EXHIBIT AJ

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15 March 2019

Mr. Stuart Gray
Partner, Cardillo Gray Partners
P.O. Box 409
Newcastle NSW 2300

Dear Mr. Gray,

Re**: Patrick FOLBIGG date of birth 3.6.1990.**

I have reviewed the documents provided by your office, and at your request provide an assessment of Patrick Folbigg's neurological condition and its likely aetiology/aetiologies. In preparing this report I have reviewed the following documents:

1. Autopsy report of Patrick Folbigg (part 1) - 4 pages, dated 14.2.1991
2. Mater Hospital autopsy report part 2.
3. Records of Dr. Colley
4. Report of Expert Certificate provided by Dr. Joseph Dezordi (P74-p79).
5. Three electronic (.pdf) files forwarded by email from your office on 7.3.19. These .pdfs contain sections of the medical records of Patrick Folbigg. These records are incomplete. Some pages have been copied more than once.
6. Two electronic (.pdf) files forwarded by your office on 12-13.3.19.

Medical history
I have been provided with photocopies of some pages from Patrick's infant health book. Some of these are indistinct.

Patrick was born by a normal delivery on 3.6.1990 with a birthweight of 3410g, and a head circumference at birth of 33.5cm. His Apgar scores were 7 and 8 at 1 and 5 minutes, respectively. There were notes in the medical record that a prior sibling had died of SIDS at age 20 days.

Patrick was reviewed in hospital by a paediatrician, Dr. Morris, on 4.6.1990. He was felt to be a vigorous, normal baby (P427). He had no issues swallowing and no stridor (P854). There was no neonatal jaundice. A baseline ECG and electrolytes were normal (P476).

Patrick was admitted to hospital electively when he was 12 days old for further investigations. The barium swallow from 15.6.90 was reported as showing some reflux of contrast into the nasal cavity- suggesting some incoordination of swallowing- but no significant pathology. A full blood count and other blood tests (CK, electrolytes and LFTS) taken during that admission were essentially normal.

Dr. Morris recorded in his infant health book at three weeks of age (27.6.1990) that Patrick's sleep study, karyotype and barium swallow were normal.

On 6.8.1990 Patrick was described as having torticollis to the left with no tumour in the sternocleidomastoid muscle. This was treated expectantly.

At 10 weeks, Patrick was said to be making sounds, smiling and attempting to laugh.

On 2.9.1990 Patrick was described in his infant health book as being 'strong in the legs'. He was trying to roll over. He noticed sounds and followed whatever was moving - in his visual fields, presumably.

On 3.9.1990 it was reported that the torticollis persisted but that his general progress was good.

On 20.9.1990, at age 15 weeks, Patrick was described in his infant health book as blowing raspberries and laughing. He could grab and play with things in his reach and sight. He was doing well with early solids (P435).

Patrick was admitted to the Mater Hospital from 18th to 29th October 1990.

Dr. Dezordi's admission note (06:00 am 18.10.1990, P81) noted that Patrick presented as a five-month-old baby after an apnoeic episode. He had been snuffly for three days, with a dry cough and some vomiting, but was otherwise well and behaving normally. He had not received any medications. He had been in contact with other children with intercurrent illnesses. Patrick had been seen by his mother at 3 a.m. because he was coughing. At 04.30 am he was heard to be gasping. He was blue around the lips, lifeless and floppy. He had minimal respiratory effort. He later made a high-pitched cry. He revived somewhat when the paramedics arrived and administered oxygen.

The contemporaneous nursing triage note (P505) made when he arrived at the Mater Hospital emergency department described him as very pale, lethargic with arching of his back when disturbed.

Dr. Dezordi noted that there was no history of epilepsy or other neurological conditions in the family. At age 5m, Patrick had received two sets of immunisations. He was felt to be developmentally normal; he had smiled before six weeks of age, laughed and vocalised and could nearly roll over. He was felt to play with fingers and to bear weight on his legs.

On examination Patrick's initial temperature was 35°. He was tachycardic with a heart rate of 160 bpm, and tachypnoeic with a respiratory rate of 60. His blood pressure of 90/50 was normal. He was pale and lethargic, responding only to painful stimuli. His pupils were felt to be dilated but reactive. He was initially saturating at 88% in room air; this improved when he was given oxygen.

After 15 minutes, he became more alert. He was described as moving his head and arching his back (it was recorded 'always does this', according to the contemporaneous note). He had a large anterior fontanelle which was neither tense nor bulging. He had a snuffly nose. He had widespread wheeze on auscultation of the chest. His liver was slightly ptosed, at 2 cm below the costal margin, but not enlarged- its span was 7 cm. His cardiovascular examination was felt to be essentially normal. His fundi were felt to be normal and he had no strabismus. His tone was described as normal other than the arching of his neck. He was described as supporting weight when on his legs and moving his limbs against gravity and resistance. His reflexes were not elicited. His plantar responses were extensor.

Patrick's chest x-ray was felt to be normal. A fingerprick glucose level was 7 mmol/L. The baseline urea, creatinine and electrolytes were normal. A full blood count showed a white cell count of 11.6 with 1% bands, 2% monocytes, 86% lymphocytes and 11% neutrophils. A blood culture was sent. A urine bag showed 4+ glucose with a small amount of blood and 2+ protein.

Dr. Dezordi also noted that Patrick had undergone an elective admission to hospital at 12 days of age for a sleep study, which was normal, and a barium swallow, which he recorded was felt to show no reflux**.**

A further note by **Dr. Ian Wilkinson,** consultant paediatric neurologist, on 18.10.90 (P84) recorded that Patrick had been coughing, leading to gasping and cyanosis, and had then become apnoeic. Dr. Wilkinson described Patrick as being subdued with little spontaneous movement. He felt that the deep tendon reflexes in the lower limbs were '*somewhat brisk'*. He felt that the disc margin of the left (optic) fundus was indistinct. He recorded a head circumference of 43.4 cm. Dr. Wilkinson's impression was that the abnormalities on the physical examination were '*probably related to the recent events in a transient fashion*'. He recommended that Patrick undergo an EEG, a head ultrasound, and further blood tests if there was another event.

A report from 18.10.1990 by Dr. J.T. Holland described the findings of an EEG, most of which was obtained in stage 2 sleep. Sleep spindles were present and well formed. These were somewhat asynchronous but not asymmetrical. The background activities were otherwise normal, and arousal at the end of recording was felt to be normal and symmetrical. This was felt to be a normal EEG for the age and state of sleep.

A blood culture from 18 October 1990 grew coagulase-negative staphylococcus from one of two bottles after one day. The significance of this was felt to be unclear.

A urine culture showed no white cells, 300 red cells, and no epithelial cells. There was no growth on the urine culture.

Blood tests obtained in the emergency department (P539) showed normal urea and creatinine, sodium and potassium, calcium and magnesium.

A nasopharyngeal aspiration showed no evidence of respiratory viruses on antigen testing or viral culture.

A chest x-ray taken at 6:20 AM on 18 October 1990 was felt to show increased lung markings compatible with bronchiolitis.

A head ultrasound obtained at 09:44 on 18 October 1990 was normal. (P544).

The nursing note from 3:15 PM on 18 October 1990 described a quiet baby whose observations were satisfactory. The baby was felt to be feeding well. His urinalysis, on admission to the ward, showed a specific gravity of 1025 with a pH of 5 and no other abnormalities on the urine dipstick (i.e. there was no proteinuria or haematuria at that point).

A medical note (P532, continued on P552, also presented as P579) from Dr. Pallas later on 18 October 1990 described persistent crying and irritability. He was described as 'pale and crying non-stop for one minute then stopping for another minute'. He was pale and sweaty.

Dr. Pallas raised the possibilities of Sandifer syndrome and the anomalous coronary artery syndrome.

Patrick's nursing observations over the course of 18 October 1990 showed that his temperature normalised, whilst he remained intermittently tachypnoeic and tachycardic.

Overnight on 18-19.10.1990 the nursing and medical notes describe Patrick as very unsettled and irritable until he was given a sedative, Panquill. He was described as arching his back when fed (P580).

A renal ultrasound obtained on 19.10.1990- because of the red cells seen on the initial urine microscopy - was normal (P602).

On 19 October 1990 Patrick's general observation sheet (P549) and nursing/medical notes (P581-582, P609-610) recorded multiple (at least 10) seizures with jerking of the legs, back arching and staring upwards for between 40 seconds and 3-4 minutes at a time. The medical notes described multifocal seizures variously involving the right leg, left arm, then left leg and right arm, with or without eye deviation to the left (P581).

Patrick received intravenous diazepam (Valium) overnight on 19 October 1990. He then received intravenous phenobarbitone.

He continued to have seizures on 20.10.1990, again with variable semiology, with eye deviation to the right and twitching of the right or both legs. Because of ongoing seizures, he was also treated with intravenous phenytoin.

A lumbar puncture on 20.10.1990 was normal, showing zero white cells, three red cells, protein of 0.22 g/L and glucose of 4.2 mmol/L. The CSF viral culture was negative (P604).

Patrick had ongoing seizures on the morning of 21 October, at which time treatment with acyclovir was initiated.

A repeat EEG was requested. The EEG was obtained on 22 October, with a recorded indication of focal fitting on the right side of the body. The EEG was abnormal for asymmetry of the background rhythms, with sharply contoured activity over the left frontoparietal region which represented '*a potential epileptogenic focus*'.

