRACP, MD

I am a Fellow of the Cardiac Society of Australia and New Zealand (FCSANZ), and am in my fifth year as chair of the genetics council of CSANZ. I am a Fellow of the Heart Rhythm Society (FHRS).

Relevant experience

Since 1998 I have worked as a paediatric electrophysiologist (heart rhythm specialist) leading a national heart rhythm service for children and also for adults with congenital heart disease. In 2003 I established and now currently chair the national cardiac inherited disease group (CIDG) which provides a coordinated national service for the detection and screening of families with inherited heart conditions, most of which can cause sudden death in young people. The attached clinical registry in New Zealand now holds more than 3,800 people.

In 2006-2008 I led a combined Australian and New Zealand Task multidisciplinary task force with the aim of reducing sudden cardiac death in young people, and chaired the writing group which wrote the best practice document to guide the appropriate investigation of young sudden death. Published in 2008, this was endorsed by the Royal College of Pathologists of Asutralasia, the Human Genetics Society of Australasia and the Cardiac Society of Australia and New Zealand.[1] This same group published a 3 year all cause study of young sudden death in 1-35 year olds in Australia and New Zealand, and CIDG published on the role of genetic testing for long QT syndrome in sudden infant death syndrome.[2 3] I have authored 130 peer-reviewed publications, more than 60 related to inherited heart conditions and young sudden death, with a focus on long QT syndrome, and the value of genetic testing in unexplained sudden death.[4-15]

I have expertise in the interpretation of ECGs in rhythm disorders in children,[16-18] particularly in the long QT syndrome, and have co-authored guidelines as to how to make such measurements.[19]

Since 2008 CIDG (NZ) have worked with the NZ national coronial and forensic services for the investigation of young sudden natural deaths, administering a government budget for the genetic

testing of those who have died suddenly where no cause has been found on autopsy. This involves the multidisciplinary review of all sudden unexplained deaths in young people referred via the coronial service (698 cases since 2008). I run a cardiac genetic clinic which assesses families who have experienced a young sudden death. Through this I have become experienced in the assessment of both adults and children for features of inherited heart conditions, particularly cardiac ion channelopathies (see below).

What cardiac genetic conditions cause sudden death where the autopsy is uninformative?

Most sudden cardiac deaths in children occur in children with a structurally normal heart which, after death, looks normal both to the naked eye and under the microscope. The genetic defects are collectively known as :"Cardiac Ion Channelopathies". The disorders are in the cardiac cells at a sub microscopic level. Rapid movement of sodium, potassium and calcium ions across the cardiac cell wall are required for depolarisation and repolarisation of the cardiac cells with every heart beat. If the channels through which the ions travel are defective, then repolarisation or depolarisation is abnormal and there is a risk of a serious ventricular arrhythmia - a rhythm so fast and uncoordinated that there is no output from the heart. Sudden syncope (blackout), cardiac arrest or sudden death occurs.

The conditions most commonly implicated are long QT syndrome, CPVT (catecholaminergic polymorphic ventricular tachycardia), and Brugada syndrome. A long list of genes can cause sub-types of these conditions.[20]

The mode of death, and the rhythm recorded during transient loss of consciousness due to these conditions, is ventricular tachycardia and/or ventricular fibrillation (VT or VF) (and not asystole or a slowing of the heart).

The appropriate investigation of young sudden unexplained deaths in 2019 and how this is different from the 1989-1999.

The Australia and New Zealand Best practice guidelines (2008) mandate several things that were not routine at the time of these deaths. These include:

1 A through family history taken to look for evidence of young sudden death or syncope/seizures.

2 A thorough investigation of the first degree relatives for clinical signs of an inherited condition (including a thorough clinical history and cardiac tests such as ECG, an ECG stress test and echocardiogram) .

3. Preservation of blood/tissue for genetic analysis

Other international authorities support this approach.[21]

Such investigations were not mandated prior to this, and as a consequence retrospective review of such cases often reveals gaps in the investigation by today's standards.

The investigation in this family

The advances in DNA extraction and genetic testing technologies have, remarkably, allowed for new extensive testing on Kathleen Folbigg and the four deceased children. This aspect of the investigation is brought up to date. However the lack of thorough cardiac and other physical assessment of the children during life (subsequent to a death of their previous sibling) and both parents means the cardiac investigation will always be less thorough and lower quality than by today's standards.

General comments about this multiple death scenario

In my more than 15 years investigating young sudden natural deaths and managing families with cardiac conditions which can cause sudden death, I have never encountered four sudden deaths of children of any age within one family unit. When there have been more than two *infant* deaths, there has been circumstantial evidence to suggest suboptimal sleep environment for the infants in such cases. However, there is a small number of cardiac inherited conditions where such a scenario of multiple sudden deaths in infancy is theoretically possible, so I enter this investigation with an open mind. (We have, like others, seen families with multiple deaths within the wider family, though to date none have arisen where such a young age has been a consistent feature, rather the age and circumstances of death have been more varied.)

Type of inheritance

For a genetic cardiac condition to cause such sudden deaths in children at such a young age, the condition must be highly malignant, and thus *its presence in either parent would seem very unlikely*, since they would not be expected to be alive, or would have features of severe disease. In general such malignant cardiac genetic conditions arise in three circumstances

1. "de novo"- the mutation arising first in the child- such that only that single child would be affected.
2. as a consequence of germ-line mosaicism - where the mutation has occurred within the gonads of either parent (ie in the ovaries or testes) giving an up to 1 in 2 chance of each child being affected,
3. As a combination of a recessive gene , one coming from each parent, (giving a 1 in 4 chance of each child being affected)

The most common (but still rare) example of 1 and 2 would be Catecholamine sensitive polymorphic ventricular tachycardia (CPVT), most commonly due to mutations in the RYR2 gene (which have not been identified in anyone here). In this condition the resting 12 lead ECG is typically normal. Death in infancy is uncommon, usually occurring after the age of 4 years, the median age at presentation being 11 to 15 years.[22 23] Another condition would be Calmodulin type long QT syndrome, (mutations in the CALM genes) which would be associated with an abnormal ECG (which was not the case for Patrick for whom there is an available 12 lead ECG for review).

For number 3 (recessive conditions), the example would be "Triadin knockout syndrome", a highly lethal condition causing sudden cardiac death in infants and toddlers. No Triadin gene mutations were found, and the ECGS are profoundly abnormal in this condition, again ruling out Patrick. Another would be Jervell and Lange Nielsen syndrome, where long QT syndrome comes down from both sides of the family. Its prevalence is about 1 per million. But the children would have been deaf,

and ECGs would have shown gross QT prolongation, and some QT prolongation in Kathleen, which was not the case. Another would be a rare form of CPVT with mutations in CASQ2, again not found in anyone tested in this family.

In passing I mention that recessive conditions which can cause sudden unexpected death are more common among metabolic and neurological disease. Those conditions causing sudden deaths usually have autopsy findings to suggest such an abnormality, or abnormal metabolic screening tests. Investigation of a near-miss case (such as Patrick) usually reveals abnormal metabolic test results. Others are better qualified to address this and have already done so in previous reports.

The cardiac phenotype (clinical features suggesting heart disease in this family)

Craig

No cardiac testing has been presented on Craig. The same investigations would be recommended in him as in Kathleen.

Caleb

No Cardiac tests available for review.

Patrick

Following his first presentation with collapse, 9 lead ECG was done, dated "(?)18/10/1990":

Normal sinus rhythm, no abnormality detected. No features of long QT or Brugada syndrome, No AV conduction block.

Baseline wander makes some leads difficult to measure but QT intervals measured in lead I, V3 and V6.

Lead 1 QT 0.25/R-R 0.4 **QTc 0.395sec**, V3 QT 0.23 R-R 0.46 **QTc 0.0.34sec,** V6 QT R-R 0.44 **QTc 0.38sec.**

Another ECG, with separate short strips stuck into the notes is not dated but appears in the notes just prior to 5/6/1990. The labelling is difficult to read but lead II is quite well seen. QT 0.3 R-R 0.56 QTc 0.40 sec

Echocardiogram (16/11/1990):

An echocardiogram was done and reported by Dr Gary Warner, an experienced cardiologist with a great deal of experience in the interpretation of childhood echocardiograms in Newcastle. He reports that the echocardiogram was normal

Comment:

Patrick had normal ECGs for age, no abnormality seen. In particular there are no features of sinus node disease or atrioventricular conduction block, there are normal repolarisation patterns, QT intervals in the lower end of the normal range; effectively excluding Long QT syndrome. The echocardiogram showed no evidence of heart muscle disease.

Sarah

I have seen no cardiac test results for Sarah.

Laura

Laura had a single ECG rhythm strip recorded during polysomonography in Augusts 1997 and February 1998. They were reported by Dr Chris Seton in the sleep disorders unit at Westmead as being normal, revealing no abnormal rhythms. I have reviewed some preserved extracts of these recordings and can state that the rhythm is normal sinus rhythm throughout, no pauses, no abnormal extra beats, no evidence of atrioventricular block. Such recordings are not of sufficient quality for the reliable assessment of the QT interval or other indices of repolarisation.

I have not seen a 12 lead ECG on Laura.

Extended single lead ECG strips recorded during the resuscitation efforts by the ambulance crew are available for review. (1/3/1999 from 12:17 onwards). These are difficult to interpret because the dominant signals are most probably artefact from regular chest compressions at a rate of about 100-110 compressions per minute, with intermittent pauses which presumably represent the periods when the ambulance officers evaluated the rhythm to see if electrical cardioversion would be helpful. In these gaps we see a mixture of asystole, and periods of a very slow broad QRS complex rhythm (about 1-2 beats every 9 second screen shot), likely to represent agonal rhythm and unlikely to be associated with a cardiac output .

Significance of the agonal rhythm- does it point to a particular diagnosis?

I was asked to address this particular question.

An agonal heart rhythm like this can be seen after any sudden death, be it cardiologic, respiratory or neurologic. It is a sign of a very sick, dying heart.

Clinicians see this rhythm in children most commonly during or after a failed resuscitation after a respiratory arrest, or asphyxia, or from a neurological cause. However it can also occur following a primary cardiac arrest most typically in people with an already sick heart from a heart muscle disorder (cardiomyopathy).

These days many people of all ages at risk of cardiac arrest have an indwelling device which records the rhythm before, during and after a lethal arrhythmia. However when the devices we implant see VT or VF, they deliver a shock. *So it is uncommon to analyse a rhythm strip from someone who is known to have had VF who has not had a shock, some time after the event*. It is most likely that if Laura's death was due to VF or VT, that rhythm (VT or VF) would still have been be running when the ambulance crew arrived. However we do know that VT and even VF do sometimes spontaneously stop, and it is conceivable that this would result in asystole, and/or an agonal rhythm because the heart was now too sick to function normally.

I consider that the presence of this rhythm makes a non-cardiac death more likely than one from a primary cardiac arrhythmia, but I don't think that is conclusive.

Impression- Laura's cardiac phenotype

The ECGs available are not of a quality whereby a cardiac ion channelopathy can either be diagnosed or excluded.

Kathleen- cardiac phenotype

I have not been able to interview Kathleen, and to my knowledge she has not been examined by a cardiologist specialising in arrhythmia or inherited heart conditions. Making a judgement about the nature of cardiac symptoms is a highly skilled process and pivotally depends on the right clinical history being taken. Syncope (sudden loss of consciousness) or presyncope (almost losing consciousness), are both common, and usually are not related to heart rhythm disturbance.

Features which point to a cardiac arrhythmia as a cause of syncope would be:[24 25]

1 The coincident presence of palpitations

2 Sudden syncope without warning, particularly during exercise

3 Injury occurring at the time of syncope, especially to the face (unconscious before hitting the ground)

4 Absence of nausea, prior

5 Absence of factors likely to lead to a fall in blood pressure (eg heat, dehydration, pregnancy, vomiting illnesses and certain medications)

Kathleen

I have read through the bundle provided which includes some of Kathleen's clinical history. This is a poor substitute for direct questioning. Nevertheless, taking each event in the notes at a time:

15.2.89- Kathleen had an EEG following a blackout during pregnancy. I don't have further details of what led up to this. There is a comment on the referral "? syncope and fitting".

Comment: Fainting is common during pregnancy, usually a haemodynamic /blood pressure issue and rarely due to a rhythm disturbance, though this is possible. Some minor seizure activity can occur following a common faint (due to a reduced cerebral blood flow at the time) and would not be helpful in differentiating the cause of the syncope.

2004 there is a self-filled Health report form apparently signed by Kathleen where she reports "no seizures".

8/3/09 Kathleen presented for review with feeling dizzy 2-3 times a day after hitting her head on a shower.

Comment: It seems very unlikely these events were cardiac related.

2/5/08 Here there was an "Alleged" collapse in her cell. The recorded notes describe "Collapse was after a vomit. Found sweaty and with reduced blood pressure." She was given fluid and electrolytes with gradual resolution of her blood pressure and her symptoms.

Comment: This would be consistent with a common situational vaso-vagal common faint related to dehydration with nausea and vomiting.

May 2011- Kathleen was reviewed by a cardiology registrar at Westmead Hospital following five epsiodes of chest pain over the previous year. Her chest pain was described as atypical for angina, and occurred at times of stress. Her doctor describes a normal resting 12 lead ECG, and a normal exercise test with no ECG changes diagnostic of ischaemia. I have not seen the ECG from that exercise test but it was reported as normal in the letter by Dr Adriani.

Comment: I would not have confidence that the ECGs would have been reviewed for QT length given that the test was to look for ischaemia, though it is likely a profound abnormality would be recognised. I strongly suspect though that any significant arrhythmia (such as may occur with CPVT for example) would have been reported.

I have reviewed a poor quality copy of a **12 lead ECG** from 31 January 2011:

The rhythm is normal sinus rhythm, the depolarisation and repolarisation patterns are normal, with nothing to suggest Brugada syndrome, long QT syndrome, early repolarisation or cardiomyopathy.

The automated QTc is 405msec. My measurements are

Lead 2 QT 380 R-R 880 **QTc 0.41**

V5 QT 400 R-R 940 **QTc 0.41**

Comment: Normal ECG without any features of long QT syndrome or other ion channelopathy.

An **echocardiogram** was reported to reveal a structurally and functionally normal heart with normal wall thicknesses and chamber dimensions.

23/06/2017- Clinical notes reveal that Kathleen turned white whilst in bathroom and "grabbed the bar of the toilet", "slowly turned white and began to collapse". She did not injure herself but was found to be drowsy, and pale and "actively vomiting". Note was made that during her medical evaluation her heart rate rose from 67 to 113 on standing and she felt "washed out and thirsty". She was encouraged to drink more.