The medical notes from 24.10 (P562) noted that Patrick had undergone a CT scan on 23.10.1990 that showed hypodense areas in both temporal and occipital lobes consistent with but not altogether typical of herpesvirus infection.

The formal report from the CT (P614) read:

> *"In the pre-contrast scan there is a decrease in attenuation seen in both occipital lobes, temporal lobe and left frontal lobe. The grey/white matter differentiation is lost. Ventricular system not dilated. No haemorrhage seen. Minimal widening of the peripheral cerebral sulci is seen in the frontal and the parietal lobes.*

Post contrast scan with thin cuts over the posterior cranial fossa and temporal lobe shows the hypodense areas involving both posterior parts of the temporal lobes and occipital lobes. Abnormal enhancement demonstrated. The intra-cranial vessels are well enhanced. No abnormal fluid collection seen.

The picture is compatible with encephalitis involving both temporal lobes, occipital lobes and left frontal lobe. Herpes encephalitis has to be considered.'

Of note, the CT scan did not show any evidence of haemorrhage or other traumatic brain injury.

On 24.10.1990, there was a note from the nursing staff that Patrick was persistently irritable with frequent episodes of staring, during which his pupils were dilated and his legs extended.

On 25.10 (P563, also P590) Dr. Wilkinson wrote a note in which he recorded '*the seizure disorder is certainly pernicious. I have shown the CT to John ?Bean ?Dean who agrees about a very definite abnormality in occipital lobes- possibly to do with H. simplex but not typical and certainly raising the possibility of a metabolic disorder*.'

There was a plan to send additional blood tests including lysosomal enzymes, long chain fatty acids and a urine metabolic screen. Patrick also underwent a rectal biopsy. All of these tests were negative (P618-619).

Dr. Joseph Dezordi provided an expert certificate in this matter at the St Kilda Police Station on 17 March 2000 (P74-79). Dr. Dezordi was working as the paediatric night resident at the Mater Hospital in Newcastle on 18 October 1990.

Dr. Dezordi reported that at about 5 am that day, he was called to an emergency in the casualty department, where he examined Patrick Folbigg. The history given at that time was that he had been seen by his mother at 3 a.m, at which time Patrick he was coughing. At 4:30 a.m. Patrick's mother heard him asking and found him blue around the lips. He was lifeless and floppy and making minimal respiratory effort. Soon after this he gave a high-pitched cry. Cardiopulmonary resuscitation was not attempted. The paramedics arrived approximately 20 minutes later, and administered oxygen.

Dr. Dezordi administered further oxygen and noted that after about 15 minutes Patrick became more alert, and remained so even when the oxygen was removed, suggesting that the problem was not primarily respiratory.

Dr. Dezordi reported that Patrick was an appropriately-grown male infant who was arching his back. There were no signs of upper airway obstruction or aspiration. Dr. Dezordi felt that there was no evidence of trauma or other injuries, and no signs to suggest any other serious illness. Blood tests were normal but there was significant glycosuria in the absence of hypoglycaemia, suggesting to Dr. Dezordi the possibility of an acute asphyxiating event, 'possibly a seizure of some kind'.

During the early period in hospital Patrick vomited three times, but had no respiratory distress with this. Dr. Dezordi felt that the chest x-ray was normal, although it was later reported as showing signs consistent with bronchiolitis.

Dr. Dezordi next encountered Patrick at 6 a.m. on 20th October 1990, by which time 'it was well-established that he was having frequent seizures in hospital'. Dr. Dezordi noted then that Patrick was fitting, and that his eyes were deviated to the right.

Dr. Dezordi subsequently noted in the Mater Hospital medical notes that a CT scan on 24 October 1990 had shown a pathological process involving the occipital and temporal lobes of the brain. The cause of these CT findings was unclear.

Dr. Dezordi organised a repeat CT scan on 5 November 1990, which showed that the abnormalities seen on the previous scan seemed to have worsened. He interpreted the second scan as showing a progressive 'loss of brain substance'.

Dr. Dezordi was asked by Dr. Ian Wilkinson, consultant neurologist, to forward the scans to an expert radiologist in Sydney for a second opinion. Dr. Dezordi then noted that on the afternoon of Wednesday, 21 November 1990 he called Prof. Merle De Silva, a consultant radiologist at the Children's Hospital at Westmead, about this CT scan. Dr. DeSilva reportedly stated that he did not feel that the CT findings were suggestive of encephalitis, and asked Dr. Dezordi if he had considered that the baby might have been subjected to child abuse.

An **interim discharge summary** from the hospital admission of 18.10-29.10.90 (P80, P632) recorded final diagnoses of intractable seizures, probable viral encephalitis and bronchiolitis. It was noted that while in hospital Patrick had developed generalized and focal seizures, associated with low-grade fevers, and had required large doses of intravenous phenobarbitone and phenytoin for seizure control. He had received seven days of intravenous acyclovir, and had been seizure-free from the third day of acyclovir treatment. Investigations undertaken during the admission included CSF examination which was felt to be normal. The CSF herpes culture was negative. The serum herpes IgM was pending at the time of discharge (but was subsequently negative). The serum lactate was mildly elevated (1.6 mmol/L) on 20th October, whilst a blood ammonia taken on the same day was 16 (normal 29-57 µmol/L). A head ultrasound was felt to be normal, whilst a cranial CT scan showed hypodense areas in the temporal and occipital lobes possibly consistent with encephalitis or demyelination. It was recorded that an EEG had shown left frontal lobe discharges and a chest x-ray slight hyperinflation and increased markings in keeping with bronchiolitis. A metabolic work-up and rectal biopsy were pending; the urine metabolic screen was negative while the serum lactate, ammonia, calcium, magnesium and glucose were normal.

Patrick was discharged home on treatment with phenytoin and phenobarbitone.

Blood was sent on 25.10.1990 for leucocyte enzyme analysis (for lysosomal disorders) and measurement of the very long chain fatty acids (for peroxisomal disorders) (P684). These profiles - which were relatively restricted in 1990 compared to those undertaken now - were negative.

The urine mucopolysaccharide screen and plasma carnitine were normal (P685).

Dr Wilkinson wrote to Dr Morris on 30 October 1990 (P633-634), describing a baby who presented "*with what sounded initially like apnoea, but he subsequently in the ward demonstrated that he was clearly having seizures, mainly right sided. On examination I could not find any neurological problem. His tone and deep tendon reflexes are normal, and he appears active and interested. There was some suggestion of the right side not functioning as well as the left, but the signs are not marked.*"

Dr Wilkinson noted that a number of investigations were still outstanding at that time, and felt that although Herpes encephalitis could not be ruled out absolutely, the changes on the CT scan suggested the possibility of a metabolic disorder.

Patrick was re-admitted to hospital from 4-10.11.1990 with '*recurrent seizures resembling an oculogyric crises (sic*)'. He had conjunctivitis, cough and a rash (P650), and was described on arrival of having episodes of tonic upward eye deviation, without jerking of the extremities or abnormalities of truncal or axial tone. His eyes remained deviated for about an hour, and then - shortly after administration of Panadol- returned to normal. Between episodes, his reflexes and tone were felt to be normal (P651).

On P660, the progress note from the Mater Emergency Department from 4 November 1990 includes a note from Dr Robert Smith suggesting that the diagnosis was '*oculogyric crises ?post encephalitis BG (basal ganglia) involvement'*. Dr Smith was at that time a paediatric registrar. He has subsequently become a paediatric neurologist.

Patrick was felt to be having seizures precipitated by episodes of illness or fever. His full blood count, electrolytes, blood glucose and calcium were essentially normal. A repeat lumbar puncture on 4 November 1990 was normal.

A repeat CT scan of the brain on 5.11.1990 (P640) was reported as showing '*mild generalized widening of the subarachnoid space'* without dilatation of the ventricles. There was high signal in both occipital lobes. The grey-white matter differentiation was otherwise normal. The post-contrast views showed abnormal enhancement in both occipital lobes, which was described as patchy in areas and distributed in both grey and white matter. It was felt that the high density seen in the pre-contrast scan might be due to dystrophic calcification, while the contrast enhancement might be '*post inflammatory*'.

Dr. Morris wrote to the general practitioner on 5.11.90, noting that Patrick remained on anticonvulsant therapy and had been seizure-free, at that time, for 10 days. He remained *'bright and alert and reasonably contented'*. There was further written addendum on the letter that Patrick had been readmitted to hospital on 4.11.90 with 'oculo-gyric episodes.'

On 6 November 1990 Dr. Morris recorded in the progress notes that Patrick was, according to his mother, '*his usual self*'. The nursing staff on the same day, however, noted that he was irritable at times, appeared vacant at others, and did not make eye contact with nursing staff (P654).

An eye swab taken on 5.11.1990 was positive for adenovirus infection (P677).

A chest X-ray on 5.11.1990 was normal.

A repeat EEG on 5.11.1990 – for which the indication was '*recurrent fits, focal fits and oculogyric episodes'* – was reported by Dr J.T. Holland as showing asymmetric slowing (right>left), with frequent multifocal spikes/polyspike activity throughout both hemispheres. The sleep transients were asynchronous, but were overall symmetrical and well-formed. A brief electrographic seizure was recorded from the left parietal region towards the end of the EEG.