Comment: The presence of nausea and vomiting, thirst and tachycardia on standing all point to a decreased intravascular volume. This was likely a common faint, although she didn't completely lose consciousness (presyncope).

24/12/2018

A Good quality **12 lead ECG** is done as part of the current investigation. I have reviewed this carefully.

Heart rate 64 beats per minute in normal sinus rhythm. The automated QTc interval is 422msec, (0.42 sec) well within the normal range (upper limit of normal being 0.47 for an adult female). My manual measurements are:

Lead 2 QT 0.39 R-R 0.95 **QTc 0.40sec** lead V5 QT 0.39 R-R 0.87 **QTc 0.42sec**

Comment: The ECG is normal. Important negative findings are the lack of a Brugada signature, conduction disease and early repolarisation. The depolarisation and repolarisation patterns are normal, in particular the T wave shape is normal.

27/12/2018 A **chest X Ray** reports a normal cardiac size and contour.

22/02/2019 An **echocardiogram** is reported as normal by Dr Mikhail Altman at Westmead hospital. Important negative findings include the normal chamber size and normal left ventricular septal and free wall thickness, and normal left ventricular function.

Comment on the cardiac phenotype in Kathleen:

A complete investigation by today's standards would include tests looking for occult disease. This would include a detailed cardiac history taken by a specialist, an exercise test with analysis of the ECG during the test and during recovery, a standing ECG test, and a 24 hour ECG. (Holter). Extending tests to Cardiac MRI, and drug challenge tests (Ajmaline and Adrenaline) would also be optimal.

However some conclusions can be drawn.

Kathleen is now over 50 years of age. This is 50 years over which an inherited heart condition can present, and signs on cardiac tests can develop. Conditions such as hypertrophic or dilated cardiomyopathy tend to progress over time, and if she was going to develop these conditions, by 50 we would reasonably expect clinical signs or abnormal ECG or echocardiogram by now. She has no features of these cardiomyopathies. These were not suspected in the deceased children at autopsy so it is reasonable to exclude these as possible cause of death in the children.

Regarding cardiac ion channelopathies, she has not had a cardiac arrest, and the syncopal episodes described would be consistent with situational or vasovagal syncope rather than arrhythmic syncope. The ECGs show no features to suggest Brugada syndrome or long QT syndrome, nor sinus node disease or other conduction system disease.

Gene carriage for long QT syndrome can be revealed by provocation ECG, such as an exercise test. However, Kathleen's QT intervals are at the lower end of the normal range, and the T wave shape does not reveal any abnormality, as is common in gene carriers with occult disease.

My opinion is that Kathleen does not have long QT syndrome. Furthermore she has no ECG features to suggest she is a gene carrier for it on two 12 lead ECGs, and so I think it is unlikely that she carries a pathogenic long QT gene.

Comments regarding the genetic findings and the clinical features

Regarding the variants reported as likely pathogenic by Professors Vinuesa and Cook.

I am not a specialist in variant determination and will leave that to the genetic experts. I make comments about the relevant clinical phenotype which would be associated if they were truly pathogenic and disease causing variants.

IDS: Variant found in Patrick only. Not my area of expertise. Since this is X-linked it would not be a plausible cause of death in Sarah or Laura, and it was absent in Caleb. This condition has an overt clinical phenotype and is not a known cause of sudden unexplained death to my knowledge.

CALM2: Variant found in Laura and Sarah and Kathleen.

Calmodulin disease would potentially be a plausible cause of death in the two carriers (Laura and Sarah). The ECGs are typically very abnormal in this condition. Given we do not have 12 lead ECGs in either Laura or Sarah, who are carriers, this cannot be excluded by this method.

However to date, when a variant has been found to be disease causing, the variants cause severe disease, resulting in cardiac arrest or death at a young age and almost invariably with severe QT prolongation when discovered prior to death. Such variants occur denovo and are not compatible with long life without treatment.[26-28] The presence of the same variant in Kathleen, who has not suffered a cardiac arrest, nor typical arrhythmic syncope, and has a normal ECG, effectively excludes this variant as a cause of disease in these children.

(I note also that in assessing pathogenicity of this variant a statement is made that no missense benign variants are described in CALM2. In fact there are many described in the population databases.[29])

MYH6: A Variant found in Kathleen, Caleb and Laura. This variant is present in Kathleen who at aged over 50 has no evidence of hypertophic cardiomyopathy (HCM) or conduction disease. There was no evidence of HCM on any child's autopsy, and no evidence of conduction disease in Patrick or Laura (based on a rhythm strip ECG). This can be effectively eliminated as a possible cause.

Regarding the VUS findings, I shall restrict my comments to KCNQ1, since this is a gene linked to long QT syndrome, a condition I have a good deal of experience with.

KCNQ1: A novel intronic variant is found all of the children and in Kathleen.

The risk of death due to long QT type 1 (which is the type linked to KCNQ1 mutations) is associated with the length of the QT interval. Cardiac events are typically triggered by stress; and death during sleep is uncommon with long QT type 1. Cardiac arrest as the first presentation is also very uncommon in long QT type 1, with recurrent syncope during exercise being the commonest presentation. [30]

KCNQ1 variants have rarely been found in SIDS cohorts.[3 31-33] However cardiac events including sudden death due to long QT type 1 are much more common in boys between the age of 15 and 15 and adult women.[34 35]

Death or near miss episodes in infancy from long QT syndrome are very rare indeed and usually associated with de novo, very severe mutations and gross QT prolongation, and much more commonly associated with KCNH2 and SCN5A (long QT type 2 and 3).[36 37] The presence of this variant in the mother rules out this possibility. Furthermore Kathleen has a normal QT interval and T wave shape, as did Patrick.

I have never seen or heard of a cluster of infant deaths in a family with long QT type 1 and consider this to be extremely unlikely, and confidently rule it out given the normal ECGs in Patrick and Kathleen.

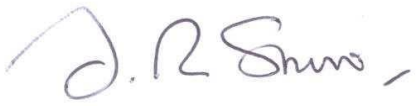
Summary

The available cardiac tests in Kathleen and in the children provide no evidence of a cardiac inherited disease.

Further cardiac evaluation of both parents would complete an optimal investigation. However, the severity of any putative cardiac condition, sufficient to cause death in four such young children, means that these conditions would not likely be present in either of the parents in some minor form only manifest by provocative testing or high quality imaging.

Genetic testing, when combined with the available phenotypic data, has not revealed a cardiac genetic cause for the deaths.

My opinion is that the available clinical data, and genetic analyses, provide no convincing evidence for the presence of any known form of cardiac inherited disease as a potential cause for the sudden deaths of these four children.



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32. Tester DJ, Wong LCH, Chanana P, et al. Cardiac Genetic Predisposition in Sudden Infant Death Syndrome. J Am Coll Cardiol 2018;**71**(11):1217-27 doi: 10.1016/j.jacc.2018.01.030[published Online First: Epub Date]|.
33. Arnestad M, Crotti L, Rognum TO, et al. Prevalence of long-QT syndrome gene variants in sudden infant death syndrome. Circulation 2007;**115**(3):361-7 doi: CIRCULATIONAHA.106.658021 [pii]

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34. Hobbs JB, Peterson DR, Moss AJ, et al. Risk of aborted cardiac arrest or sudden cardiac death during adolescence in the long-QT syndrome. JAMA 2006;**296**(10):1249-54 doi: 10.1001/jama.296.10.1249[published Online First: Epub Date]|.
35. Sauer AJ, Moss AJ, McNitt S, et al. Long QT syndrome in adults. J Am Coll Cardiol 2007;**49**(3):329-37 doi: S0735-1097(06)02651-9 [pii]

10.1016/j.jacc.2006.08.057[published Online First: Epub Date]|.
36. Schwartz PJ, Priori SG, Dumaine R, et al. A molecular link between the sudden infant death syndrome and the long-QT syndrome. N Engl J Med 2000;**343**(4):262-7 doi: MJBA-430405 [pii][published Online First: Epub Date]|.

37. Christiansen M, Tonder N, Larsen LA, et al. Mutations in the HERG K+-ion channel: a novel link between long QT syndrome and sudden infant death syndrome. Am J Cardiol 2005;**95**(3):433-4 doi: 10.1016/j.amjcard.2004.09.054[published Online First: Epub Date]|.

University of Auckland
Standard
ACADEMIC CV

THE UNIVERSITY OF AUCKLAND
NEW ZEALAND

NAME: **Jonathan Robert Skinner**

CURRENT POSITION: **Honorary Professor**

DEPARTMENT: **Department of Paediatrics: Child and Youth Health**

FACULTY: **Medicine**

CLINICAL POSITION: **Consultant Paediatric Cardiologist/Electrophysiologist**

DEPARTMENT **Paediatric and Congenital Cardiac Services**
Starship Childrens Hospital, Auckland

EDUCATIONAL QUALIFICATIONS: [Tertiary only]

Postgraduate Career - Qualifications

2014	FHRS (Fellow of the Heart Rhythm Society)
2002	FCSANZ
2002	FRACP (Paediatrics)
1999	Vocational registration in New Zealand
1997	Cert. Instructor in Advanced Paediatric Life Support
1996	UK Accreditation in Paediatric Cardiology (Cert. by Royal Coll. Specialist Training Auth, 10th March 1997)
1994	MD (Leicester, UK)
1994	European Accreditation in Paediatrics
1985	DCH.RCP (Lond)
1985	MRCP (UK)

Undergraduate Career - Qualifications

1982	MBChB (Leicester, UK)
1976 – 1982	Medical School: Leicester University Medical School

CURRENT APPOINTMENT DATES:

2016	Appointment to Honorary Professor of Paediatrics Child and Youth Health, University of Auckland
2008	Honorary Associate Professor University of Auckland

1998 Appointment as specialist paediatric cardiologist and electrophysiologist, Greenlane paediatric and Congenital Cardiac Services
Honorary Senior Lecturer**,** University of Auckland

PREVIOUS APPOINTMENTS (Clinical):

1996 – 1997	Clinical Fellow, Electrophysiology (one year during Senior Registrar post) – St Georg Hospital, Hamburg, Germany
1994 – 1998	Senior Registrar, Paediatric cardiology (3.7 years) – Royal Hospital for Sick Children, Bristol, UK
1994	Paediatric cardiology, Oberarzt (3.5 months Invited position) – Deutches Herzzentrum, Berlin, Germany
1992 – 1994	Clinical Registrar, Paediatric cardiology (21 months) – Children's Hospital, Birmingham, UK
1992	Locum consultant, Paediatric cardiology (1 month) – University Hospital of Wales, UK
1989 – 1992	Research Fellow, Paediatric Cardiology (National Heart Research Fund) and Clinical Research Associate and Honorary Senior Registrar in Child Health (3 years and two months) – Department of Child Health, University of Newcastle upon Tyne, Newcastle, UK
1988	Locum Registrar, General and Neonatal Paediatrics (3 months) – Poole General Hospital, Dorset, UK
1986 – 1988	Registrar/Senior Registrar, Neonatology, General Paediatrics and Child Development (22 months), Mater Misericordiae Hospital, Newcastle, New South Wales, Australia
1984 – 1986	Senior House Officer (18 months), Newcastle Paediatric rotation General Paediatrics – Shotley Bridge General Hospital, Consett, UK Paediatric Cardiology – Freeman Hospital, Newcastle upon Tyne, UK Community Paediatrics – North Shields, Tyne and Wear, UK
1983 – 1984	Senior House Officer, General Paediatrics, General, Chest and Renal, General and Neurology and Neonatal Intensive Care (12 months) – Nottingham City Hospital (Teaching), Nottingham, UK
1983	House Surgeon, Orthopaedics and General Surgery (6 months) – Leicester Royal Infirmary, Leicester, UK
1982 – 1983	House Physician, Cardiology and Chest and General Medicine (6 months) – Leicester Regional Cardiothoracic Unit (Teaching), Leicester, UK

SIGNIFICANT DISTINCTIONS / AWARDS: (see also Grants/Research Funding)

2017 The Profesor Durrer visiting Professorship. University Medical Centre Amsterdam.
2015 Recipient Auckland Academic Health Alliance Award
1999 Pfizer Academic Research award, best junior UK presentation to the European Society of Paediatric Research

PROFESSIONAL SOCIETIES / SERVICE / OTHER ACTIVITIES:

New Zealand

2012– Paediatric Representative, Cardiac Society of Australia and New Zealand, NZ Branch
2011– Committee member, Child Youth Mortality Group, Northern Region
2009– Committee member, Resuscitation committee, Starship Children's Hospital
2009– Advisory Board Member, Children's Research Centre, Starship Children's Hospital
2000– Chairman and Founder of the Cardiac Inherited Disease Group for New Zealand

Australasian

2013– Chairman, Genetics Council of the Cardiac Society of Australia and New Zealand, Australian Branch
2008– Committee Member of the neonatal section of the CCPU (Certificate in Clinician Performed ultrasound), the Australasian Society for Ultrasound in Medicine (ASUM)
2005– Founder and Chairman of the Trans-Tasman Response Against sudden Death in the Young (TRAGADY)

International

2015– International Advisor – New England Inherited Heart Disease Network
2014– International Advisor – Canadian Inherited Heart Disease Network
2014– Committee member (International Representative), PACES (Pediatric and Congenital Electrophysiology Society) Research
2011– Editorial Board Member, Heart Rhythm

Membership of Learned Societies

2014 Promotion to Fellow of Heart Rhythm Society
2008– Australian Society of Ultrasound in Medicine
2005– Founder and Chairman of the Trans-Tasman Response Against sudden Death in the Young (TRAGADY)
2003– Heart Rhythm Society
2002– Council Member of Genetic Working Group of Cardiac Society of Australia and New Zealand
2000– Chair and Co-founder, Cardiac Inherited Disease Group
2000– Cardiac Society of Australia and New Zealand
2000– New Zealand Pacing and Electrophysiology Group (now Heart Rhythm New Zealand)
1998– New Zealand Paediatric Society
1998– Fellow Royal College of Paediatrics and Child Health
1993 – 95 Council Member, British Paediatric Cardiac Association
1991– British Paediatric Cardiac Association

1991– British Society of Echocardiography
1990– Neonatal Society (UK)
1990– Paediatric Research Society (UK)
1990– European Society for Paediatric Research

CLINICAL INNOVATIONS: New Services and technologies introduced to New Zealand:

2016 Introduction of zero flourosopic ablation technique to New Zealand
2014 First Implantation of Reveal linq micro-digital loop recorder
2011 Appointment of two regional CIDG coordinators establishing cardiac genetic services in Waikato and Wellington
2009 First combined genetic arrhythmia disorders clinic (with Dr Ian Hayes, clinical genetics)
2009 Multi-regional ethical approval achieved for a national inherited heart disease and sudden death registry
2008 Government funding achieved for the molecular autopsy (world first).
2008 TRAGADY (Australia and New Zealand) best practice guidelines for the investigation of young sudden death are published.
2006 Introduction of cryoablation catheter techniques to New Zealand
2005 First cardiac resynchronisation for cardiac failure in a child.
2004 Northern regional ethics approval for an inherited heart disease registry
2003 Introduction of 3D electrophysiologic mapping to the North island (Starship Foundation grant, Ensite/NavX system)
2001 Establishment of an annual paediatric cardiology visiting clinic to French Polynesia
2001 Surgical collaboration-(with Mrs K Finucane) first epicardial hybrid ICD for a child post cardiac arrest using a novel system
2000 First genetic test for long QT syndrome (collaboration with lab plus, Madhuri Hegde)
2000 First implanted digital loop recorder in a child
2000 CIDG core team is formed, first national teleconference
1999 First paediatric echocardiography course
1999 Surgical collaboration (with Mrs K Finucane)- first atrial maze procedure for intractable atrial arrhythmias post Fontan
1998 Establishment of a New Zealand paediatric arrhythmia clinic and paediatric pacing review service
1998 First radiofrequency catheter ablation of tachycardia in a child

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TEACHING: [Undergraduate Programmes, Postgraduates – number, dates, achievements, development, qualitative evidence, teaching profile, summary of evaluations, etc]

Teaching Activities

1999 – 2012 (convenor) FRACP course in cardiology, Green Lane Hospital, Auckland
1999 – 2005 Coordination of and participation in Registrar teaching Green Lane and Starship Hospitals, Auckland

1998 – 2003 5th year students Paediatric Heart Disease Green Lane Hospital, Auckland
2000 , 2003,2005 Teaching at Neonatal Nurse Practitioners' Day, Auckland twice since
2000 – 2004 Arrhythmia Workshop x2 – Paediatric update for NZ Paediatricians, Starship Hospitals, Auckland
2000 – 2003 Ward 9 Nursing Tutorials (x2), Green lane then Starship Hospital, Auckland
1999 Teaching in Paediatric Cardiology Emergencies, Green Lane Hospital, Auckland
1999 Teaching Paediatric Registrars Emergency Department Arrhythmias, Starship ED, Auckland
1999 ICR Nurses Course on arrhythmias, Auckland
1999-2001 Teaching Electrophysiology technicians, Green Lane Auckland, Setting exam questions for physiology technicians (national exam)

National and International

2014 Demonstrator/Lecturer, "The neonatal circulation", Melbourne, Australia
2010 Demonstrator/Lecturer, "Functional echocardiography in the neonate", Melbourne, Australia
2008 Demonstrator/Lecturer advanced course in Clinician Practice in Ultrasound (neonatology), Sydney, Australia
2007 Examiner: Paediatric Diploma (OSCE) University of Auckland, Auckland, New Zealand
2001 Lecturer, "Adult Congenital Heart Disease and Arrhythmia", Electrophysiology Conference for Advanced Cardiology Trainees, Christchurch, New Zealand
1999 Instructor on Advanced Paediatric Life Support Course, Wellington, New Zealand (also 2001).

Courses/Meetings Organised

2018 Convenor of the International Training day in Inherited Heart Disease. Brisbane, Australia
2016 Convenor of the International Training day in Inherited Heart Disease. Brisbane, Australia
2014 Convenor of the First International training day in cardiovascular genetics. Brisbane, Australia
2006 First meeting of Australia and New Zealand task force to prevent sudden death in the young (subsequently named TRAGADY), Sydney Australia. Numerous meetings since (8 in total).
2006 Convenor of "Echocardiography for Neonatologists" (three day course with 21 delegates), Paediatricians and Sonographers, from Australia and New Zealand, CEC Auckland City Hospital, Auckland, New Zealand
2001 Committee member for the Joint Paediatric and Adult Physician Meeting of the RACP, "Mind the Gaps", Auckland, New Zealand
2000 Co-Convenor "Echocardiography of Congenital Heart Disease", Green Lane Hospital, Auckland, New Zealand
1999 Co-Convenor "Echocardiography of Congenital Heart Disease", Green Lane Hospital, Auckland, New Zealand
1999 Convenor of "Echocardiography for Neonatologists", Green Lane Hospital, Auckland, New Zealand

1997	Convenor of "Echocardiography for Neonatologists", Bristol Royal Hospital for Sick Children, Bristol, UK
1995	Convenor of "Echocardiography for Neonatologists", Bristol Royal Hospital for Sick Children, Bristol, UK
1994	Convenor of "Echocardiography for Neonatologists", Freeman Hospital, Newcastle Upon Tyne, Newcastle, UK
1992	Convenor of "Echocardiography for Neonatologists", Freeman Hospital, Newcastle Upon Tyne, Newcastle, UK
1991	Convenor of "Echocardiography for Neonatologists", Freeman Hospital, Newcastle Upon Tyne, Newcastle, UK

Post Doctorate Fellowship supervised

Dr Annika Winbo
Establishment of pluripotential cell lines and cellular electrophysiology for long QT syndrome (With Johanna Montgomery) University of Auckland
Commenced 2015

Thesis Projects Supervised (6 doctorates, one M Phil)

Claire ODonovan (PhD, Auckland NZ)
Psychological impact of sudden death syndromes
Co-supervisor Elizabeth Broadbent
Started 2016

Kathryn Rice (PhD, Auckland, NZ)
Diastolic function in Fontan Survivors
Co-supervisors: Tom Gentles.
Started 2015

Kathryn Waddell-Smith (PhD, Auckland, NZ)
Identifying and reducing risk in familial long QT syndrome
Co-supervisors: Don Love, Ed Mitchell.
Started 2013 , awarded 2018.

Nikki Earle (PhD, Auckland, NZ))
Role of genetic modifiers in long QT syndrome
Co-Supervisors: Rob Dougherty. Andrew Shelling, Donald Love
Thesis Underway 2012 – completed end of 2014

Carey-Anne Eddy (Evans) (PhD, Auckland New Zealand)
The Role of Molecular Genetics in Sudden Death Syndromes
Co-Supervisors: Andrew Shelling
Thesis awarded 2011

Karen Searby (M Phil).

Auckland University of Technology "Epicardial pacing – A 40 year experience"
Co-Supervisor: Associate Professor Elaine Rush.
Thesis Awarded 2006

Alan Groves (MD, Auckland NZ).
"Echocardiographic prediction of poor outcome in preterm infants".
Co-Supervisors: Carl Kuschel, Jane Harding.
MD Awarded 2006

Beverley Tsai-Goodman (MD, Bristol UK). "Development of a precordial Doppler system to evaluate cardiac output continuously in the newborn". Co-Supervisors: Rob Martin.
MD Awarded 2003 Bristol UK.

RESEARCH CAREER:

Summary Statement:

Most of my research falls into two sections; the circulation in the newborn (1992-1998), and the genetics of sudden death syndromes (1998-current).

During training in neonatology, I became interested in haemodynamics. The recent development of Doppler echocardiography allowed me to be among the first to use it to study the circulation in the newborn in a way never possible previously in the human. I was fortunate to be supervised by the UKs leading paediatric echocardiologist (Stewart Hunter) and a remarkable and greatly admired neonatologist (Edmund Hey). After completing my thesis in this area I authored a textbook to guide other neonatologists how to do this in clinical practice. I however then moved from neonatology into paediatric cardiology and electrophysiology.

I realised that genetics was the fastest growing area in world medicine and that this would help us diagnose and track families with sudden death syndromes, and also possibly explain sudden unexplained death in infancy and beyond. After arriving in New Zealand in 1998 I formed an allegiance with two other clinicians (John French and Jackie Crawford), and a molecular geneticist (Mark Rees). We named ourselves the Cardiac Inherited Disease group. This framework grew nationally, crossed university/hospital interface and spread to Australia when I formed a similar Trans-Tasman group which we called TRAGADY (Trans-Tasman Research Against sudden Death in the Young). By forming allegiance with forensic pathologists and the coronial service, we were able to study sudden unexplained death in a way unique in the world. In 2006, New Zealand was the first country to have a formal investigative service for sudden deaths, initially funded by Cure Kids, this funding was taken over by the ministry of justice in 2008, and continues today with CIDG administrating. The growth of a now national inherited heart disease and sudden death registry (currently almost 2000 registrants) was paralleled by new allegiances at the university and lab Plus ((Andrew Shelling, Don Love) allowing us to study these registry populations. We were excited to be the first to discover large gene deletions causing long QT syndrome, and the first to use the neonatal screening card as a

DNA source to ascribe cause of death up to 35 years after death. We have probably the highest rate of detection of long QT in the world here in Auckland, and the model of care we have established here is being emulated elsewhere. The 3 year all cause Australia/New Zealand study of sudden death is the first research achievement for TRAGADY and was published last year in the NEJM. The registry phenotype/genotype work is just beginning, as CIDG collaborates internationally with GWAS, whole genome and gene discovery programmes, and develops pluripotential cell lines for study here in Auckland with Johanna Montgomery working closely with current leaders in cellular long QT electrophysiology in Sydney and elsewhere.

Along the way I have encouraged juniors working with me into research by publish interesting case series and clinical scenarios, audits and observations.

RESEARCH PUBLICATIONS

129 peer reviewed publications (99 original research papers , 30 reviews guidelines and editorials, letters). (H- index 33)
One book and 7 chapters.

Also listed are published abstracts, alternative presentations and media.

Original Research in Peer Reviewed Journals (93)

1. Gardner RJM, Crozier IG, Binfield AL, Love DR, Lehnert K, Gibson K, Lintott CJ, Snell RG, Jacobsen JC, Jones PP, Waddell-Smith KE, Kennedy MA, **Skinner JR**. Penetrance and expressivity of the R858H CACNA1C variant in a five-generation pedigree segregating an arrhythmogenic channelopathy. *Molecular Genetics and Genomic Medicine*. 2018 Oct 21. doi: 10.1002/mgg3.476. [Epub ahead of print]PMID: 30345660
2. Robertson SP, Hindmarsh JH, Berry S, Cameron VA, Cox MP, Dewes O, Doughty RN, Gray G, Jacobsen JC, Laurence A, Matisoo-Smith E, Morton S, Shelling AN, Sika-Paotonu D, Rolleston A, **Skinner JR**, Snell RG, Sporle A, Print C, Merriman TR, Hudson M, Wilcox P.Genomic medicine must reduce, not compound, health inequities: the case for hauora-enhancing genomic resources for New Zealand. *New Zealand Medical Journal*. 2018 Aug 17;131(1480):81-89.PMID:30116069
3. Dufendach KA, Timothy K, Ackerman MJ, Blevins B, Pflaumer A, Etheridge S, Perry J, Blom NA, Temple J, Chowdhury D, **Skinner JR**, Johnsrude C, Bratincsak A, Bos JM, Shah M. Clinical Outcomes and Modes of Death in Timothy Syndrome: A Multicenter International Study of a Rare Disorder. *JACC Clinical Electrophysiology*. 2018 Apr;4(4):459-466. doi: 10.1016/j.jacep.2017.08.007. Epub 2017 Nov 6.PMID:30067485
4. Life-Threatening Event Risk in Children With Wolff-Parkinson-White Syndrome: A Multicenter International Study. Etheridge SP, Escudero CA, Blaufox AD, Law IH, Dechert-Crooks BE, Stephenson EA, Dubin AM, Ceresnak SR, Motonaga KS, **Skinner JR**, Marcondes LD, Perry JC, Collins KK, Seslar SP, Cabrera M, Uzun O, Cannon BC, Aziz PF, Kubuš P, Tanel RE, Valdes SO, Sami S, Kertesz NJ, Maldonado J, Erickson C, Moore JP, Asakai H, Mill L, Abcede M, Spector ZZ,

Menon S, Shwayder M, Bradley DJ, Cohen MI, Sanatani S. *JACC Clinical Electrophysiology* 2018 Apr;4(4):433-444.doi:10.1016/j.jacep.2017.10.009. Epub 2017 Nov 15.PMID:30067481

5. Baruteau AE, Kyndt F, Behr ER, Vink AS, Lachaud M, Joong A, Schott JJ, Horie M, Denjoy I, Crotti L, Shimizu W, Bos JM, Stephenson EA, Wong L, Abrams DJ, Davis AM, Winbo A, Dubin AM, Sanatani S, Liberman L, Kaski JP, Rudic B, Kwok SY, Rieubland C, Tfelt-Hansen J, Van Hare GF, Guyomarc'h-Delasalle B, Blom NA, Wijeyeratne YD, Gourraud JB, Le Marec H, Ozawa J, Fressart V, Lupoglazoff JM, Dagradi F, Spazzolini C, Aiba T, Tester DJ, Zahavich LA, Beauséjour-Ladouceur V, Jadhav M, **Skinner JR**, Franciosi S, Krahn AD, Abdelsayed M, Ruben PC, Yung TC, Ackerman MJ, Wilde AA, Schwartz PJ, Probst V.SCN5A mutations in 442 neonates and children: genotype-phenotype correlation and identification of higher-risk subgroups. *European Heart Journal*. 2018 Aug 14;39(31):2879-2887. doi: 10.1093/eurheartj/ehy412.PMID: 30059973
6. Bellissima BL, Garavan F, **Skinner JR**, Tingle MD. Use of Coronial Post-mortem Tissue for Research in New Zealand. *Journal of Law in Medicine*. 2017 Nov;25(1):205-209.PMID:29978632
7. Marcondes L, Crawford J, Earle N, Smith W, Hayes I, Morrow P, Donoghue T, Graham A, Love D, **Skinner JR**; Cardiac Inherited Disease Group New Zealand. Long QT molecular autopsy in sudden unexplained death in the young (1-40 years old): lessons from and eight year experience in New Zealand. PLoS One. 2018 Apr19:13(4):e0196078. Doi: 10.1371/journal.pone.0196078. eCollection 2018. PMID: 29672598
8. Lahrouchi N, Raju H, Lodder EM, PapatheodorouE, Ware JS, Papadakis M, Tadros R, Cole D, **Skinner JR**, Crawford J, Love DR, Pua CJ, Soh BY, Bhalshankar JD, Govind R, Tfelt-Hansen J, Winkel BG, van der Werf C, Wijeyeratne YD, Mellor G, Till L, Cohen MC, Tome-Estaban M, Sjarma S, Wilde AAM, Cook SA, Bezzina CR, Shappard MN, Behr ER. Utility of Post-Mortem Genetic Testing in Cases of Sudden Arrhythmic Death Syndrome. *Journal of the American College of Cardiology*. 2017;69(17):2134-2145. Doi:10.1016/jacc.2017.02.046 PMID 28449774
9. Leong IUS, Dryland PA, Prosser DO, Lai SW, Graham M, Stiles M, Crawford J, **Skinner JR**, Love DR. Splice Site Variants in the *KCNQ1* and *SCN5A* Genes: Transcript Analysis as a Tool in Supporting Pathogenicity. *J Clin Med Res*. 2017 Aug;9(8):709-718. doi: 10.14740/jocmr2894w. Epub 2017 Jul 1. PMID: 28725320
10. Leong IUS, Stuckey A, Belluoccio D, Fan V, **Skinner JR**, Prosser DO, Love DR. Massively Parallel Sequencing of Genes Implicated in Heritable Cardiac Disorders: A Strategy for a Small Diagnostic Laboratory. *Med Sci (Basel).* 2017 Oct 10;5(4). pii: E22. doi: 10.3390/medsci5040022. PMID:29099038
11. **Skinner JR.** Detection of pneumothorax in the preterm infant with ultrasound. *Journal of Paediatrics and Child Health*. 2016:52(12):1122. Doi: 10.1111/jpc.13362 PMID 27989007
12. Earle N, Crawford J, Gibson K, Love D, Hayes I, Neas K, Stiles M, Graham M, Donoghue T, Aitken A, **Skinner JR**. Detection of sudden death syndromes in New Zealand. *New Zealand Medical Journal* 2016:129(1445): 67-74
13. Waddell-Smith KE, Smith W, Crawford J, **Skinner JR**. Beta-Blocker adherence in familial long QT syndrome. *Circulation : Arrhythmia and Electrophysiology* 2016:9 doi: 10.1161/CIRCEP.115.003591