Dr. Holland commented that '*…there does appear to have been some deterioration in the record since the previous two. Review of the original one is again absolutely normal. The second one, I think,*

is borderline and this one frankly abnormal. The picture suggests an ongoing encephalopathic process.'

Dr. Smith wrote a further note regarding a re-admission via the emergency department on 14 November 1990 (P712-713). In that note, Dr. Smith reviewed the medical history as summarised above. On 14 November, he recorded that Patrick had had worsening rhinorrhea. His mother had left him for five minutes on the couch. When she returned, his eyes were open and '*looking up into head".* He had stridulous breathing. He then vomited. After this he was sleepy. By the time of arrival to the emergency department he was pale. He had increased tone in his left arm. His tone was otherwise normal/high. His strength was normal and his reflexes symmetrically normal. Dr. Smith noted that Patrick did not fix or follow, but that he reacted to sound. His face moved symmetrically, but Dr. Smith wondered if he had a "? droopy left lid". Dr. Smith felt that Patrick's optic discs possibly had a 'bluish tinge'. Patrick's examination was otherwise normal other than perianal thrush. His head circumference on this admission was 44.1cm.

Dr. Smith concluded that Patrick had had a seizure secondary to sleep deprivation and an upper respiratory tract infection. He noted that he was clinically anaemic and wondered if he was blind.

Dr. Smith noted that Patrick's full blood count on admission showed a haemoglobin of 10.2 g/L, and wondered if Patrick might be iron deficient. An ESR (erythrocyte sedimentation rate) was 58mm/h, which was markedly elevated. A repeat cerebral lactate was felt to be slightly elevated at 1.6 mmol/L; this normalized on repeat testing two days later. A blood gas that day was essentially normal, showing a pH of 7.37.

On 16.11 Dr. Dezordi wrote in the progress notes that Patrick was alert and responsive but not reacting to visual stimuli (P715). An echocardiogram was normal.

On 19 November a medical resident wrote a note regarding "staring attacks" (P718), in which Patrick became quiet and stiff, his eyes staring straight ahead, toes pointed and arms and legs held rigid. This lasted a few seconds and were followed by crying and arching. The episodes ceased after Panadol and anticonvulsants were given.

Further blood tests showed therapeutic anticonvulsants levels - Patrick was at that time being treated with carbamazepine and phenobarbitone. His repeat ESR on 20 November (P719) remained elevated at 35. His haemoglobin was 10.1. The samples for planned iron studies were insufficient.

Patrick was seen for an ophthalmology review on 21 November by Dr. Challinor (P720, P767). Dr. Challinor noted that Patrick's mother felt that he had had normal visual reactions until about a month before this assessment. It was noted that he was not visually responsive and did not fix or follow, but that he had continual ocular movements. Dr. Challinor further noted "*These were not nystagmoid or roving in nature but consisted of changes of conjuate gaze direction in a random manner. Patrick did not suppress his vestibular nystagmus*". Patrick's ocular examination was felt to be normal; the discs were normal with normal maculae and retinas. His visual loss was therefore ascribed to cortical visual disturbance rather than a structural ophthalmological abnormality.

A blood test for leukocyte inclusions was negative (P735, P741).

Testing for congenital (TORCH) viral infections was negative (P742).

X-rays of the cervical spine and skull base obtained on 22 November were normal. (P770)

Dr. Smith completed an interim discharge summary on 22 November 1990 (P769) in which he noted that Patrick had been admitted with a seizure disorder and visual disturbance which he attributed to occipital infarction. He also noted that this did not appear to be a progressive condition, despite the relatively recent diagnosis of visual loss.

Dr. Wilkinson wrote to Dr. Marley on November 30, 1990, and reported that Patrick had had further seizures, with a repeat CT confirming further changes. Dr. Wilkinson noted *'There was some concern about the possibility of a degenerative disease, but John Bear felt that this was probably just vascular. We sent them down to Camperdown Children's Hospital to get another opinion just to be certain, in the opinion was the same as John's. Basically it looks as though there has been some impairment of the blood supply in the basilar territories. I believe that parents had some concern that this might have taken place during that prolonged seizure, but in fact he already had changes in that area at the time of the first presentation. I am not really quite sure what caused this problem, but all of our tests for degenerative disease have come back negative.'*

Patrick again presented to hospital on 22 December 1990 at which time the admission diagnosis was 'Oculogyric crisis' (P780). Dr. Smith's admission note on that date described Patrick as having a history of encephalitis with generalized seizures and occipital infarcts. He presented with an episode of prolonged upward gaze in association with a viral illness and fever. It was noted that he had had a previous episode of tonic upgaze. His discharge summary described him as having a "*post-encencephalitic basal ganglia problem provoked by fever'.* There was a plan that any further episodes of tonic upgaze during this admission should be treated with benztropine (Cogentin), but there were none (P787).

Patrick underwent a physiotherapy assessment on 14 January 1991 (P796) on which he was found to have increased muscle tone, more so in the lower limbs in the upper limbs, and more on the left than the right. He had a residual plantar grasp bilaterally. He had immature reactions to changes in position, and had not yet developed good righting responses. It was noted that he was not visually responsive. It was felt that his hearing was normal. He could roll to the left and the right and when positioned in a sitting position could support his head. He could sit independently when placed but was not yet getting to sit himself, or crawling. He could reach for objects with either hand. When held in supported standing, he tended to go up onto his toes and bounce.

A later note from the same physiotherapist (date obscured, P801) noted a marked improvement in Patrick's visual responsiveness; he was felt to be fixing and following thorough 180 degrees.

An emergency department note (P507) recorded that Patrick presented on 13 February 1991 at 10:20am in asystolic arrest. The preceding night he had had a possible seizure, and a mildly increased temperature. At 07:30 on the morning of 13 February he was put back to sleep and was well. At around 09:30 am his mother found him unresponsive. She called her husband, who rushed home from work. Patrick had no heartbeat and was not breathing. CPR was commenced and an ambulance phoned. When the paramedics arrived, there was no heartbeat and Patrick was not breathing. His colour was good and he was described as still being warm.

Patrick arrived at the Mater Hospital casualty department at 10:20. He was asystolic. He was intubated and given adrenaline via his endotracheal tube and intravenously. He was given two doses

of calcium gluconate, followed by two further doses of intravenous adrenaline. At 10:40 resuscitation was ceased and Patrick was pronounced deceased.

The death certificate completed by Dr. Wilkinson gave the cause of death as asphyxia due to airway obstruction of one-hour duration, on the background of epilepsy of four months' duration.

The **autopsy report** (Part 1) dated 14.2.1991 records the clinical diagnosis of encephalopathic disorder leading to intractable seizures, the underlying cause of the encephalopathy being unknown.

The autopsy report describes an external appearance of a normally formed, well-nourished infant weighing 8.57kg, with a head circumference of 44cm and crown-heel length of 77cm. There was no evidence of trauma or other external abnormalities.

The brain weighed 750g (noted normal average brain weight at this age approximately 714g). The brain was fixed for later dissection. The spinal cord looked macroscopically normal and was also fixed for later dissection.

The post-mortem examination identified congestion in the posterior-basal lung segments, with no other macroscopic abnormalities in the heart or lungs. The liver was felt to look congested and weighed 284g (average normal weight at this age 254g). The kidneys were macroscopically normal, as was the skeletal muscle.

Blood was collected for chromosomal analysis, culture, and drug levels. Tissue was stored for further histological and metabolic studies. The chromosomal analysis of the skin biopsy showed a normal male karyotype (46XY).

Post-mortem blood cultures grew E. coli from one bottle and Enterococcus faecalis from another bottle. A third organism was isolated and identified as Enterococcus avium.

Subsequent examination of the brain showed marked shrinking of both occipital lobes which were described as being thinner and more undulated than normal, with widened sulci. The frontal, parietal and temporal lobes showed no macroscopic abnormalities. On sectioning, the grey matter of the visual cortex in both hemispheres was thinner than normal, showing cystic degeneration. The cysts measured 1-2 mm in diameter and were present in a linear pattern at the junction of the grey and white matter. The underlying white matter was whiter and firmer than normal and appeared to be expanded. The affected areas in the right and left occipital hemispheres measured approximately 45x35x35 mm and 35x35x30 mm, respectively. Similar areas of abnormally firm white matter were present in the left frontal and both parietal lobes. The midbrain, pons and medulla and cerebellum were normal, as was the spinal cord.

Histological examination of the brain was reported separately by Dr. Alex Kahn, a pathologist from Sydney. Dr. Kahn reported on 24 June 1991 that he had not found any convincing evidence of neuronal storage disease or leukodystrophy on the brain sections forwarded to him. He felt that the major changes in the brain were old infarcts and gliosis, mostly in the form of old laminar necrosis which was most severe in the parieto-occipital area. He felt that the cerebellar cortex was unaffected. There was some atrophy of neurones in the deeper parts of the cerebrum, cerebellum and brainstem, which he commented could have resulted from the child's epileptic seizures. He also found a light lymphoid infiltrate in the leptomeninges. Dr. Kahn identified a small amount of linear cortical calcification in the occipital region, which he felt to be part of the laminar cortical necrosis.

He saw no evidence of congenital infection and suggested that the distribution of the lesions was unusual for herpes simplex encephalitis; he felt it appeared much more likely to be '*the result of the cardio-respiratory arrest suffered at approximately five months of age'* (P842).
Microscopic examination of the lungs showed no significant abnormalities other than small foci of alveolar collapse in the periphery. The heart, skeletal muscle, liver, spleen, thymus, pancreas, kidneys, thyroid, adrenal, testes and gastrointestinal tract were all normal. The mixed growth on blood cultures taken post-mortem was felt to represent contamination.

The handwritten medical records from the Regional Medical Genetics Unit for Newcastle and northern New South Wales (P1909-P1911) were provided. **Dr. Colley**, a geneticist, first saw Mr. and Mrs. Folbigg on 12.11.1991. Dr. Colley saw them on a number of subsequent occasions, undertaking testing for MCAD. She saw them again after Sarah Folbigg was born on 14.10.1992, and recorded that the Sarah's MCAD screen and UMS were negative.

On 4th of December 1991 Dr. Colley wrote to Dr. Bridget Wilken at the Oliver Latham laboratory in Sydney requesting her opinion as to whether or not further metabolic investigations were warranted (P855).

Dr. Wilken responded on 10 December 1991, in which she noted that samples sent from Patrick in October 1990 had contained no abnormal metabolic substances suggestive of a fatty acid oxidation disorder. Overall she felt that further testing for MCAD was unlikely to be helpful.

Opinion
Patrick was born at term after an uncomplicated pregnancy and delivery. At birth, he appeared a normal child. His medical and developmental progress in the first months of life was unremarkable.

At two months of age Patrick was noted to have torticollis (a tendency to an abnormal head posture often related to shortening of the sterncleidomastoid muscle). No other abnormalities were identified on examination, and he was felt to be otherwise normal at that time.

At five months of age, in October 1990, Patrick had an acute illness associated with initial breathing difficulties. He was admitted to hospital emergently. He was described at that time as being very pale, and lethargic, with a tendency to back-arch.

An initial head ultrasound and EEG were felt to be normal, but within a few days he had developed a refractory focal seizure disorder and had occipital abnormalities on a CT scan, the cause of which was unclear. Extensive investigations for an infectious or metabolic cause of this presentation were unrewarding.

Patrick subsequently developed a refractory seizure disorder requiring ongoing treatment with anticonvulsant medications. His EEG evolved to show multifocal epileptiform discharges, and a repeat CT scan showed some progression of the abnormalities seen on the first scan, with probable calcification in the occipital regions.

In addition to typical epileptic seizures with clonic (jerking) movements, he had at least two atypical episodes with tonic upward eye deviation. These were felt to represent possible oculogyric crises. On at least one occasion consideration was given to treating these episodes with benztropine.

Patrick had slowing of his development related to his poorly controlled epilepsy. By six months of age it was apparent that he had markedly decreased visual responsiveness, although a subsequent note suggested that this possibly fluctuated to some degree. A consultant ophthalmologist felt that he was cortically blind.

Patrick was admitted to hospital on multiple occasions. On no occasion was he reported to have shown any obvious evidence of inflicted injury. His CT scans and ophthalmological examination did not show the changes often seen in children subjected to non-accidental injuries.

In response to your specific questions:

1. ***What is encephalopathy and what are its causes?***
   Encephalopathy is a general term inferring brain disease or damage, or altered mental state. The major symptom of any encephalopathy is an altered level of consciousness or altered mental state. The causes of encephalopathy are protean, but can include infections, hypoxic-ischaemic brain injury, inborn errors of metabolism, trauma, toxic injury, and genetic conditions affecting brain development or other aspects of cerebral function.

2. ***Does the exclusion of a viral cause exclude an encephalopathic process?***
   Viral encephalitis is a relatively common cause of encephalopathy in childhood. Patrick was investigated for herpes encephalitis, the most common and most severe cause of viral encephalitis in the first year of life. This testing included CSF examination on two occasions, viral cultures of the CSF, and testing of the herpes simplex IgM. This was, in 1990, the extent of the investigations possible for herpes encephalitis in early infancy. That testing was negative in Patrick's case, showing no evidence of viral encephalitis.

   Contemporary testing for herpes encephalitis would include PCR studies on the cerebrospinal fluid. These studies are much more sensitive than the testing- by viral cultures and serology - that was available in 1990. Even in 2019, however, that testing is not 100% sensitive or specific for herpes encephalitis. This diagnosis can sometimes be difficult to confirm or exclude with certainty.

   There are many other causes of infantile encephalopathy other than viral encephalitis. Exclusion of a viral cause therefore fails to exclude other causes of infantile encephalopathies. Other causes of refractory infantile epileptic encephalopathies include genetic disorders affecting brain development and metabolism, conditions affecting neurotransmitter synthesis and function, and inborn errors of metabolism affecting other aspects of cerebral function (Pearl 2016).

3. ***What tests, within your knowledge and experience, are available to treating clinicians now to test for the causes of encephalopathy in children that were not available in 1990? Please provide details of and about these tests.***
   Were a child to present in 2019 with an acute life-threatening event of the type that Patrick experienced in November 1990, the first line investigation would be magnetic resonance imaging (MRI). MRI is the optimal imaging modality for identification of structural (developmental) abnormalities in the brain, and for changes suggestive of infection, hypoxic-ischaemic injury, trauma or other injury. MRI gives insight into the extent and severity of such changes. MR imaging was not generally available in Australia in 1990. CT imaging is much less informative, in that tissue definition is very poor with CT scans of the brain.

In an infant presenting in 2019 with refractory seizures and developmental delay/ regression of unknown cause, MR spectroscopy (MRS), a slightly different form of imaging, would also be undertaken for identification of metabolic or other functional changes in cerebral function. Some of these conditions - such as the various disorders of creatine metabolism – can present in early infancy with epilepsy, developmental arrest, neurologic deterioration and drug-resistant seizures. EEG and MRI are not diagnostic. Diagnosis is contingent on MRS and targeted biochemical and/or genetic testing (Leuzzi et al. 2016).

Analysis of the CSF in 2019, in addition to the investigations undertaken in Patrick's case, would include measurement of the CSF amino acids and neurotransmitters. These measurements might identify abnormalities of the biogenic amines suggestive of a monoamine neurotransmitter disorder. Analysis of urine pterins and amine metabolites would also be undertaken. These investigations were not available in 1990. The conditions for which they test were not recognized at that time (Pearl 2016; Ng et al. 2015).

In 1990, Patrick underwent very basic genetic testing in the form of a karyotype – a standard chromosomal analysis. Additional testing which would be undertaken in 2019 would include a chromosomal microarray (molecular karyotype) – a much higher resolution study able to identify more subtle chromosomal changes. Many children with previously undiagnosed neurological conditions are found to have pathogenic changes on microarray (Berg et al. 2017; Howell et al. 2018).

In a case such as Patrick's, investigated in 2019, next generation sequencing (by whole exome sequencing (WES), or, ideally, whole genome sequencing (WGS) - would be undertaken in order to identify a possible genetic epileptic encephalopathy or cardiac genetic condition predisposing to acute life-threatening events (ALTEs) or sudden unexpected death (Berg et al 2017; Howell et al. 2018; Brownstein et al 2018). There are a number of such conditions. Most affect sodium, potassium or calcium channel function in the brain, causing fluctuating symptoms and potentially predisposing to epilepsy or cardiac rhythm disturbances (Holt et al. 2016; Bagnall et al 2016). Many cannot be easily identified other than by targeted or panel genetic testing, and hence were not diagnosable in 1990.

4. ***Assume that whole genome sequencing has been done on Patrick. Are there any mutations that could produce an encephalopathy that might be identified by such sequencing?***

As listed above, there are a number of neurologic conditions causing infantile encephalopathies- possibly associated with epilepsy and fluctuating neurologic symptoms such as hypotonia, weakness, ptosis and oculogyric crises- which were not tested for in Patrick in 1990 but which might be identified by means of whole genome sequencing in 2019. These conditions include but are not limited to disorders of creatine metabolism, alternating hemiplegia of childhood, neurotransmitter disorders, and genetic channelopathies causing infantile encephalopathies and cardiac arrhythmias.

Two particularly relevant examples are alternating hemiplegia of childhood (Jaffer et al. 2015; Sweney et al. 2015) and the genetic disorders of neurotransmitter metabolism (Ng et al. 2015). These conditions often present in infancy with episodic weakness with fluctuating abnormalities of tone and responsiveness. Monoamine neurotransmitter disorders are under-recognized and often misdiagnosed, as they can mimic cerebral palsy and other neurological disorders. 'Red flag' symptoms of monoamine neurotransmitter disorders

include diurnal variation of symptoms, autonomic disturbance, involvement of the eyes (ptosis, oculogyric crisis) and levodopa responsiveness. Patrick was felt have possible ptosis on one occasion, and was suspected to be experiencing oculogyric crises on two occasions. Treatment with Cogentin was proposed on one occasion; treatment with levodopa was not attempted. Neurotransmitter disorders are clinically and genetic heterogeneous (see figure below, from Ng et al. 2015) and were much more challenging to diagnose before next-generation sequencing (by WES or WGS) became available.