14. Bagnall RD, Weintraub RG, Ingles J, Duflou J, Yeates L, Lien Lam, Ph.D., Davis A, Thompson T, Connell V, Wallace J, Naylor C, Crawford J, Love DR, Hallam L, White J, Lawrence C, Lynch M, Morgan N, James P, du Sart D, Puranik R, Langlois N, Vohra J, Winship I, Atherton J, McGaughran J, **Skinner JR**, Semsarian C. A Prospective Study of Sudden Cardiac Death in Children and Young Adults. *New England Journal of Medicine* 2016;374:2441-52
15. Waddell-Smith KE, Donoghue T, Oates S, Graham A, Crawford J, Stiles MK, Aitken A, **Skinner JR**. Inpatient detection of cardiac-inherited disease: the impact of improving family history taking. *Open Heart*. 2016 Feb 16;3(1):e000329. doi: 10.1136/openhrt-2015-000329. PMID: 26925241
16. Martin A, Crawford J, **Skinner JR**, Smith W. High Arrhythmic Burden but Low Mortality during Long-term Follow-up in Arrhythmogenic Right Ventricular Cardiomyopathy. *Heart Lung and Circulation*. 2016 Mar;25(3):275-81. doi: 10.1016/j.hlc.2015.08.019. Epub 2015 Sep 28.PMID: 26546095
17. Earle N, Ingles J, Bagnall RD, Gray B, Crawford J, Smith W, Shelling AN, Love DR, Semsarian C, **Skinner JR**.NOS1AP polymorphism alter QTc interval duration but not cardiac arrest risk in hypertrophic cardiomyopathy. *Journal of Cardiovascular Electrophysiology*. 2015 Dec;26(12):1346-51 2015 PMID:26332198
18. Waddell-Smith KE, Ertresvaag KN, Li J, Chaudhuri K, Crawford JR, Hamill JK, Haydock D, **Skinner JR**; Cardiac Inherited Disease Group New Zealand. Physical and psychological consequences of left cardiac sympathetic denervation for long QT syndrome and catecholaminergic polymorphic ventricular tachycardia. *Circulation Arrhythmia and Electrophysiology*. 2015 Jul 29. PMID;26224781
19. Leong IU, Stuckey A, Lai D, **Skinner JR**, Love DR. Assessment of the predictive accuracy of five in silico prediction tools, alone or in combination, and two metaservers to classify long QT syndrome mutations. *BMC Medical Genetics* 16(1); 34, 2015.
20. Herath VC, Gentles TL, **Skinner JR**. Dilated cardiomyopathy in children: Review of all presentations to a children's hospital over a 5-year period and the impact of family cardiac screening. *Journal of Paediatrics and Child Health* 51(6); 595-599, 2015.
21. Earle N, Poppe K, Pilbrow A, Shelling AN, Cameron V, Troughton R, Whalley G, Love DR, **Skinner JR**, Doughty R, Richardson AM. Genetic markers of repolarization and arrhythmic events following acute coronary syndromes. *American Heart Journal* 169
R(4); 579-586.e3, 2015.
22. Leong IUS, **Skinner JR** and Love DR. Application of massively parallel sequencing in the clinical diagnostic testing of inherited cardiac conditions. *Medical Sciences* 2(2); 98-126, 2014.
23. Leong IU, Sucich J, Prosser D, **Skinner JR**, Crawford J, Higgins C, Love D. Array comparative genetic hybridization identifies a heterozygous deletion of exon 3 of the RyR2 gene. *Upsala Journal of Medical Sciences*, 120:3,190-197, 2015. http://www.tandfonline.com/doi/full/10.3109/03009734.2015.1029101, 2015.
24. Glengarry J, Crawford J, Morrow PL, Stables SR, Love DR, **Skinner JR**. Long QT molecular autopsy in sudden infant death syndrome. *Archives of Disease in Childhood* 99(7); 635-640, 2014.

25. **Skinner JR**, Marquis-Nicholson RM, Luangpraseuth A, Cutfield R, Crawford J, Love DR. Diabetic Dead-in-Bed syndrome; a possible link to a cardiac ion channelopathy. *Case Reports in Medicine* 2014(Article ID 647252); 5 pages, 2014.
26. Leong IU, **Skinner JR**, Shelling AN, Love DR. Expression of a mutant KCNJ2 Gene Transcript in Zebrafish. *ISRN Molecular Biology* 2014(Article ID 324839); 14 pages, 2014. http://dx.doi.org/10.1155/2014/324839
27. Marquis-Nicholson R, Prosser DO, Lowe JM, Zhang L, George AM, Crawford JR, **Skinner JR**, Love DR. Array comparative hybridization identifies a heterozygous deletion of the entire KCNJ2 gene as a cause of sudden cardiac death. *Circulation Cardiovascular Genetics* 7(1); 17-22 2014.
28. Baskin B, **Skinner JR**, Sanatani S, Terespolsky D, Krahn AD, Ray PN, Cherer SW, Hamilton RM. TMEM43 mutations associated with arrhythmogenic right ventricular cardiomyopathy in non-Newfoundland populations. *Human Genetics* 132(11); 1245-1252, 2013.
29. **Skinner JR**, Waddell-Smith K, Anderson BJ. Reply to the Editor: Amiloride concentrations in clinical practice. *Heart Rhythm* 10(11); e82-e83, 2013.
30. Earle N, Yeo Han D, Pilbrow A, Crawford J, Smith W, Shelling AN, Cameron V, Love DR, **Skinner JR**. Single nucleotide polymorphisms modify the risk of cardiac events and sudden death in long QT syndrome. *Heart Rhythm* 11(1); 76-82, 2014.
31. de Al Meida T, Pennock V, **Skinner JR** Medical Image: Reflex anoxic seizures in a toddler. *New Zealand Medical Journal* 126(1369); 83-85, 2013.
32. Earle N, Crawford J, Smith W, Hayes I, Shelling A, Hood M, Stiles M, Maxwell F, Heaven D, Love DR, **Skinner JR**. Community detection of long QT syndrome with a clinical registry: An alternative to ECG screening programs? *Heart Rhythm* 10(2); 233-238, 2013.
33. Yeong M, Rumball E, Sinclair S, **Skinner JR**. Emergency cardiac resynchronisation in a 4kg infant post-surgical closure of ventricular septal defect. *Heart Lung and Circulation* 22; 317-319, 2013.
34. Wilms H, Midgley D, Morrow P, Stables S, Crawford J, **Skinner JR**. Evaluation of autopsy and police reports in the investigation of sudden unexplained death in the young. *Forensic Science, Medicine, and Pathology* 8(4); 380-389, 2012.
35. Leong IU, Lan CC, **Skinner JR**, Shelling AN, Love DR. In vivo testing of microRNA-mediated gene knockdown in zebra fish. *Journal of Biomedicine & Biotechnology* 2012(Article ID 350352); 7 pages, 2012.
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Media

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- 11 may 2017 Radio New Zealand Interview regarding a Samoan Patient's desire to raise awareness of long Qt syndrome among Polynesians.
- 5 May 2017 Lead feature article in New Zealand Herald regarding a good outcome in child after 2 weeks on ECMO
- 6 July 2016 Interview with WTV (Chinese TV) regarding the NEJM article on SCD
- 27 June 2016 Interview with news Hub (featured on TV3 news) regarding the NEJM article on SCD http://www.newshub.co.nz/nznews/genetic-link-to-sudden-cardiac-deaths-2016062717#axzz4ClBGBsdq
- 17th June 2016. TV One News feature on a court case of a young school student who died suddenly in an altercation
- 12th November 2015 Interview with Fairfax media, featuring in Stuff, and NZ Herald feature on the sudden death of Laurent Vidal, a high end athlete who died in his bed. http://www.stuff.co.nz/national/health/73918658/cardiac-arrests-mostly-kill-young-people-in-their-sleep-or-doing-daily-tasks

- 7th December 2014
  A feature by Tony Wall on a young man of 24 who died in his sleep. Interview with myself around the key issues regarding sudden death in the young, its causes, and its investigation. In Stuff on line, and in the Star on Sunday.
  http://www.stuff.co.nz/national/health/63873750/Mother-wants-answers-over-sons-death
- 6th December 2014
  Seven Sharp, TV One. Interview with myself regarding the frequency of sudden death in the young, and its causes. Signs to watch for in the family etc. Story was based around two young men who were resuscitated during sports activity.
- 27th February 2014
  "Tackling unexpected and unexplained sudden death in 1-35 year olds". A 15 minute interview. Katherine Ryan interviews Jon Skinner on the Nine to Noon programme on Radio New Zealand National.
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  "Clinical genetic LQT testing now available in NZ" - The TV3 news.
- May 2006

TV One News, Christchurch Press, Sunday Herald. All featured our recent publication on "Sibutamine and risk of cardiac arrest". I was interviewed by each of the above media group.

- October 2004
  TVNZ, "Documentary NZ" - Interview with Jude Dobson re sudden infant death syndrome screened May 2005.
- Saturday/Monday 18 October 2004
  Radio New Zealand "Health Matters" - Broadcast Interview with Ms Wallace re Sudden death in the young.

Medico Political

- 17th May 2007 - Wellington.
  Presentation to the Child Youth Mortality Meeting re data collation for young sudden death.

- 2008 – Wellington/Auckland.
  Multiple meetings with Moh and LECG (consultancy firm employed by MoH) regarding the NSTR (National Services and Technology review) of Cardiac Genetics services and registry for New Zealand.

- June 2010 - Auckland.
  Presentation to chair of DHBNZ regarding development of inherited heart disease services for New Zealand.

- February 2015 - Auckland.
  Presentation to New Zealand Coroners regarding refinement of services for sudden death victims in New Zealand.

Published Abstracts

1. Waddell-Smith K, Ertresvaag KN, Li J, Crawford J, Hamil J, Haydock D, **Skinner JR**. Physical and psychological consequences of left cardiac sympathetic denervation in inherited heart diseases. Heart Rhythm 12(5) Suppl 2; PO05-150, 2015.
2. Skinner J, Crawford J, Waddell-Smith K, Earle N, Donoghue T, Graham M, et al. Development of the New Zealand cardiac inherited disease registry. Heart Lung and Circulation 23(Suppl 2); e4, 2014.
3. Earle N, Smith W, Crawford J, Love D, Hayes I, Graham M, Skinner J. The prevalence of emerging genotypic risk factors in patients with long QT syndrome. Heart Lung and Circulation 23(Suppl 2); e9, 2014.
4. Earle N, Poppe K, Cameron V, Pilbrow A, Troughton R, Skinner J, et al. Risk of sudden cardiac death following acute coronary syndromes: Interplay of single nucleotide polymorphisms and the presence of heart failure. Heart Lung and Circulation 23(Suppl 2); e9, 2014.
5. Marcondes L, Crawford J, Earle N, Smith W, Donoghue T, Love D, Skinner J. Long QT gene testing in autopsy-negative sudden death in the young: Influence of age and gender on diagnostic yield. Heart Lung and Circulation 23(Suppl 2); e13-4, 2014.
6. Waddell-Smith K, Li J, Hinds H, Crawford J, Skinner J. Identification of high risk long QT syndrome patients using mean Holter QTc. Heart Lung and Circulation 23(Suppl 2); e18, 2014.
7. Waddell-Smith K, Donoghue T, Graham M, Oates S, Crawford J, Li J, Skinner J. The inpatient cardiology visit: Missing the opportunity to detect inherited heart conditions. Heart Lung and Circulation 23(Suppl 2); e18-9, 2014.
8. Earle N, Crawford J, Smith W, Love DR, Hayes I, Shelling A, Hood M, **Skinner JR**. Community detection of long QT syndrome with a clinical registry: A practical alternative to ECG screening programs. Circulation 126; A11218, 2012.
9. Watanabe H, Yang T, Atack T, Hasdemir C, Crawford J, **Skinner JR**, Roden D. Divergent biophysical effects of cardiac sodium channel mutations in the N-terminus. Heart Rhythm 6(5 Suppl); S-354: PO05-73, 2009.

10. **Skinner JR**, Crawford J, Vaughan A, Gladding P, Eddy C-A, Love D, Rees M, Shelling A. Posthumous diagnosis of long QT syndrome from the neonatal screening card. Heart Lung and Circulation 18(Suppl 3); S82, 2009.
11. **Skinner JR**, Eddy CA, MacCormick J, Chung SK, Crawford JR, Love DR, Rees MI, Shelling AN. Identification of large gene deletions and duplications in patients with long QT syndrome. Heart Rhythm 5 (5 Suppl); S-75: AB36-2, 2008.
12. Long S, Fong D, Webb R, Gentles T, **Skinner JR**, O'Donnell C, Hornung T, Wilson, N. Echocardiographic assessment of early changes of left heart function in patients receiving prednisolone for acute rheumatic fever. Heart Lung and Circulation 16(Suppl 2); S219, 2007.
13. Al-Salama T, West T, Riddell F, Searby K, Kerr A, Finucane K, **Skinner JR**. Outcome for children receiving a pacemaker implantation at Green Lane Hospital over a 40 year period. Heart Lung and Circulation 14(Suppl 1); S141, 2005.
14. **Skinner JR**, McCulley C, Crawford J, Chung S-K, McAllister H, French J, Shelling AN, Rees MI. Spectrum of mutations in long-QT genes in New Zealand long-QT syndrome cases. Heart Lung and Circulation 13(Suppl 2); S89, 2004.
15. Maher LM, Jones , Smith RF, Tripp I, Smith W, **Skinner JR**. Accuracy and repeatability of algorithms to predict Accessory pathway location in children with WPW syndrome. Heart Lung and Circulation 13(Suppl 2); S23, 2004.
16. Borthwick H, Bhala C, McCulley C, Crawford J, Chung S-K, French J, Rees MI, **Skinner JR**. 24 hour beat to beat QT analysis in genetically characterised families with long-QT syndrome. Heart Lung and Circulation 13(Suppl 2); S87, 2004.
17. **Skinner JR**, Chung SK, Crawford J, French J, Rees MI. SCN5A Mutations – A link to febrile convulsions and sudden infant death. Heart Lung and Circulation 13(Suppl 2); S40, 2004.
18. Allchorne L, Smith W, Hood M, Skinner J, Heaven D. WPW with prognostically significant conduction. PACE 26(Part II); S106: 423, 2003.
19. Borthwick H, **Skinner JR**. 24 hour beat to beat measurement of the QT interval. Journal of the American College of Cardiology 39(Suppl 2), 2359, 2002.
20. Wilson N, **Skinner JR**, Gentles T. Late closure of Fontan fenestrations. Cardiology in the Young 11(Supp 1); 2001.
21. Searancke K, Riddell F, Kerr A, Finucane K, **Skinner JR**. Medium term follow-up comparison of steroid and non-steroid eluting epicardial pacing leads. Cardiology in the Young 11(Supp 1); 52: P238, 2001.
22. Shanahan CL, Wilson N, Gentles T, **Skinner JR**. Are estimated indices of pulmonary vascular resistance (PVR) reliable in pre-Fontan assessment in patients with a bidirectional Glenn. Cardiology in the Young 11(Supp 1); 184: P489, 2001.
23. Finucane K, Setty SP, Kerr AR, **Skinner JR**. Conversion to an extracardiac conduit with a limited right atrial maze procedure for the failing Fontan with atrial tachycardias. Cardiology in the Young 11(Supp 1); 113: P137, 2001.
24. **Skinner JR**, Wakley CM, Hayes A, Joffe H, Martin RP. Difficulty and inequality in the acquisition of life insurance amongst adults with congenital heart disease. European Heart Journal 18(Supplement): P2304, 1997.
25. Wakley CM, **Skinner JR**, Hayes A, Joffe H, Martin RP. Health awareness in adults with congenital heart disease. In: Y Imai, K Momma (Eds.) Proceedings of the Second World Congress of Paediatric Cardiology and Cardiac Surgery. Futura Publishing Co, Armonk (NY); 1209 p, Abstract 269N, 1997.

26. **Skinner JR**, Wakley CM, Hayes A, Joffe H, Martin RP. Acquisition of life insurance amongst adults with congenital heart disease. In: Y Imai, K Momma (Eds.) Proceedings of the Second World Congress of Paediatric Cardiology and Cardiac Surgery. Futura Publishing Co, Armonk (NY); 1209 p, Abstract P554, 1997.
27. Tsai-Goodman B, **Skinner JR**, Whittingham A, Halliwell M, Marlow N, Martin RP. Continuous recording of Doppler echocardiographic signals in the newborn. In: Y Imai, K Momma (Eds.) Proceedings of the Second World Congress of Paediatric Cardiology and Cardiac Surgery. Futura Publishing Co, Armonk (NY); 1209 p, Abstract P169, 1997.
28. Hebe J, Siebels J, **Skinner JR**, Volkmer M, Ouyang F, Kuck KH. Atrial reentry tachycardias after surgery for congenital heart disease: treatment using radiofrequency current. In: Y Imai, K Momma (Eds.) Proceedings of the Second World Congress of Paediatric Cardiology and Cardiac Surgery. Futura Publishing Co, Armonk (NY); 1209 p, Abstract 195, 1997.
29. **Skinner JR**, Wakley CM, Hayes A, Joffe H, Martin RP. Knowledge of diagnosis amongst adults with congenital heart disease. In: Y Imai, K Momma (Eds.) Proceedings of the Second World Congress of Paediatric Cardiology and Cardiac Surgery. Futura Publishing Co, Armonk (NY); 1209 p, Abstract 389, 1997.
30. **Skinner JR**, Hebe J, Volkmer M, Ouyang F, Braun E, Cappato R, Siebels J, Kuck K-H. The influence of Amiodarone on the isthmus between the inferior vena cava and the tricuspid annulus in atrial flutter. PACE 20(4, part II); 278, 1997.
31. Hebe J, **Skinner JR**, Siebels J, Braun E, Kuck K-H. Arrhythmogenic substrates and outcome of radiofrequency current ablation in children with and without congenital heart disease. PACE 20(4, part II); 297, 1997.
32. **Skinner JR**, Hebe J, Volkmer M, Ouyang F, Braun E, Cappato R, Siebels J, Kuck KH. Einfluss von Amiodaron aund den Isthmus zwischen V.cava Inf. und Trikuspidklappenanulus bei Patienten mit Vorhofflattern. Zeitschrift für Kardiologie 86(Supp 2); A6-385: 163, 1997.
33. **Skinner JR**, Martin RP, Hayes A, Joffe H, Jordan SC. Early mortality in infants with hypertrophic cardiomyopathy 1969-1993. Cardiology in the Young 5(Supp 1); 45, 1995.
34. **Skinner JR**, Sreeram N, Wright JG, DeGiovanni J, Silove ED, Brawn W, Sethia B. Acute failure of the Fontan operation: Indications and outcome of early intervention. Zeitschrift für Kardiologie 83(Suppl 1); 1-243: 41, 1994.
35. Skinner J, Hunter S, Hey EN. Low left ventricular output predicts fatal outcome in babies with persistent transitional circulation. Pediatric Research 35; 278: 117, 1994.
36. Skinner J, Hunter S, Hey EN. Pulmonary arterial pressure in the newborn and the time to peak velocity/right ventricular ejection time ratio - influence of gestation and ductal patency. Cardiology in the Young 3(Supp 1); 410, 1993.
37. Stuart AG, MacIver DH, **Skinner JR**, Sethia B, Brawn WJ, DeGiovanni JV, Wright JGC, Silove ED. Balloon angioplasty of aortic constriction after repair of interrupted aortic arch. Cardiology in the Young 3(Supp 1); 257, 1993.
38. **Skinner JR**, Adwani S, Wright JGC, DeGiovanni JV, Silove ED, Brawn WJ, Sethia B. Echocardiographic assessment of right ventricular area in pre-operative assessment of pulmonary atresia and intact ventricular septum. Cardiology in the Young 3(Supp 1); 460, 1993.
39. Skinner J, Hunter S, Hey EN. Evaluation of the TPV/RVET ratio in determining pulmonary arterial pressure in the neonate. Pediatric Research 32; 614, 1992.

40. **Skinner JR**, Stuart AGS, O'Sullivan J, Hunter S. Validation of right heart pressure determination by Doppler in infants with tricuspid regurgitation. Paediatric Cardiology 13(4); 272-3, 1992.
41. Morris KP, **Skinner JR**, Wren C, Hunter S, Coulthard M. Erythropoietin improves cardiac function in children with end-stage renal failure. Pediatric Nephrology 5; C51, 1991.
42. Skinner J, Boys RJ, Hunter S, Hey EN. Non-invasive determination of pulmonary arterial pressure in healthy neonates. Pediatrics Digest 2; 4-5: (Invited abstract), 1992.
43. **Skinner JR**, Hunter S, Hey EN. Regulation of cardiac output in the premature neonate: stroke volume or heart rate? Pediatric Cardiology 12(4); 258, 1991.
44. **Skinner JR**, Hunter S, Hey EN. A new method to assess ductal shunting. Paediatric Review Community 6; 58-59, 1991.
45. **Skinner JR**, Hunter S, Hey EN. Cardiac Output in the Premature Neonate: The Influence of Ductus Arteriosus. British Journal of Radiology 64; 660, 1991.
46. **Skinner JR**, Hunter S, Hey EN. Cardiorespiratory collapse and the silent ductus. Klinische Padiatrie 203; 52, 1991.
47. **Skinner JR**, Hunter S, Hey EN. Pulmonary artery pressure in hyaline membrane disease. Pediatric Research 28; 286: 58, 1990.
48. **Skinner JR**, Hunter S, Hey EN. Tricuspid regurgitation and pulmonary artery pressure in hyaline membrane disease. Early Human Development 24(2); 169, 1990.
49. **Skinner JR**, Hunter S, Hey EN. Tricuspid regurgitation in the newborn: Doppler estimation of pulmonary artery pressure. Early Human Development 22(2); 108-109, 1990.

RESEARCH GRANTS AND FUNDING

2018 Green Lane Fund Limited Budget grant (co PI)
NZ$20,000 "Familial DCM among the transplant population" Peter Ruygrok, Jon Skinner

2017 Hugh Green Foundation (AI)
NZ$300,000 research Fellowship for Annika Winbo (PI)), research into the cellular basis of long QT syndrome using human pluripotential cells.

2017 SJ Taylor Fund, School of Medicine, University of Auckland (AI)
NZ$285,952 salary and working expenses for Annika Winbo (PI)), research into the cellular basis of long QT syndrome using human pluripotential cells.

2016 Green Lane Fund Limited Budget grant (co PI)
NZ$20,000 16/28/4123 "A whole genome pedigree based gene and mutation discovery study". Kathryn Waddell-Smith, Jon Skinner

2015 Auckland Academic Health Alliance (co PI)
NZ$99,860 "Joining of hearts and minds to address the cellular basis of Long QT Syndrome" modelling a sympathetically triggered disorder in cardiomyocytes derived from patient-induced pluripotent stem cells. Joint PI with Johanna Montgomery, with assistant investigator Annika Winbo.

2015 TM Hoskins Trust (co PI)
NZ$20,000 "Salary for lab technician and genetic testing to be carried out on families who have lost a loved one to an inherited heart condition." PI Application with Don Love, for cascade testing in families of SUD victims with a novel gene found to have caused the death.

2015 Green Lane Fund Limited Budget grant (co PI)
NZ$19,150 To facilitate training in cellular cardiac electrophysiology, specifically acquiring skills regarding performing quality recordings (patching and multielectrode array) from beating cardiac myocytes derived from patient- induced pluripotent stem cells (iPS cells). *Supervisor to successful applicant, Annika Winbo.*

2014 New Zealand Medical and Scientific Research Grant 2014 $20,000 (co PI)-"QT and continuous glucose monitoring study", with Dr Craig Jefferies

2014 Green Lane Fund (PI)
NZ$98,000 Third year inherited Heart Disease Fellowship, Kathryn Waddell Smith. Study of adherence and environmental influences in long QT syndrome. *Successful applicant as PI.*

2014 Cure Kids (T.H. Hosking Charitable Trust) (PI)
NZ$39,200 – "Holter recordings in children and adolescents with long QT syndrome".

2014 National Heart Foundation Grant-in-aid (co PI)
NZ$15,000 – Equipment purchase to enable whole exome sequencing for genetic causes of long QT syndrome (Ivone Leong, Don Love).

2014 New Zealand Medical and Scientific Research Grant 2014
NZ$20,000 – "QT and continuous glucose monitoring study", with Dr Craig Jefferies.

2013 Green Lane Fund – Grant 13/60/4100
NZ$48,882 – "Project support application for 'Targeted Next Generation Sequencing to identify the genetic causes of heritable cardiac disorders" September 2013 (Jon Skinner, Don Love).

2012 National Heart Foundation
Fellowship in Inherited Heart Disease. Successful applicant as supervisor to establish this competitive post. April 2013 for two years. (Supervisor of Kathryn Waddell- Smith)

2012 National Heart Foundation – Grant 1508
NZ$126,048 – "Mutation screening of cardiac genes in SUDY and SIDS" (Co-investigator: PI A/Prof Don Love, Nikki Earle, Andrew Shelling).

2011 AMRF – Grant 1/211/002
NZ $122,000 – 3 year Doctoral scholarship Dr Nikki Earle. "The role of SNPs in long QT genes in sudden cardiac death". (Supervisor of Dr Nikki Earle).

2009 NHMRC (Australia) – Grant 632575

AUD$658,500 – "Investigation of Sudden Cardiac Death in the Young" (Co-investigator: PI Ass. Prof Chris Semsarian, Agnes Ginges Centre, Sydney, NSW; other CI's, Dr Robert Weintraub, Melbourne; Dr Raj Puranik, Sydney; Ass Prof Jo duFlou, Sydney)

2008 Cure Kids
Four year commitment to 20% FTE support for Dr Jon Skinner as Chair of CIDG.

2008 National Heart Foundation – Grant 1347
NZ$15,000 – "Do large deletions and/or duplications in LQTS genes cause sudden death in young New Zealanders" (Carey Anne Eddy, Andrew Shelling, Jackie Crawford Jon Skinner)

2005/2007 – Cure Kids/Todd Foundation/ASB Trust
NZ$1,115,000 – A combination of three grant applications which have been combined into one under Cure Kids. "Two year prospective national testing of LQT genes on Sudden Death victims in New Zealand" (Jon Skinner, Andrew Shelling, Mark Rees, Carey Nel, Jackie Crawford)

2005 Green Lane Research and Education Fund
NZ$47,500 – "Ryanodine mutations in Sudden Death victims" (Jon Skinner, Andrew Shelling, Mark Rees, Carey Nel, Jackie Crawford)

2005 Medtronic Special Bursary
NZ$130,000 - Two year salary support for Inherited Cardiac Disease Coordinator. Auckland, New Zealand

2005 National Heart Foundation – Grant 1119
NZ$15,000 – "Economic evaluation of molecular screening for SADS" (Jon Skinner, Richard Milne, Jackie Crawford, Andrew Shelling)

2005 Southern Trust
NZ$65,000 – Supervisor of Dr Judith MacCormick. Fellowship researching the Phenotype/genotype correlations in long-QT syndrome.