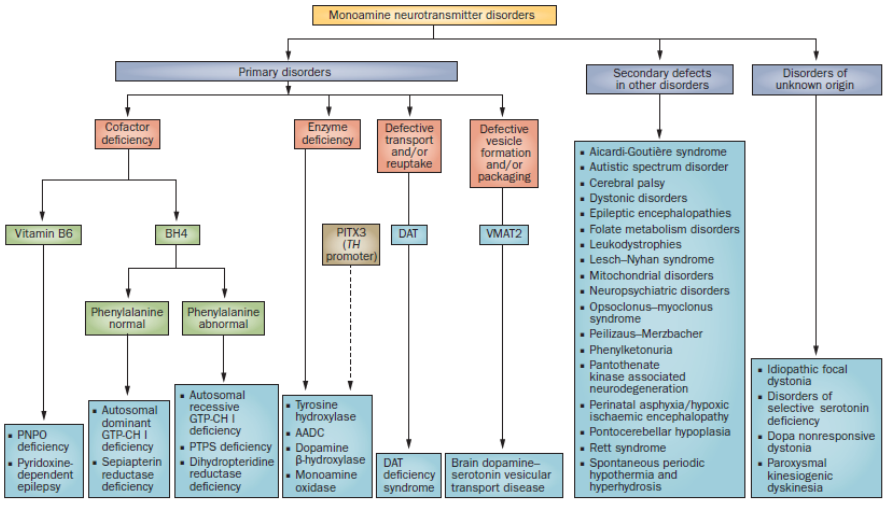


Figure 1 | Overview of primary and secondary monoamine neurotransmitter disorders. Primary disorders of dopamine and serotonin metabolism are attributable to enzyme or cofactor deficiencies, defective neurotransmitter transport and/or reuptake or defective vesicle formation and/or packaging. Neurotransmitter abnormalities are becoming increasingly recognized as secondary phenomena that result from other neurological disorders. Abbreviations: BH4, tetrahydrobiopterin; DAT, dopamine transporter; GTP-CH 1, GTP cyclohydrolase 1; PITX3, pituitary homeobox 3; PTPS, 6-pyruvoyl tetrahydropterin; VMAT2, vesicular monoamine transporter 2.

Figure from Ng et al. 2015

5. *Please read the clinical file provided to you with regards to Patrick. In your opinion, is the clinical and radiological presentation recorded in that material consistent with a single hypoxic episode on 18 October 1990? Please provide reasons for your answer.*
I have reviewed the clinical file provided regarding this case. I am not convinced that Patrick's clinical history is consistent with him having neurologic deficits resulting from a single hypoxic-ischaemic episode occurring on October 18, 1990.

   On that date, when Patrick was first brought to the ED, he was pale and lethargic, but had some back arching. He was hypothermic, tachycardia and tachypnoeic. He was therefore very unwell at the time of presentation. On the same day, however, a head ultrasound and EEG were normal, and within a few hours of admission he was described in the nursing notes as feeding well. Had Patrick sustained a severe hypoxic-ischaemic insult on the morning on 18.10-1990- one sufficiently severe to cause the changes seen on his subsequent imaging and his post-mortem examination- it is difficult to imagine that he would have been able to feed well that day, and that his EEG could have been entirely normal.

   During that admission Patrick was subsequently described as intermittently very irritable, and he developed refractory partial seizures. His lumbar puncture was normal. A repeat EEG on 22.10 showed no slowing of the background, which might be expected with a hypoxic

encephalopathy, but did show focal changes which were left frontal (i.e. not co-localized with the abnormalities seen on the CT of 24.10.1990). Dr Wilkinson reported- relative to that admission; "*On examination I could not find any neurological problem. His tone and deep tendon reflexes are normal, and he appears active and interested."* During the admission, therefore, despite the development of a refractory seizure disorder Patrick was not presenting like a child who had sustained a significant hypoxic-ischaemic insult.

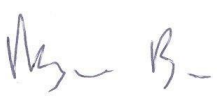
Patrick was readmitted to hospital on 4.11.1990 with what, may have been a seizure but might also have been an oculogyric crisis. This episode consisted of tonic upward eye deviation lasting an hour, without other features of an epileptic seizure. Oculogyric crises- in which there is tonic gaze deviation for long periods of time- are rare in infants but important to recognize because they can reflect an underlying (genetic) disorder of dopamine metabolism (Grattan-Smith 2010). These episodes can also occur after encephalitis- as was postulated in this case- but the CSF and CT findings, and the post-mortem examination, were not consistent with an encephalitis affecting the basal ganglia.

By November 1990 there were concerns regarding Patrick's visual responsiveness. His CT scan showed evidence of cerebral atrophy and more marked changes than were reported on his initial imaging. There was contrast enhancement on both scans. I have not seen these scans but there seems to be at least a suggestion of progression of the radiologic changes, and it appears that his loss of visual responsiveness occurred after the admission of October 1990.

Patrick's physiotherapy assessment of 14.1.1991 documented a child in who there were no fixed severe abnormalities of tone or reflexes- such as would be expected after a significant hypoxic-ischaemic brain injury. The major finding was his visual loss. Isolated visual loss is not a common finding after hypoxic brain injury. On a physiotherapy review some time (but necessarily less than one month) later, Patrick's vision was felt to be much better than had previously been the case. This suggests that he had a fluctuating picture- potentially more consistent with a metabolic or other encephalopathy- rather than a fixed neurologic deficit related to a static hypoxic-ischaemic injury sustained some months earlier.

6. *If, in your opinion, the clinical and radiological presentation recorded in that material in regards to Patrick **is not** consistent with a single hypoxic episode on 18 October 1990, then what is the clinical documentation and records consistent with?*

As described above, there are a number of alternative diagnoses potentially causative of Patrick's neurological condition which have not been excluded by his previous testing. These conditions include but are not limited to disorders of creatine metabolism, alternating hemiplegia of childhood, neurotransmitter disorders, and genetic channelopathies causing infantile encephalopathies and cardiac arrhythmias. All of these conditions can cause progressive neurologic deficit in infancy, can be associated with epilepsy or seizure-like episodes, and can result in the premature death of affected children. Further testing for these conditions would best be accomplished by whole genome sequencing.



Prof Monique Ryan

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Berg AT, Coryell J, Saneto RP, et al. Early-life epilepsies and the emerging role of genetic testing. JAMA Pediatr. 2017;171:863-871.

Brownstein CA, Poduri A, Goldstein RD, Holm IA. The Genetics of Sudden Infant Death Syndrome. In: Duncan JR, Byard RW, editors. SIDS Sudden Infant and Early Childhood Death: The Past, the Present and the Future. Adelaide (AU): University of Adelaide Press; 2018 May. Chapter 31.

Grattan-Smith PJ. Oculogyric crises in infants. J Pediatr Neurol 2010;8:39-40.

Howell KB, Eggers S, Dalziel K, et al. A population-based cost-effectiveness study of early genetic testing in severe epilepsies of infancy. Epilepsia. 2018;59:1177-1187.

Jaffer F, Avbersek A, Vavassori R, et al. Faulty cardiac repolarization reserve in alternating hemiplegia of childhood broadens the phenotype. Brain.2015;138:2859-2874.

Leuzzi V, Mastrangelo M, Battini R, Cioni G. Inborn errors of creatine metabolism and epilepsy. Epilepsia. 2013;54:217-227.

Ng J, Papandreou A, Heales SJ, Kurian MA. Monoamine neurotransmitter disorders-clinical advances and future perspectives. Nat Rev Neurol. 2015;11:567-584.

Pearl PL. Amenable treatable severe pediatric epilepsies. Semin Pediatr Neurol. 2016;23:158-166.

Sweney MT, Newcomb TM, Swoboda KJ. The expanding spectrum of neurological phenotypes in children with ATP1A3 mutations, Alternating Hemiplegia of Childhood, Rapid-onset Dystonia-Parkinsonism, CAPOS and beyond. Pediatr Neurol. 2015;52:56-64.

Appendix:Personal Details (detailed CV available on request)

Professor Monique Ryan is a senior paediatric neurologist with qualifications including Bachelor's degrees in Medicine and Surgery (University of Melbourne, 1991), Fellowship of the Royal Australasian College of Physicians (1998) and a Master's degree in Medicine (University of Sydney 2001). She is a member of the Australia and New Zealand Child Neurology Society.

Prof. Ryan is Director of the Department of Neurology at the Royal Children's Hospital, Melbourne, and holds honorary appointments with the Murdoch Children's Research Institute, Monash University and the University of Melbourne. Her research interests include paediatric neuromuscular disorders and clinical trials of new treatments for neurological disorders of childhood. She has written more than 120 peer-reviewed publications and over 25 book chapters, and is author of a recent textbook on paediatric neuromuscular conditions.

Please be advised that the writer of this report has read Schedule K of the Supreme Court Rules, the Expert Witness Code of Conduct, and agrees to comply with its requirements.

As an expert paediatric neurologist, I have specialized knowledge based on your training, study or experience, which is set out in the Report. My opinion as an expert is wholly based on that specialized knowledge.

EXPERT CERTIFICATE

S177 EVIDENCE ACT 1995

The Expert Certificate is given by me pursuant to s177 of the Evidence Act that the defendant proposes to tender this Expert Certificate concerning my attached report dated which is signed by me as an expert and:

- States my name and address;
- States that I have specialised knowledge based on my training, study or experience as specified in the report attached to this certificate; and,
- Set out an opinion that I hold, and which is wholly or substantially based on that knowledge.

Dated: 15.3.16

Signed:

Name:

Professor Monique M Ryan
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Royal Children's Hospital
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Provider No: 0649634K

PROFESSOR MONIQUE MARIE RYAN

CURRICULUM VITAE

1.3.2019

NAME **MONIQUE MARIE RYAN**

QUALIFICATIONS MB BS, M Med, FRACP

CONTACT DETAILS
Department of Neurology
Royal Children's Hospital
Melbourne, Australia
Phone: 03 9345 4916
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Pager: 0425 763 592

DATE OF BIRTH 20.1.1967

CURRENT APPOINTMENTS
Director, Department of Neurology, The Royal Children's Hospital, Melbourne
Honorary Clinical Professor, Faculty of Medicine, University of Melbourne
Adjunct Clinical Professor, School of Clinical Sciences, Monash University

Major Achievements and Summary

PUBLICATIONS
Professor Ryan has published more than 155 peer-reviewed articles, many in high-impact journals including Nature Genetics, the New England Journal of Medicine, Lancet Neurology and the American Journal of Human Genetics. Her publications have been cited over 900 times and she has an *h-index* of > 29.

Journal publication summary as follows:

	Published (MEDLINE)	In Press	Submitted
Primary Manuscripts	155	2	3
Reviews	12	-	1
Letters	3	-	-
Group studies*	13	1	3

* Listed in paper as part of multinational study group

Top 3 papers based on Journal Impact Factor
Finkel RS, Mercuri E, Darras BT, Connolly AM, Kuntz NL, Kirschner J, Chiriboga CA, Saito K, Servais L, Tizzano E, Topaloglu H, Tulinius M, Montes J, Glanzman AM, Bishop K, Zhong ZJ, Gheuens S, Bennett CF, Schneider E, Farwell W, De Vivo DC; **ENDEAR Study Group**. Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. N Engl J Med. 2017;377:1723-1732.

Wan J, Yourshaw M, Mamsa H, Rudnik-Schöneborn S, Menezes MP, Hong JE, Leong DW, Senderek J, Salman MS, Chitayat D, Seeman P, von Moers A, Graul-Neumann L, Kornberg AJ, Castro-Gago M, Sobrido MJ, Sanefuji M, Shieh PB, Salamon N, Kim RC, Vinters HV, Chen Z, Zerres K, **Ryan MM**, Nelson SF, Jen JC. Mutations in the RNA exosome component gene *EXOSC3* cause pontocerebellar hypoplasia and spinal motor neuron degeneration. Nat Genetics. 2012. 44:704. Impact factor = 36.4

Hogarth MW, Houweling PJ, Thomas KC, Gordish-Dressman H, Bello L; **Cooperative International Neuromuscular Research Group (CINRG**), Pegoraro E, Hoffman EP, Head SI, North KN. Evidence for ACTN3 as a genetic modifier of Duchenne muscular dystrophy. Nat Commun. 2017 Jan 31;8:14143.

Top 3 papers based on number of citations

Ilkovski B, Cooper ST, Nowak K, **Ryan MM,** Yang N, Schnell C, Durling HJ, Roddick LG, Wilkinson I, Kornberg AJ, Collins KJ, Wallace G, Gunning P, Hardeman EC, Laing NG, North KN. Nemaline myopathy caused by mutations in the muscle α-skeletal-actin gene. Am J Hum Genet 2001. 68:1333-1343. Citations = 81

Quijano-Roy S, Mbieleu B, Bönnemann CG, Jeannet PY, Colomer J, Clarke NF, Cuisset JM, Roper H, De Meirleir L, D'Amico A, Ben Yaou R, Nascimento A, Barois A, Demay L, Bertini E, Ferreiro A, Sewry CA, Romero NB, **Ryan M,** Muntoni F, Guicheney P, Richard P, Bonne G, Estournet B. De novo *LMNA* mutations cause a new form of congenital muscular dystrophy. Ann Neurol. 2008;64:177-186. Citations: 80

Burns J, Ouvrier RA, Yiu EM, Joseph PD, Kornberg AJ, Fahey MC, **Ryan MM.** Ascorbic acid for Charcot-Marie-Tooth disease type 1A in children: a randomised, double-blind, placebo-controlled, safety and efficacy trial. Lancet Neurol. 2009. 8:537-544. Citations = 55

Leadership

2015- Director, Department of Neurology, The Royal Children's Hospital
2008- Head, Royal Children's Hospital Multidisciplinary Neuromuscular Clinic
2010-2013, 2015- Member, Executive Board, Cooperative International Neuromuscular Research Group (CINRG) USA)
2010- Executive, Australasian Neuromuscular Network (ANN)
2012- Member, Therapeutics Subcommittee, CINRG (USA)

Research Highlights

- Senior investigator on the first randomised placebo-controlled clinical trial in paediatric Charcot-Marie-Tooth disease (results published in *Lancet Neurol*)
- Site Principal Investigator for 17 industry-driven international clinical trials of novel therapies for Duchenne muscular dystrophy: Translarna/Ataluren, PTC Therapeutics, 2007-2010, 2012-; Prosensa/ GSK, 2010-2012, 2012-2014, Roche 2017-, Reveragen 2016-, Polaris 2018-, Sarepta 2018-.
- Site Principal Investigator for 5 industry-driven international clinical trials of novel therapies for spinal muscular atrophy (Ionis/Biogen ENDEAR 2014-2016; NURTURE 2015-, SHINE 2016-)
  and AveXIs (AveXis 2015-2018).
- Australasian representative, Therapeutics Advisory Committee, TREAT-NMD 2010-
- Chief Investigator, NHMRC Centre of Research Excellence grant awarded $2.5M (2012-2016)
- Associate Investigator, European Union-NHMRC RareBestPractice grant $614K (2013-2016)
- Flagship leader, Melbourne Genomics Health Alliance, 2013-2016

- Associate Investigator, Australian Genomics Health alliance, awarded NHMRC TCR grant of $25m, 2015.

Major External Collaborations

- Site Principal Investigator for numerous international neuromuscular research consortia including TREAT-NMD, the Cooperative International Neuromuscular Research Group, the International Neuropathy Consortium and International Guillian-Barré Outcome Study.
- Principal investigator, Neuromuscular Disorders of Childhood study, Australian Paediatric Surveillance Unit, 2006-2009
- Investigator, Acute Flaccid Paralysis Study, Australian Paediatric Surveillance Unit, 2006-2015

Teaching

Undergraduate

2010-2018 Theme Champion, Muscular Dystrophy, 'Molecules to Malady', University of Melbourne Bachelor of Biomedical Sciences)

Post-graduate

2011-2017 Annual lectures to the Specialist Certificate in Palliative Care, University of Melbourne
2007- Annual lecture series to Victorian paediatric FRACP candidates
2007- Annual bedside and clinical teaching sessions to Part II FRACP candidates
2009, 2014 Orthopaedic Trainee Course, Royal Australasian College of Surgeons
2005, 2008 Australian Association of Neurologists Trainee Update, Canberra Australia
2003-2006 Deputy Head and Subdean of Post-Graduate Research, Discipline of Paediatrics and Child Health, Faculty of Medicine, University of Sydney.

Professional Service

International

2012- Therapeutics Strategy Committee, FSH Global Research Foundation
2009- Australasian Representative, Therapeutics Advisory Committee, TREAT-NMD
2010- Clinical Care Steering Committee, Australasian Neuromuscular Network
2010- Research Steering Committee, Australasian Neuromuscular Network
2007-2014 Scientific Program Committee Australian and New Zealand Association of Neurologists
2007-2014 Paediatric Written Examination Committee Royal Australasian College of Physicians
2011-2014 Executive Board Australia and New Zealand Child Neurology Society
2012 Scientific Programme Committee, 12th International Child Neurology Congress
2012 Planning Committee, 12th International Child Neurology Congress
2009 Planning Committee Asian-Oceanic Myology Congress
2007- 2010 Paediatric Neurology Curriculum Committee, Australian and New Zealand Association of Neurologists
2005-2007 Policy Group for Decision-Making at the End of Life in Children, Royal Australasian College of Physicians

National

2015- Victorian curator, National Myotonic Dystrophy Registry
2015- Victorian curator, National FSHD Registry
2013- Victorian curator, National Spinal Muscular Atrophy Registry
2010- Victorian curator, National Duchenne Muscular Dystrophy Registry
2012, 2014- Reviewer, Paediatric Dosing Resource Australian Medicines Handbook
2006-2015 Australian Polio Expert Committee, Australian Government Dept Of Health and Ageing
2006-2015 Investigator, Acute Flaccid Paralysis Study, Australian Paediatric Surveillance Unit
2009-2010 Principal Committee, National Duchenne Muscular Dystrophy Registry Working Group

Institutional
2009-2012, 2015- Clinical Ethics Case Response Group, Royal Children's Hospital
2008- Board Member, Medical Staff Association, Royal Children's Hospital
2007-2008 Clinical Ethics Management Group, Royal Children's Hospital

CV in detail

FELLOWSHIPS AND MEMBERSHIPS
2000 Fellow, Royal Australasian College of Physicians

UNDERGRADUATE AND POSTGRADUATE DEGREES
2001 Master of Medicine, University of Sydney
Nemaline myopathy: a clinical, pathologic and genetic study
1991 Bachelor of Medicine and Surgery, University of Melbourne