2005 Green Lane Research and Education Fund
NZ$19,500 – "The mutation analysis of SCN5A in 25 Sudden Infant Death Syndrome cases" (Jon Skinner, Andrew Shelling, Mark Rees, Carey Nel, Jackie Crawford)

2003 Southern Trust
NZ$95,000 – Co-Supervisor of Dr Alan Groves. Fellowship researching the use of Doppler echocardiographic techniques in assessment of cerebral and systemic blood flow in neonates. (Alan Groves, David Knight, Carl Kuschel, Jon Skinner).

2003 Child Health Research Foundation
NZ$90,000 – "Genetic analysis of long-QT Syndrome in New Zealand and their relation to sudden unexpected death syndromes". Collaborative study with University of Auckland, Child Health and Molecular Biology Departments. (Mark Rees, John French, Jon Skinner, Jackie Crawford)

2001 Green Lane Research Fund
NZ$5, 000 – Grant to Subsidise Reagents for LQT gene testing in SIDs (SCN5A)

2001 Cot Death Association
NZ$30, 000 – "Association of SIDS with LQTs". Collaborative Study with University of Auckland, Child Health and Molecular Biology Departments

2001 Lion Foundation
NZ$90,000 – Development of an Inherited Cardiac Disease database

2000 Green Lane Research Fund
NZ$5, 000 – Seeding grant to establish Long QT Registry for Patients in Auckland Region

1996 Sir Jules Thorn Charitable Trust
£78,000 – Project grant for two and a half years (1996-98) – Principle Applicant as Supervisor in Bristol. "Developing a new method for the continuous non-invasive measurement of pulmonary arterial pressure and cardiac output in the newborn".

1991 Pfizer Academic Research Award
£450 – Regarding work presented to The European Society of Paediatric Research

1990 National Heart Research Fund (UK)
£49,984 – Project Grant for two years (Non-invasive determination of pulmonary arterial pressure in the newborn).

1989-90 Local Scientific and Research Committee of Newcastle Upon Tyne
£21,145 – 14 Months Project Grant

Invited Lectures (since 1999).
143; 36 at international meetings outside Australia and New Zealand.

1. " The New Zealand Cardiac Inherited Disease Registry; what we have learnt and what we can do better"-presented to the Southern Regional Cardiology Forum, Wellington.NZ.
2. "A quiz on congenital heart disease"- presented to the physicians and paediatricians , Papeete, Tahiti , October 2018
3. "Catecholaminergic polymorphic VT: Flecainide, ICD, both or neither?" Cardiac Society Australia/New Zealand. Brisbane, August 2018
4. "Sudden death in the young- an overview" Cardiac Society Australia/New Zealand. Brisbane, August 2018
5. "What's hot in cardiac genetics" Cardiac Society Australia/New Zealand. Brisbane, August 2018
6. "The humble ECG in inherited heart disease: Tips and Tricks" Cardiac Society Australia/New Zealand. Brisbane, August 2018
7. "Just another pathway" Case presentation at specialist cardiac electrophysiology symposium, (pre Cardiac Society Australia/New Zealand). Brisbane, August 2018

8. "The New Zealand experience of long QT genetic testing" International Cardiovascular Genetics Conference. Brisbane May 24 2018.
9. "Cardiac Ion Channelopathies" Education day of International Cardiovascular Genetics Conference. Brisbane May 23 2018.
10. "The investigation of SUDY. Act locally think globally" Ted-style talk opening the first paediatric session at Heart Rhythm, Boston May 2018.
11. "Arrhythmias-for the paediatric anaesthetists" Lecture to paed anaesthesia dept Starship Childrens Hospital April 9 2018
12. "Investigation after sudden cardiac death" to Australia and New Zealand Intensive care society Auckland, March 2018.
13. "The Cardiac Inherited Disease registry" Public lecture, and hosting multi speaker event Auckland City Hospital November 23 2017..
14. " Cardiac Inherited Diseases" Lecture to trainee adult cardiology registrars November 21 2017
15. " Investigation of resuscitated Sudden Cardiac Death" To Middlemore Hopsital ICU and Cardiology services November 17 2017
16. "This is your life" –stylistic presentation regarding Dr. Warren Smith and the history of EP in New Zealand. Auckland City Hospital Grand Round November 15 2017
17. "Paediatric Cardiology –lessons for the general practitioner" Heart Group GP symposium August 2017
18. "I don't have to blame myself anymore". Inaugural professorial lecture, University of Auckland, August, 2017.
19. "Ten years of a molecular autopsy service". Presented to the cardiac Genetic Service, Stanford University, California USA, August 2017
20. "QT drugs, Whats the problem and when to worry?" World Congress of the Paediatric and Congenital Cardiac Society, Barcelona, July 2017.
21. "Ten years of a molecular autopsy service". Presented to Cardiologists and research scientists in experimental cardiology at the Academic medical centre Amsterdam. July 2017
22. "Sudden death in Athletes". Presentation to Tahitian Paediatrics/Cardiology. Papeete, Tahiti, September 2016
23. " Sudden Cardiac death in Australia and New Zealand" Forum, Paediatric and Congenital Cardiac Services, Starship Childrens Hospital, September 2016.
24. "Cardiac Ion channelopathies and sudden death: ICDs, risk stratification and sport" Cardiac Society Australia/New Zealand. Adelaide, New Zealand. June 2015
25. "Whats new in the diagnosis and management of long QT syndrome?" International Cardiovascular Genetics Conference, Brisbane, Australia. May 2016
26. "Cardiac Ion Channelopathies" Educational symposium at ?" International Cardiovascular Genetics Conference, Brisbane, Australia. May 2016
27. "Aggressive cascade screening is the better way to detect Inherited Heart Conditions (Hyde Park Corner presentation) Heart Rhythm, San Fransisco, USA, May 2016
28. " Which 3D mapping tools do we need to ablate complex atrial arrhythmias?" Heart Rhythm, San Fransisco, USA, May 2016
29. "Designing the sudden Death Investigation" Heart Rhythm, San Fransisco, USA, May 2016
30. "Heart Rhythms. What is the Research telling us?". Heart Kidz Conference. Auckland, New Zealand. September 2015

31. "Inherited Heart Diseases – so who is dying suddenly?" Cardiac Society Australia/New Zealand, New Zealand Meeting. Auckland, New Zealand. June 2015.
32. "Should we be screening for inherited heart Diseases?" Cardiac Society Australia/New Zealand, New Zealand Meeting. Auckland, New Zealand. June 2015.
33. "Sudden Death in the Very Young". Paediatric Update (National NZ, video conference). Auckland, New Zealand. June 2015.
34. "Early repolarisation; when do I worry?" Heart Rhythm. Boston, USA. May 2015.
35. "SIDS and the long QT phenotype". Heart Rhythm. Boston, USA. May 2015.
36. "Investigation after sudden cardiac death". Australia and New Zealand Intensive Care Society, Auckland, October, 2015.
37. "Left Cardiac Sympathetic Denervation: which side of me do you like the best?" Greenlane Scientific Sessions, Auckland City Hospital, October 2015
38. "Clinical profile of long QT type 2". Cardiac Physiome Workshop (International), Viaduct Events Centre. Auckland, New Zealand. April 2015.
39. "Progress in the prevention of young sudden death". To Paediatricians and Cardiologists in Papeete. Papeete, Tahiti. September 2014.
40. "TRAGADY and CIDG". Monash Cardiovascular Symposium. Melbourne, Australia. September 2014.
41. "Infant ECG screening for long QT syndrome" Heart Rhythm NZ National Meeting. Auckland, New Zealand. September 2014.
42. "CPVT: A clinical Perspective". Joint meeting of the International Cardiovascular Genetics Conference and IUPAB (International Union of Pure and Applied Biophysics). Brisbane, Australia. August 2014.
43. "Genetics of long QT syndrome: what's new?" International Cardiovascular Genetics Conference. Brisbane, Australia. August 2014.
44. "Genetics of Cardiac Ion Channelopathies." Educational symposium for the International Cardiovascular Genetics Conference. Brisbane, Australia. August 2014.
45. "Paediatric electrophysiological and ablation." Post graduate Paediatric Cardiac Nursing course. Auckland, New Zealand. July 2014.
46. "Genetics of Brugada Syndrome". Cardiac Society of Australia and New Zealand. New Zealand Meeting. Dunedin, New Zealand. June 2014.
47. "Development of the Cardiac Inherited disease Service". Presented to Cardiologists, Allied Professional and Trainees. Wellington Hospital. Wellington, New Zealand. June 2014.
48. "Long QT syndrome in Children (They are not just little channelopathies)". Heart Rhythm. San Francisco, USA. May 2014.
49. "Investigations after Cardiac Arrest – a guide for clinicians". Presented to Cardiologists and Trainees, Middlemore Hospital. Auckland, New Zealand. April 2014.
50. "Workshop on Interpretation of ECGs". Asia Pacific Paediatric Cardiac Society. New Delhi, India. March 2014.
51. "Practical tips in drug management of tachyarrhythmia's". Asia Pacific Paediatric Cardiac Society. New Delhi, India. March 2014.
52. "Treatment of asymptomatic Children with cardiac Ion Channelopathies". Asia Pacific Paediatric Cardiac Society. New Delhi, India. March 2014.
53. "Heart Rhythm problems in children, from nuisance to fatality". Presented to @Heart, Parent Support Group. Christchurch, New Zealand. October 2013.

54. "Investigation of young sudden death, the New Zealand experience" Presented to Tahitian Cardiologists. Papeete, Tahiti. October 2013.
55. "SNPs and risk of sudden cardiac death". Heart Rhythm New Zealand National Meeting. Tauranga, New Zealand. September 2013.
56. "Early repolarisation syndromes". Heart Rhythm New Zealand National Meeting. Tauranga, New Zealand. September 2013.
57. "Long QT, Brugada syndrome and other inherited arrhythmia syndromes". In Session: Advice for the General Cardiologist. Cardiac society of Australia and New Zealand. Brisbane, Australia. August 2013.
58. "Challenges in patients with long QT syndrome". Cardiac Society of Australia and New Zealand. New Zealand Meeting. Wellington, New Zealand. June 2013.
59. Debate– (protagonist): "Long QT1 subjects adequately treated with beta blockers should not be allowed to do elite sports" v Sami Viskin. Heart Rhythm. Denver, USA. May 2013.
60. "Update on Long QT syndrome". Channelopathy Session at Heart Rhythm. Denver, USA. May 2013.
61. "Resuscitated Sudden Cardiac Death– what next?" World Congress of Paediatric Cardiology and Cardiac Surgery. Cape Town, South Africa. February 2013.
62. "Early repolarisation syndromes such as short QT syndrome". World Congress of Paediatric Cardiology and Cardiac Surgery. Cape Town, South Africa. February 2013.
63. "Functional echocardiography– what the neonatologist wants to know". World Congress of Paediatric Cardiology and Cardiac Surgery. Cape Town, South Africa, February 2013.
64. "Cardiac Genetic Investigation of young sudden death". Presented to the NZ National Coroner's Update meeting. Crowne Plaza Hotel. Auckland, New Zealand. November 2012.
65. "SUDEP: Sudden unexplained death in people misdiagnosed with epilepsy?" Greenlane Scientific Sessions. Auckland City Hospital. Auckland, New Zealand. October 2012.
66. "Assessment of long QTc". CSANZ: Paediatric Cardiology Session, Brisbane, Australia. August 2012.
67. "Paediatric Pacemaker Mischief". Heart Rhythm New Zealand Cardiac Physiologist Meeting. Auckland, New Zealand. August 2012.
68. "Clinical application of cardiac genetic testing". Co-hosted seminar ASEANZ Cardiovascular and Metabolic Forum. Melbourne, Australia. June 2012.
69. "Sudden Death in the young: the role of the Cardiac Ion Channelopathies". Auckland Cardiac Mechanics Workshop, International Society for Heart Research, Australasian Section. University of Auckland, Auckland, New Zealand. June 2012.
70. "Pacemakers and ICDs: running the interference". NZ Paediatric Society. Annual Meeting. Auckland, New Zealand. November 2011.
71. "ECG masterclass". Heart Rhythm NZ Meeting. Wellington, New Zealand. October 2011.
72. "Bicuspid Aortic Valve Disease". Study day in Adult Congenital Heart Disease. Taunton, UK. September 2011.
73. "Inherited Heart Diseases". Advanced Cardiac Nurse Training Course. University of Auckland, Auckland, New Zealand. July 2011.
74. "What to do after a near miss-sudden death in intensive care". Paediatric Cardiac Intensive Care Society. Cambridge, UK. September 2011.
75. "Childhood Brugada: Diagnosis and Therapy". Heart Rhythm Society Meeting. San Francisco, USA. May 2011.

76. "Management of infants having LQTS". PACES symposium on "Difficult management issues in the Young Patient Having a channelopathy". San Francisco, USA. May 2011.
77. "Echocardiography on the neonatal unit; Benefits Barriers and Pitfalls". Second Functional echocardiography course (Neonatology). Monash Medical Centre. Melbourne, Australia. October 2010.
78. "Pulmonary Hypertension in Children". Cardiology/Paediatrics CME Meeting. Mamao Hospital, Papeete, Tahiti. September 2010.
79. "Sudden Death in the Young". Cardiovascular Forum. Marion Davis Centre, Auckland, New Zealand. September 2010.
80. "Identifying heart disease in children". Taranaki (Care4kids) Nurse Study Day. Taranaki Base Hospital. Taranaki, New Zealand. August 2010.
81. "Epidemiology and genetics of sudden cardiac death". ASEANZ conference. Melbourne, Australia. June 2010.
82. "Investigation of Sudden Death in the Young- implications for PICU". Paediatric Intensive Care Unit Staff, Starship Hospital. Auckland, New Zealand. March 2010.
83. "Cardiac/Genetic Investigation of Sudden Death in the Young". Waikato Cardiology and Pathology Group Meeting. Waikato Public Hospital. Hamilton, New Zealand. February 2010.
84. "Sudden Death in the Young- from the lab to the community". Second New Zealand Round Table on Human Genomics. The Law Foundation. Wellington, New Zealand. November 2009. (Speaker and panel discussant).
85. "Implications of Genetic testing: managing and disclosing risk". Session on Sudden Cardiac Death. Cardiac Society of Australia and New Zealand. Sydney, Australia. August 2009.
86. Debate "Mass infant ECG screening is a waste of health service resources". World Congress of Paediatric Cardiology and Cardiac Surgery. Cairns, Australia. June 2009.
87. "Investigation of young sudden death- integration of family investigations" World Congress of Paediatric Cardiology and Cardiac Surgery. Cairns, Australia. June 2009.
88. "Cardiac Sodium channel disease" World Congress of Paediatric Cardiology and Cardiac Surgery. Cairns, Australia. June 2009.
89. "Cardiac Inherited Diseases: what the adult cardiologist needs to know." Cardiac Society of Australia and New Zealand. New Zealand Meeting. Wellington, June 2009.
90. "Sudden Death of Children In New Zealand" Paediatric Society. Paihia, Bay of Island, New Zealand. October 2008
91. "Cardiac Inherited Diseases- from the cradle to the grave". New Zealand Pacing and Electrophysiology Group. Christchurch, New Zealand. September 2008.
92. "Paediatric Sudden Death" in Congenital arrhythmias. Paediatric Cardiology Session. Cardiac Society of Australia and New Zealand. Adelaide, Australia. May 2008.
93. "Sodium Channel Mutations and Arrhythmias". Monogenetically Mediated Heart Disease Session in Clinical (adult) Cardiology. Cardiac Society of Australia and New Zealand. Adelaide, Australia. May 2008.
94. "Mechanisms of arrhythmia". Post Graduate Nursing Course (730) in Paediatric Cardiology and Cardiac Surgery. Auckland School of Nursing. Auckland, New Zealand. August 2008.
95. "Inherited Heart Diseases" Post Graduate Nursing Course in Cardiology. Auckland school of nursing. Auckland, New Zealand. July 2008.