AWARDS / SCHOLARSHIPS
2008 Best Poster Presentation, World Muscle Society, Newcastle UK
2006 Best Oral Presentation (Nerve or Neuromuscular Junction), XIth International Congress on Neuromuscular Disorders, Istanbul
2004 Sylvia and Charles Viertel Charitable Foundation Clinical Investigatorship
2002 Founders' Award for Clinical Research by a Neurologist-in-Training, American Association of Neurologists
2001 Von L Meyer Fellowship, Children's Hospital Boston
2000 Caroline Margaret Duncan Award, Southern Pediatric Neurology Society USA
2000 Von L Meyer Fellowship, Children's Hospital Boston
2000 Outstanding Junior Member, Child Neurology Society USA
1999 Australian Association of Neurologists Young Investigator (Poster) Award
1999 United Medical Protection Society (Australia) Conference Prize
1998 John Yu Scholarship, The Children's Hospital at Westmead
1998 Royal Australasian College of Physicians Travelling Paediatric Fellowship
1989 Community Medicine and Clinical Practice Prize, University of Melbourne

EXTERNAL PEER REVIEWED FUNDED GRANTS

2016	FSHD Global Research Foundation	$203 409
2015	National Health and Medical Research Council TCR in Genomics (AGHA) I.D.1113531 (AI)	$25M
2014	CMT Association of Australia	$10,420
2013-2016	EU FP7 program (Rare Disease Best Practice) (AIG)	$614,128
2013	Brain Foundation	$50,000
2012	Epilepsy Tasmania, The Tasmanian Infantile Epileptic Encephalopathy Project	$80,000
2011-2016	National Health and Medical Research Council Centre for Research Excellence in Neuromuscular Disorders (CIE) I.D. 1031893	$2.5M
2006-2009	The March of Dimes (US)(CID)	US$250,000
2007	Australian Podiatry Education & Research Foundation	$7,980
2007	NSW Health, Podiatrists' Registration Board of NSW	$12,640
2004	GlaxoSmithKline Fellowship in Neurology, Royal Australasian College of Physicians	$25,000
2004	Sylvia and Charles Viertel Charitable Foundation Clinical Investigatorship	$55,000
1999	Muscular Dystrophy Association of Australia	$3,000

INTERNAL PEER REVIEWED FUNDED GRANTS

2014	Murdoch Children's Research Institute	$30,000
2008	Murdoch Children's Research Institute	$10,000
2007	Murdoch Children's Research Institute	$10,000
2006	The University of Sydney Research and Development Scheme (CIB)	$48,000
2004	The University of Sydney Research and Development Scheme ECR Researcher	$47,500
1998	The Children's Hospital Small Grants Committee Grant	$2,500

EDITORIAL/ REVIEWER ROLES

Journals

2018- Associate Editor, Frontiers in Neurology (USA)
2016-2018 Associate Editor, Oxford Journal of Rare Disorders (UK)
2014-2018 Associate Editor, Journal of Paediatrics and Child Health (Australasia)
2008- Editorial Board, Journal of Clinical Neuroscience (Australasia)
2006-2011 Associate Editor, Journal of Pediatric Neurology (Turkey)

***Ad hoc* reviewer**

Annals of Neurology
Annals of Clinical and Translational Neurology
Annals of Indian Academic Neurology
BMC Neurology
Brain and Development
Canadian J Neurol Science
Clinical Allergy and Immunology
Clinical and Developmental Immunology
Clinical Science
Current PLOS
Developmental Medicine and Child Neurology

European Journal of Human Genetics
Future Neurology
Journal of Child Neurology
Journal of Neuromuscular Disorders
Lancet Neurology
Medical Journal of Australia
Muscle and Nerve
Neurology
Neurology- Clinical Practice
Neuromuscular Disorders
Pediatric Pulmonology
Pediatrics
J Paediatrics and Child Health
Sleep Medicine
Therapeutic Advances in Neurological Disorders

Reviewer of Online Resources
Genetics Home Reference resources, National Library of Medicine, U.S. National Institutes of Health

Grant panels
2015 Flanders Medical Research Fund
2015 Daniel Bravo-Andreu Private Foundation (Spain)
2011- Scientific Grants Review Committee, FSHD Global
2010, 2015 Agency for Health Quality and Assessment of Catalonia (Spain)
2014 Health Research Council of New Zealand
2013 Telethon-UILDM (Italy)
2007-2010 Murdoch Children's Research Institute
2009, 2010 Pfizer Neuroscience Research Grants

Conference committees
Australian Neuromuscular Network 2010-
Australian and New Zealand Child Neurology Society 2011- 2014
International Child Neurology Association 2012
Asian-Oceanic Association of Myology 2009

TRAINEE SUPERVISION
Trainees in Paediatric Neurology
Australasian Association of Neurology/ Royal Australasian College of Physicians
2019 Dr. Kate Irving 2015-2018 Dr Ian Woodcock
2013-2015 Dr. Erik Andersen 2012-2013 Dr. Eunice Chan
2011 Dr. Tyson Ware 2010 Dr. Damian Clark
2010-2012 Dr. Katherine Howell 2006-2008 Dr. Eppie Yiu
2006 Dr. Suzanna Maclennan 2005-2006 Dr. Mahendra Moharir
2003-2005 Dr. Helen Young

Basic trainees in Paediatrics
At least one RACP basic trainee supervised annually since 2011

PhD SUPERVISION
2017- Dr Ian Woodcock
2012-2015 Dr Manoj Menezes PhD 2015, University of Sydney

2010-2014 Dr Eppie M Yiu PhD 2014, University of Melbourne
PA fellowship, MCRI Research Fellowship, Stella Mary Langford Scholarship, University of Melbourne, NHMRC Scholarship, Elsevier Prize

2006-2009 Dr Joshua Burns PhD 2006, University of Sydney
NHMRC Australian Clinical Research Fellowship, Fulbright scholarship 2009

2006-2010 Dr Paula Bray PhD 2010, University of Sydney
Douglas and Lola Douglas Scholarship, University of Sydney, John Yu Scholarship, The Children's Hospital at Westmead

MASTERS DEGREE SUPERVISION

2018- Ms. Kate de Valle
2013- 2017 Ms Mary Roberts M Phil, University of Melbourne
2013-2015 Ms Joy Goubran M Sc 2015, University of Melbourne
2004-2010 Dr Simon Grew M Med 2010, University of Sydney
2003-2006 Dr Helen Young M Med 2006, University of Sydney
- APA fellowship

BACHELOR SCIENCE HONOURS STUDENT SUPERVISION

2006-2007 Dr Colleen D'Arcy B Med Sci 2007, Monash University
- MA-EH Embley Memorial Medal

NATIONAL/ INTERNATIONAL TRAINING VISITORS

Dr. Steven DeRoos (USA) 2005 Dr. Iain Horrocks (Scotland) 2005-2006
Dr. Eric Payne (Canada) 2009 Dr. Jane MacLean (USA) 2013
Dr. Trupti Jadhav (India) 2012-2014 Dr. Loudella Callotes-Castillo (Phillipines) 2013-2015
Dr. Sanne Hobbelink (Netherlands) 2015

PUBLICATIONS

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155. **Ryan MM**, Ouvrier RA, Procopis PG. Movement disorder in influenza A encephalitis. Pediatr Neurol 1999;21:669-673.

Review articles

1. Moore GE, Lindenmayer AW, McConchie GA**, Ryan MM**, Davidson ZE. Describing nutrition in spinal muscular atrophy: A systematic review. Neuromusc Disord. 2016;26:395-404.

2. Chan EK, Kornberg AJ, **Ryan MM**. A diagnostic approach to recurrent myalgia and rhabdomyolysis in children. Arch Dis Child 2015;100:793-799.

3. **Ryan MM.** Pediatric Guillian-Barré syndrome. Curr Opin Pediatr 2013;25:689-693.

4. Mohamed A, **Ryan MM**. Neuromuscular complications of intensive care. Handb Clin Neurol. 2013;113:1481-1483.

5. Yiu EM, **Ryan MM**. Demyelinating prenatal and infantile developmental neuropathies. J Peripher Nerv Syst. 2012;17:32-52.

6. Yiu EM, **Ryan MM**. Genetic axonal neuropathies and neuronopathies of pre-natal and infantile onset. J Periph Nerv Syst 2012;17:285-300.

7. Cardamone M, Darras BT, **Ryan MM**. Inherited myopathies and muscular dystrophies. Semin Neurol. 2008;28:250-259.

8. Ouvrier RA, Geevasinga N, **Ryan MM**. Autosomal recessive and X-linked forms of hereditary motor and sensory neuropathy in childhood. Muscle Nerve 2007;36:131-143.

9. Williams S, Horrocks IA, Ouvrier RA, Gillis J, **Ryan MM**. Critical illness polyneuropathy and myopathy in pediatric intensive care: a review. Pediatr Crit Care Med 2007;8:18-22.

10. Burns J, Landorf K, Ouvrier R, Crosbie J, **Ryan MM**. Interventions for the treatment of pes cavus.

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11. **Ryan MM**, Ouvrier R. Hereditary peripheral neuropathies of childhood. Curr Opin Neurol 2005;18:105-110.
12. R**yan MM**. Guillain-Barré syndrome in childhood. J Paediatr Child Health 2005;41:237-241.
13. **Ryan MM**, Engle EC. Acute ataxia in childhood. J Child Neurol 2003;18:309-316.