96. "Modern techniques in catheter ablation of cardiac arrhythmias". New Zealand National Paediatric Update Weekly Meeting (teleconference). Fisher and Paykel Education Centre, Auckland City Hospital. Auckland, New Zealand. April 2008.
97. "Blackouts, funny turns and indifferent ECGs – Picking out the bad ones". Emergency Paediatricians. Emergency Department, Starship Children's Hospital. Auckland, New Zealand. April 2008.
98. "Post-operative Echocardiography". Advanced Paediatric Echocardiography Course, Fisher and Paykel Education Centre. Auckland City Hospital. Auckland, New Zealand. March 2007.
99. "Arterial Abnormalities". Advanced Paediatric Echocardiography Course. Fisher and Paykel Education Centre. Auckland City Hospital. Auckland, New Zealand. March. 2007.
100. "Blackouts, funny turns and indifferent ECGs – Picking out the bad ones". Paediatric Update for Senior Paediatricians. Fisher and Paykel Education Centre. Auckland City Hospital. Auckland, New Zealand. March 2008.
101. "Investigation of young sudden death". Royal College of Pathologists of Australasia. Napier, New Zealand. September 2007.
102. "Genetics of long QT syndrome". Murdoch Genetics Institute. Melbourne, Australia. September 2007.
103. "Burning and freezing in four dimensions". Green Lane Scientific Meeting (GLOBS). Auckland City Hospital, Auckland, New Zealand. September 2007.
104. "Long QT and Ion Channelopathies". Cardiac Genetics Symposium, Cardiac Society of Australia and New Zealand. New Zealand Meeting. Christchurch, New Zealand. August 2007.
105. "Problems with the investigation of young sudden death". New Zealand Pacing and Electrophysiology Group. Rotorua, New Zealand. August – September 2007.
106. "How can genetic diagnoses reduce sudden death in the young?" Human Genetic Society of Australasia. Sky City Auckland Convention Centre. Auckland, New Zealand. July 2007.
107. "Genetic diagnosis in sudden death". Boston Scientific Sudden Cardiac Death Symposium - Sponsored Meeting Sanctuary Cove. Queensland, Australia. June 2007.
108. "Long QT and sudden death in New Zealand". Green Lane Scientific Meeting (GLOBS). Auckland City Hospital, Auckland, New Zealand. November 2006.
109. "Molecular genetics of Long QT syndrome in sudden adult death syndromes". LabPlus User Forum. Fisher and Paykel Education Centre. Auckland City Hospital. Auckland, New Zealand. November 2006.
110. "Arrhythmic cause of syncope in children". Paediatricians and Cardiologists. Auckland City Hospital, Auckland, New Zealand. October 2006.
111. "Arrhythmic cause of syncope in children". Papeete, Tahiti. October 2006.
112. "Inheriting and living with inherited sudden death syndromes". Paediatric Grand Round. Auckland City Hospital, Auckland, New Zealand. September 2006.
113. "Long QT syndrome and sudden unexpected death". NZPEG meeting. Mt Ruapehu, Ohakune, New Zealand. September 2006.
114. "Genetic investigation of cardiac channelopathies: Service or research?" Cardiostim. Nice, France. June 2006.
115. "Syncope in children". CIR Nurses. Auckland City Hospital. Auckland, New Zealand. April 2006.

116. "Syncope in children". The Goodfellow Symposium" (GP CME Program). School of Population Health, Medical and Health Sciences, University of Auckland. Auckland, New Zealand. March 2006.
117. Expert Panel Member - "Young sudden death" and "What is the cost of young sudden death" and "How much does it cost to prevent it?" Symposium on Genetics and Sudden Death. National Heart Meeting. Sydney Convention Centre. Sydney, Australia. March 2006.
118. "Cardiac channelopathies". Royal College of Pathology of Australasia Meeting. Sydney, Australia. March 2006.
119. "Sudden death in children". New Zealand Resuscitation Council (NZRC) Meeting. Wellington, New Zealand. December 2005.
120. "Cardiac channelopathies and sudden death". Genetics symposium. Asia Pacific Interventional Advances (APIA). Newcastle, NSW, Australia. November 2005.
121. "Inherited arrhythmias". Heart Children Annual Conference. Wellington, New Zealand. October 2005.
122. "Cardiac arrhythmias and sudden death". Genetics symposium. Cardiac Society of Australia and New Zealand. Perth, Australia. August 2005.
123. "Cardiac Inherited Diseases". Post Graduate Cardiac Nursing Course. University of Auckland. Auckland, New Zealand. June 2005.
124. "Long QT syndrome and sudden death". Report to The Genetics Working Group for the Cardiac Society of Australia and New Zealand. Sydney Convention Centre. Sydney, Australia. November 2004.
125. "Sudden death in the young". Cure Kids "Return of The Riders" Corporate Sponsorship. Ellerslie Novotel. Auckland, New Zealand. October 2004. (Raising $253,000).
126. "Role of genetic diagnosis in syncope". Cardiac Society of Australia and New Zealand. Brisbane, Australia. August 2004.
127. "Work of the Cardiac Inherited Disease Group". New Zealand Pacing and Electrophysiology Group. Cardiac Society of New Zealand. Taupo, New Zealand. June 2004.
128. "Long QT syndrome in New Zealand". Human Genetics Society of Australasia (NZ Branch). Wellington, New Zealand. November 2003.
129. "Diagnosing the cause of sudden unexpected death when the post mortem is negative". Royal College of Pathologists of Australasia (NZ) Annual Scientific Meeting. Heritage Hotel. Auckland, New Zealand. October 2003.
130. "Syncope" and "SIDs". Meeting of Paediatricians and Cardiologists. Papeete, Tahiti. September 2003.
131. "Recognition of congenital heart disease in the newborn". New Zealand National Neonatal Nurses Conference. Auckland, New Zealand. February 2003.
132. "Recognition of congenital heart disease in the newborn". Taranaki Day. Symposium on The Newborn. New Plymouth, New Zealand. August 2003.
133. "Recognition of congenital heart disease in the newborn". Update in Neonatal Diagnosis for Midwives (National Meeting). Auckland, New Zealand. October 2003.
134. "Sudden Infant Death Syndrome". 12th World Congress on Cardiac Pacing and Electrophysiology. Hong Kong, Hong Kong. February 2003.
135. "Keyhole cardiac surgery in children". Rotary International. North Shore Branch. Takapuna, Auckland, New Zealand. November 2002.

136. "Indications for and types of intervention in ASDs and VSDs and "neonatal PDA". Meeting of Paediatricians and Cardiologists. Papeete, Tahiti. November 2002.
137. "Debate: There is no place for neonatologists doing echocardiography on the neonatal unit (Antagonist)". World Cardiology Congress. Sydney, Australia. May 2002.
138. "Children syncope and long QT syndrome: The genetic clinical interface" - World Cardiology Congress. Sydney, Australia. May 2002.
139. "Echocardiography of adult congenital heart disease". Queensland University of Technology/Agilent Echocardiography Course. Sydney, Australia. June 2001.
140. "Arrhythmias in congenital heart disease". Cardiac Society of Australia and New Zealand. New Zealand Meeting. Auckland, New Zealand. August 2001.
141. "Current status of adults with congenital heart disease". Anaesthetic Department at Auckland City Hospital. Auckland, New Zealand. February 2000.
142. "Clinical aspects of long QT syndrome". Cardiology Registrar Seminar at Heritage Hotel. Auckland, New Zealand. May 2000.
143. "Congenital heart disease as a model for cardiopulmonary interactions". Cardiopulmonary Workshop. Glaxo Wellcome. Wellington, New Zealand. 1999.
144. "Arrhythmias post-surgery for congenital heart disease" - Cardiac Society of Australia and New Zealand. New Zealand Meeting. Wellington, New Zealand. August 1999.

Convenor/Chairmanship/Faculty at Meetings

1. International Cardiovascular Genetics Conference (ICCG2018.com). Convenor of Educational Symposium. Brisbane, Australia. May 2018.
2. Heart Rhythm Society (Faculty, invited speaker). Boston, USA. May 2018
3. National CIDG meeting (with HRNZ), convenor and lecturer, chairman, Auckland, September 2017
4. National CIDG meeting (with HRNZ), convenor and lecturer, chairman, Auckland, September 2016
5. Heart Rhythm Society (Faculty, Chair, invited speaker). Denver, USA. May 2016
6. International Cardiovascular Genetics Conference (ICCG2016.com). Convenor of Educational Symposium. Brisbane, Australia. May 2016.
7. National CIDG meeting (with HRNZ), convenor and lecturer, chairman, Auckland, September 18 2015
8. Public lecture and convenor of public forum, Auckland 2015, The history and development of services for investigation of and prevention of young sudden death in New Zealand. Auckland, September 18 2015
9. International Cardiovascular Genetics Conference (ICCG2014.com). Convenor of Educational Symposium. Brisbane, Australia. August 2014.
10. Heart Rhythm Society (Faculty, invited speaker). San Francisco, USA. May 2014.
11. Heart Rhythm Society (Faculty, invited speaker x 2). Denver, USA. May 2013.
12. World Congress of Paediatric Cardiology and Cardiac Surgery. Organising faculty. Chair of Session: "What's New in electrophysiology and catheter ablation?" Invited Speaker (see Above). Cape Town, South Africa 2013.
13. TRAGADY meeting (Chair). Melbourne, Australia. November 2012.
14. Cardiac Society of Australia and New Zealand. Chair of Session: "Role of Cardiac Genetic Clinics". Brisbane, Australia. August 2012.

15. PACES (Pediatric and Congenital Electrophysiology Society) (Invited Speaker). Heart Rhythm Society Satellite. San Francisco, USA. May 2011.
16. Heart Rhythm Society (Invited Speaker). San Francisco, USA. May 2011.
17. TRAGADY meeting (Chair). Sydney, Australia. April 2010.
18. Heart Rhythm Society (Invited Expert). Panel discussion and presentation. "Genetic of sudden cardiac death. Interpretation of probable and possible and type 2 mutations". Denver, USA. May 2010.
19. "What's Hot in EP?" World Congress of Paediatric Cardiology. Cairns, Australia. June 2009.
20. New Zealand Pacing and Electrophysiology Group. Session: Cardiac Inherited Diseases. Christchurch, New Zealand. September 2008.
21. "Channelopathies in arrhythmogenesis". Electrophysiology Session. Cardiac Society of Australia and New Zealand. Adelaide, Australia. May 2008.
22. TRAGADY Meeting. Research Sub Group. Adelaide, Australia. August 2008.
23. TRAGADY Meeting. Brisbane, Australia. September 2007.
24. TRAGADY Meeting. Melbourne, Australia. April 2007.
25. "Case studies in hereditary arrhythmias". Cardiac Society of Australia and New Zealand. Canberra, Australia. August 2006.
26. TRAGADY Meeting. Sydney, Australia. August 2006.
27. Pathology sub-group of TRAGADY. Sydney, Australia. March 2006.
28. TRAGADY Meeting. Sydney, Australia. November 2005.
29. Inaugural Meeting of The Australasian Task Force on The Prevention of Sudden Death In The Young (TRAGADY). Sydney, Australia. June 2005.
30. Monthly National Meetings of The Cardiac Inherited Disease Group (www.cidg.org). April 2004- Present.
31. "Implantable and combined monitoring devices". World Congress on Cardiac Pacing and Electrophysiology. Hong Kong. February 2003.
32. "New technical advances in Paediatrics". Joint Meeting of The Paediatric Society and Physicians Societies, RCPCH. Auckland, New Zealand. October 2001.
33. "Syncope and Tachyarrhythmias in children". Cardiac Society of Australia and New Zealand. New Zealand Meeting. Auckland, New Zealand. August 2001.

Other Presentations to Learned Societies: (those without a published abstract)

1. Groves AM, Kuschel CA, Knight DB, **Skinner JR**. Relationship between blood pressure and blood flow in the first postnatal day in preterm infants. The Neonatal Society, London. 18 November 2004.
2. **Skinner JR**, Hood M, Smith WM. Catheter ablation of tachyarrhythmia's in children: Recent results from Auckland. Cardiac Society of Australia and New Zealand. New Zealand Meeting. Auckland, New Zealand. 5-8 August 2001.
3. **Skinner JR**, Hood M, Smith WM. Radiofrequency ablation of accessory pathways after the Fontan operation. Cardiac Society of Australia and New Zealand. New Zealand Meeting. Auckland, New Zealand. 5-8 August 2001.
4. **Skinner JR**. Radiofrequency ablation of tachyarrhythmia's in children: Recent results from Green Lane. New Zealand Paediatric Society. Napier, New Zealand. October 2000.

5. **Skinner JR**, Hey EN, Hunter S. Assessment of ductal shunting: The significance of the murmur. British Paediatric Cardiac Association. Bristol, UK. 24 November 1995 and also to South West Paediatric Club 17 November 1995 (Prize for best presentation).
6. **Skinner JR**, Martin RP, Hayes AM, Joffe H, Jordan SC. Early mortality in infants with hypertrophic cardiomyopathy 1969-93. British Paediatric Association. York, UK. March 1995; and Association of European Paediatric Cardiologists, Bologna, Italy. 17-20 May 1995.
7. **Skinner JR**, Hunter S. Echocardiography in the neonatal unit: A job for the cardiologist or the neonatologist? British Paediatric Association, York, UK. March 1995.
8. Skinner J, Hunter S, Hey EN. Haemodynamic effects of spontaneous and indomethacin induced ductal closure. British Paediatric Association. Warwick University, UK. 16-19 April 1991.
9. Morris K, Skinner J, Wren C, Hunter S, Coulthard M. Abnormal cardiac function in children with end stage renal failure. British Paediatric Association. Warwick University, UK. 16-19 April 1991.
10. **Skinner JR**, Hunter S, Hey EN. Tricuspid regurgitation and pulmonary artery pressure in healthy neonates. Linden Hall Paediatric Research School. UK. September 1989 (Convenors A Aynsley-Green, D Hull, R Moxon)
11. Richardson P, Skinner J, Selby C. Cortisol excretion rate in childhood. An evaluation of normal urinary cortisol excretion rate in children. Read at the British Endocrine Society Meeting 1986.

GRANT REVIEWS AND PUBLICATION REVIEWS

I have not kept an exhaustive record of reviewed papers and grants. In the last 12 months I have reviewed 32 papers submitted for publication and three international grant applications (Denmark, UK, Netherlands). Journals reviewed for include *New England journal of Medicine, Circulation, Human Genetics, Heart Rhythm, Circulation arrhythmia and electrophysiology, American Heart Journal, Archives of Disease in Childhood, American Journal of Human Genetics, Stem cells* and *development.* As editorial board member for *Heart Rhythm* I review about one new paper per 2-4 weeks.

Referees

Inquiry into the convictions of Kathleen Megan Folbigg

24 January 2019

Professor Jon Skinner
Paediatric Cardiologist
Starship Child Health
Park Road
Grafton
Auckland 1023
NEW ZEALAND

By email: jskinner@adhb.govt.nz

Dear Professor Skinner

Letter of Engagement

Summary

On 22 August 2018 the Governor of New South Wales directed that an inquiry be held into the convictions of Kathleen Megan Folbigg for three counts of murder, one count of manslaughter and one count of maliciously inflicting grievous bodily harm in respect of her four children on 21 May 2003 ("the Inquiry"). The Crown Solicitor is the Solicitor Assisting the Honourable Reginald Oliver Blanch AM QC ("the Judicial Officer") with the Inquiry and Amber Richards has carriage of the matter on the Crown Solicitor's behalf.

The scope of the Inquiry includes consideration of expert medical evidence, including:

- any new research or advances in medical science relevant to the causes of death of each child and the cause of the apparent or acute life threatening event in respect of one child, Patrick.
- expert medical opinion as to the causes of death of each child and the cause of the apparent or acute life threatening event in respect of Patrick in light of any relevant new research or advances in medical science.
- any new research or literature concerning the incidence of reported deaths of three or more infants in the same family attributed to unidentified natural causes.
- any other related expert medical evidence.

Scope of engagement

As discussed you are engaged, as one of a team of experts, to interpret raw data produced as a result of genetic sequencing and testing arranged by the Inquiry in respect of the Folbigg family, and provide an expert report to the Inquiry regarding the presence of any likely pathogenic genes, or pathogenic genetic variants in genes, that are known to be associated with sudden unexpected death in infancy.

As per our previous correspondence we anticipate the data will be available by early February 2019 and we are looking to convene a meeting of the geneticists who will be involved in the interpretation of the data on Monday 4 February 2019 to determine how the interpretation phase will proceed.

We confirm the team of experts the Inquiry is engaging to assist in the interpretation of the raw data comprises:

- Dr Michael Buckley MBChB PhD FRCPA FHGSA FRCPath: senior staff specialist genetic pathologist employed by NSW Health Pathology and currently the President of the Human Genetics Society of Australasia

- Professor Edwin Kirk MBBS PhD FRACP FRCPA: senior staff clinical geneticist and genetic pathologist employed by NSW Health Pathology and Chief Examiner in Genetics for the Royal College of Pathologists of Australia.

- Dr Alison Colley MBBS FRACP FRCPA: senior staff specialist clinical geneticist in specialist practice at Liverpool Hospital and Director of the Liverpool Genetics Service

- Professor Matthew Cook MBBS PhD FRACP FRCPA: clinical immunologist in specialist practice employed by ACT Health at Canberra Hospital and the ACT pathologist service.

It is possible that you will be required to give oral evidence at the public hearings of the Inquiry. The hearing of evidence relevant to genetics will be held in Sydney in the week starting Monday, 18 March 2019. If feasible, it would be preferable for you to give evidence in person and we can make arrangements for you to travel to Sydney at our cost. If this is not practical, arrangements can be made for you to appear by audio-visual link. Please advise us of your availability in this regard as soon as possible.

Preparation of your report

The Inquiry would be assisted if you could interpret the results of the genetic testing and prepare a report which identifies and explains whether any of the results of that testing are relevant to the causes of death of each of the children.

Your report should only offer opinions to the extent those opinions are based upon your knowledge, training and fields of specialist expertise.

In preparing your report, please:

i. identify (and reference as appropriate) any facts and assumptions from materials upon which you rely;

ii. show how those facts and assumptions relate to your opinions;

iii. provide an explanation of your reasons for each of your opinions;

iv. define and explain any technical terms; and

v. if necessary, set out any qualification or reservations you have about the opinions expressed in your report (for instance, because of reservations you hold about a fact, or if further information is required, or for any other reason).

Documents with which you are briefed

We have already provided you with a set of briefing documents. For your reference, the index to this set is set out below in **Annexure A**. We will provide you with any other necessary documents as they come to hand.

Expert code of conduct and curriculum vitae

At **Annexure B** to this letter I set out the Expert Witness Code of Conduct and ask that you read it carefully. In your report you should acknowledge that you have read the Code and agree to be bound by it. I suggest the following form of words be included in the body of the report:

> "I, Professor Jon Skinner, acknowledge that for the purpose of Rule 31.24 of the Uniform Civil Procedure Rules 2005 that I have read the Expert Witness Code of Conduct in Schedule 7 to the said rules and agree to be bound by it."

I also request that you please attach a copy of your curriculum vitae to your report.

Fees

I note that this engagement is subject to approval of your fees. As soon as possible, please advise us of your fees (including the relevant currency and whether or not they include GST) and an estimate of how many hours you anticipate spending on the preparation of your report. If you later consider further time is necessary for the completion of your report, please advise us in advance. In the event you are required to give evidence, we will agree the time to be spent separately.

If an amount for GST is to be included in your fee, you will need to cite your ABN and either indicate that the fee is inclusive of GST or separately indicate the amount of GST charged. If your fee is more than $50.00 and no ABN or compliant invoice is supplied I am required to withhold 48.5% of the fee on account of GST.

It is essential to comply with Australian Taxation Office requirements that any invoice be addressed to the business name: NSW Crown Solicitor's Office.

Confidentiality

Please ensure you keep your engagement, the documents with which you are briefed, and your report **confidential**.

Conclusion

Please do not hesitate to contact Amber Richards, Senior Solicitor, on (02) 9258 0832 or amber.richards@cso.nsw.gov.au if you have any queries or require anything further to assist in the preparation of your report.

Kind regards

Amber Richards
Senior Solicitor
for Crown Solicitor

ANNEXURE A

Index to briefing material

Tab	Document	Date	Source
1.	Medical testing of Patrick Folbigg	13 February 1991	ODPP Volume 1 of 7, Tab 29
2.	Letter to Doctor Wilcken regarding newborn blood sample	11 October 1999	ODPP SYD02582197, Tab 168
3.	Genetics report regarding death of Caleb Folbigg	13 January 2000	SC054200, Tab 211
4.	Genetics report regarding death of Patrick Folbigg	13 January 2000	SC054200, Tab 212
5.	Genetics report regarding death of Sarah Folbigg	13 January 2000	SC054200, Tab 213
6.	Genetics report re death of Laura Folbigg	13 January 2000	SC054200, Tab 214
7.	Expert Certificate / Statement of Doctor Bridget Wilcken and exhibits 7a – 7c	14 January 2000	ODPP SYD02583893, Tab 98
7a.	Letter from Doctor Alison Colley to Doctor Bridget Wilcken regarding Caleb and Patrick Folbigg	4 December 1991	ODPP SYD02583893, Tab 100
7b.	Letter from Doctor Bridget Wilcken to Doctor Alison Colley regarding Caleb and Patrick Folbigg	10 December 1991	ODPP SYD02583893, Tab 101
7c.	NSW Newborn Screening Programme Report regarding Caleb, Patrick, Sarah and Laura Folbigg	13 January 2000	ODPP SYD02583893, Tabs 103-106
8.	Report of Professor Peter Berry	November 2000	ODPP SYD2575735, Tab 25
9.	Letter from Doctor Alison Colley to Doctor Bridget Wilcken	27 February 1992	ODPP SYD02583893, Tab 102
10.	Letter from Professor David Isaacs re testing for serum levels of IgG of Folbigg children	3 March 2003	ODPP SYD02584119, Tab 109
11.	Facsimile from Dr J Vivian Wells re IgG levels	5 March 2003	ODPP SYD02584119, Tab 120
12.	Medical Testing for IgG deficiency, prolonged QT and "Druckers" Gene	7 March 2003	SC54199, Tab 27
13.	Letter from Doctor Allan Cala re IL-10 Gene theory	19 March 2003	ODPP SYD02582197, Tab 13
14.	Letter from Doctor John Christodoulou to J Culver regarding genetic causes of some cases of SIDS	18 February 2003	ODPP SYD02584119, Tab 89
15.	Summary of present situation re medical investigations prepared by Peter Krisenthal, Legal Aid	27 February 2003	ODPP SYD02584119, Tab 101

Tab	Document	Date	Source
16.	Letter from Professor John Hilton to ODPP regarding genetic testing of Folbigg Children	27 February 2003	DPP SYD02584119, Tab 100
17.	Transcript of evidence of Doctor Bridget Wilcken at trial (pages 817-823)	16 April 2003	Initial Transcripts bundle Volume 3, Tab 11 (provided with Preliminary Bundle)
18.	Supplementary Report of Professor Peter Berry regarding Doctor Drucker's work on IL-10 gene polymorphism theory	29 April 2003	ODPP SYD02584119, Tab 188
19.	Final Report of Professor Cecelia Blackwell	8 May 2014	Professor Cecelia Blackwell
20.	Article, "Exploring the risk factors for sudden infant deaths and their role in inflammatory responses to infection" (Frontiers in Immunology, Volume 6, Article 44) by Caroline Blackwell, Sophia Moscovis, Sharron Hall, Christine Burns and Rodney J Scott	March 2015	N/A
21.	Chapters from Book, *SIDS - Sudden infant and early childhood death: The past, the present and the future* edited by Jhodie R Duncan and Roger W Byard (University Adelaide Press 2018):	N/A	N/A
21a.	Chapter 2 – Sudden Infant Death Syndrome: An Overview by Jhodie R Duncan and Roger W Byard	N/A	N/A
21b.	Chapter 14 – Future Directions in Sudden Unexpected Death in Infancy Research by Heather E Jeffery	N/A	N/A
21c.	Chapter 30 – Cytokines, Infection, and Immunity by Siri Hauge Opdal	N/A	N/A
21d.	Chapter 31 – The Genetics of Sudden Infant Death Syndrome by Catherine A Brownstein, Annapurna Poduri, Richard D Goldstein and Ingrid A Holm	N/A	N/A
22	Specimen slides relating to Caleb, Sarah, Laura and Patrick Folbigg – Working Document	15 November 2018	N/A
23	NSW Forensic & Analytical Science Service - Histology Request Forms	N/A	N/A

ANNEXURE B

Uniform Civil Procedure Rules 2005, Sch 7: Expert Witness Code of Conduct

1 Application of code

This code of conduct applies to any expert witness engaged or appointed:

(a) to provide an expert's report for use as evidence in proceedings or proposed proceedings, or

(b) to give opinion evidence in proceedings or proposed proceedings.

2 General duties to the Court

An expert witness is not an advocate for a party and has a paramount duty, overriding any duty to the party to the proceedings or other person retaining the expert witness, to assist the court impartially on matters relevant to the area of expertise of the witness.

3 Content of report

Every report prepared by an expert witness for use in court must clearly state the opinion or opinions of the expert and must state, specify or provide:

(a) the name and address of the expert, and

(b) an acknowledgement that the expert has read this code and agrees to be bound by it, and

(c) the qualifications of the expert to prepare the report, and

(d) the assumptions and material facts on which each opinion expressed in the report is based (a letter of instructions may be annexed), and

(e) the reasons for and any literature or other materials utilised in support of each such opinion, and

(f) (if applicable) that a particular question, issue or matter falls outside the expert's field of expertise, and

(g) any examinations, tests or other investigations on which the expert has relied, identifying the person who carried them out and that person's qualifications, and

(h) the extent to which any opinion which the expert has expressed involves the acceptance of another person's opinion, the identification of that other person and the opinion expressed by that other person, and

(i) a declaration that the expert has made all the inquiries which the expert believes are desirable and appropriate (save for any matters identified explicitly in the report), and that no matters of significance which the expert regards as relevant have, to the knowledge of the expert, been withheld from the court, and

(j) any qualification of an opinion expressed in the report without which the report is or may be incomplete or inaccurate, and

(k) whether any opinion expressed in the report is not a concluded opinion because of insufficient research or insufficient data or for any other reason, and

(l) where the report is lengthy or complex, a brief summary of the report at the beginning of the report.

4 Supplementary report following change of opinion

(1) Where an expert witness has provided to a party (or that party's legal representative) a report for use in court, and the expert thereafter changes his or her opinion on a material matter, the expert must forthwith provide to the party (or that party's legal representative) a supplementary report which must state, specify or provide the information referred to in clause 3 (a), (d), (e), (g), (h), (i), (j), (k) and (l), and if applicable, clause 3 (f).

(2) In any subsequent report (whether prepared in accordance with subclause (1) or not), the expert may refer to material contained in the earlier report without repeating it.

5 Duty to comply with the court's directions

If directed to do so by the court, an expert witness must:

(a) confer with any other expert witness, and

(b) provide the court with a joint report specifying (as the case requires) matters agreed and matters not agreed and the reasons for the experts not agreeing, and

(c) abide in a timely way by any direction of the court.

6 Conferences of experts

Each expert witness must:

(a) exercise his or her independent judgment in relation to every conference in which the expert participates pursuant to a direction of the court and in relation to each report thereafter provided, and must not act on any instruction or request to withhold or avoid agreement, and

(b) endeavour to reach agreement with the other expert witness (or witnesses) on any issue in dispute between them, or failing agreement, endeavour to identify and clarify the basis of disagreement on the issues which are in dispute.