Books
1. Darras BT, Jones HR Jr, **Ryan MM**, De Vivo DC (editors). Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach. 2nd edition. San Diego: Elsevier, 2015.
2. Associate Editor. Netter's Neurology. Jones HR (ed). Icon Learning Systems, Teterboro, NJ 2005. (2nd edn. 2009, 3rd edn 2013).
3. Contributing Editor. The Complete Parenting Guide. The Children's Hospital at Westmead. Focus Books, 2005.

Book chapters
1. **Ryan MM,** Muntoni F, North KN. Muscle Disorders. In: Arzimanoglou A, O'Hare A, Johnston MV, Ouvrier RA (eds). Aicardi's Diseases of the Nervous System in Childhood. MacKeith Press, London, 2018.
2. Nelson's chapter x4
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6. Gillis J, **Ryan MM.** Chronic neuromuscular disease. In: Rogers' Textbook of Pediatric Intensive Care, 5th Edition. Nichols DG (ed). Lippincott Williams Wilkins, Baltimore, 2015.
7. **Ryan MM**, Engle EC. Disorders of the Ocular Motor Cranial Nerves and Extraocular Muscles In: Darras BT, Jones HR Jr, **Ryan MM**, De Vivo DC (eds). Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach. 2nd edition. Elsevier, San Diego, 2014.
8. Jones HR Jr., Grattan-Smith PJ, **Ryan MM**. Acute Polyneuropathies. In: Darras BT, Jones HR Jr, **Ryan MM**, De Vivo DC (eds). Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach. 2nd edition. Elsevier, San Diego, 2014.
9. McMillan HJ, **Ryan MM**. Overview of Pediatric Peripheral Neuropathies. In: Darras BT, Jones HR Jr, **Ryan MM,** De Vivo DC (eds). Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach. 2nd edition. Elsevier, San Diego, 2014.
10. **Ryan MM**, Jones HR. Mononeuropathies. In: Darras BT, Jones HR Jr, **Ryan MM,** De Vivo DC (eds). Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach. 2nd edition. Elsevier, San Diego, 2014.
11. Jones HR Jr, **Ryan MM**, Levin KH. Radiculopathies and Plexopathies. In: Darras BT, Jones HR Jr, **Ryan MM,** De Vivo DC (eds). Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach. 2nd edition. Elsevier, San Diego, 2014.
12. De Vivo DC, Darras BT**, Ryan MM**, Jones HR Jr. Introduction: Historical Perspectives. In: Darras BT, Jones HR Jr, **Ryan MM**, De Vivo DC (eds). Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach. 2nd edition. Elsevier, San Diego, 2014.
13. **Ryan MM**, Kornberg AJ. Neuromuscular Disorders. In: Practical Paediatrics 7th edn. South M, Isaacs D (eds). Churchill Livingstone, Melbourne, 2012.
14. Tracy JA, **Ryan MM,** Engelstad J, PJB Dyck. Peripheral Nervous System. In Netter's Atlas of Neurology. Jones HR, Burns TM, Aminoff M, Pomeroy S (eds). Elsevier, Philadelphia, 2011.
15. **Ryan MM**. Brachial and lumbosacral plexopathies. In: Netter's Neurology 2nd ed. Jones HR, Baker R, Srinivasan J, Allam G. (eds). Elsevier, Philadelphia, 2011.
16**. Ryan MM**. Hereditary polyneuropathies. In: Netter's Neurology 2nd ed. Jones HR, Srinivasan J, Allam G, Baker R, (eds.) Elsevier, Philadelphia, 2011.
17. Mohamed A, **Ryan MM.** Neuromuscular complications of intensive care. In: Paediatric Neurology (Part III). Handbook of Clinical Neurology Series, 3rd edn. Dulac O, Lassonde M, Sarnat H. (eds) Elsevier

2013.
18. **Ryan MM**, Pollard G, Ouvrier RA. Inflammatory neuropathies. In: Inflammatory and autoimmune disorders of the nervous system in children. In: Dale RC, Vincent A (eds). MacKeith Press UK 2009.
19. **Ryan MM.** Congenital muscular dystrophies. In: Current Management in Child Neurology (4th edn). Maria BL, (ed). BC Decker, Shelton 2009.
20. **Ryan MM**. Neonatal hypotonia. In: Current Management in Child Neurology (4th edn). Maria BL (ed). BC Decker, Shelton 2009.
21. Gillis J, **Ryan MM.** Chronic neuromuscular disease. In: Rogers' Textbook of Pediatric Intensive Care, 4th Edition. Nichols DG (ed). Lippincott Williams Wilkins, Baltimore, 2008.
22. **Ryan MM,** North KN. Congenital myopathies. In: Neurological Therapeutics, Principles and Practice. Noseworthy J (ed). Martin Dunitz, Rochester, 2006.
23. Ouvrier RA, **Ryan MM,** Redmond A. Treatment of peripheral neuropathies. In: Treatment of Pediatric Neurologic Disorders. Singer HS, Kossoff EH, Hartman AL, Crawford TO (eds). Taylor and Francis, Boca Raton, 2005.
24. **Ryan MM,** North KN. Congenital myopathies. In: Treatment of Pediatric Neurologic Disorders. Singer HS, Kossoff EH, Hartman AL, Crawford TO (eds). Taylor and Francis, Boca Raton 2005.
25. **Ryan MM**, Kuntz N, Burns TM. Autonomic testing in childhood. In: Clinical Neurophysiology of Infancy, Childhood and Adolescence. Holmes GL, Moshé S, Jones HR Jr (eds). Butterworth Heinemann, Philadelphia, 2005.
26. Burns TM, **Ryan MM**, Darras BT, Jones HR Jr. Peripheral neuropathies in infants and children. In: Peripheral Neuropathy. Dyck PJ, Thomas PK (eds). 4th edn. Elsevier Saunders, Philadelphia, 2005.
27. **Ryan MM**, Burns TM, Russell JA, Jones HR Jr. Multifocal neuropathies. In: Netter's Neurology. Jones HR (ed). Icon Learning Systems, Teterboro, 2005.
28. **Ryan MM**, Russell JA. Distal predominant myopathies. In: Netter's Neurology. Jones HR (ed). Icon Learning Systems, Teterboro, 2005.
30. **Ryan MM,** Russell JA. Myopathies presenting with exercise intolerance. In: Netter's Neurology. Jones HR (ed). Icon Learning Systems, Teterboro, 2005.
31. **Ryan MM,** Russell JA. The channelopathies: myopathies presenting with episodic weakness. In: Netter's Neurology. Jones HR (ed). Icon Learning Systems, Teterboro, 2005.
32. **Ryan MM**. Lumbosacral plexopathies. In: Netter's Neurology. Jones HR (ed). Icon Learning Systems, Teterboro, 2005.
33. Burns TM, **Ryan MM**, Jones HR. Myasthenia gravis. In: Netter's Neurology. Jones HR (ed). Icon Learning Systems, Teterboro, 2005.
34. **Ryan MM**. Neuromuscular junction anatomy and physiology. In: Netter's Neurology. Jones HR (ed). Icon Learning Systems, Teterboro, 2005.
35. Escolar DM, **Ryan MM,** Jones HR. Lower extremity mononeuropathies. In: Neuromuscular Disorders of Infancy, Childhood and Adolescence. Jones HR, Darras BT, DeVivo DV (eds). Butterworth Heinemann, Philadelphia, 2002.
36. **Ryan MM,** Stasheff SF, Engle EC. Disorders of the ocular motor cranial nerves and extraocular muscles. In: Neuromuscular Disorders of Infancy, Childhood and Adolescence. Jones HR, Darras BT, DeVivo DV (eds). Butterworth Heinemann, Philadelphia, 2002.
37. Andrews PI, **Ryan MM,** Kandt RS. Genetic causes of pediatric stroke. In: Genetics of Cerebrovascular Disease. Alberts MJ (ed). Futura Publishing Company, Inc. Armonk 1999.

INVITED PRESENTATIONS

International meetings/conferences

2019 European Neomuscular Consortium, Amsterdam
2018 Neurodevelopmental and Behavioural Paediatric Society of Australasia
2016 Australia and New Zealand Child Neurology Society, Auckland
2015 Asian-Oceanic Congress of Child Neurology, Taipei, Taiwan
2015 10th International Conference, Improving the Use of Electromyography in Paediatrics, UK
2015 Australia and New Zealand Child Neurology Society, Melbourne

2015 MDA New Zealand
2013 9th International Conference, Improving the Use of Electromyography in Paediatrics, UK
2012 Australasian Podiatry Conference, Melbourne
2011 International Child Neurology Congress, Brisbane
2012 Muscular Dystrophy Association of Australia, Sydney
2011 8th International Conference, Improving the Use of Electromyography in Paediatrics, UK
2011 Australia and New Zealand Association of Neurologists, Hobart Australia
2011 Australasian Podiatry Conference, Melbourne Australia
2010 Peripheral Nerve Society, Sydney Australia
2010 International Child Neurology Congress, Cairo Egypt
2009 Asian-Oceanic Myology Centre, Melbourne Australia
2008 Cooperative International Neuromuscular Research Group, Washington DC
2007 6th International Conference, Improving the Use of Electromyography in Paediatrics, UK
2006 International Child Neurology Congress, Montréal Canada
2005 5th International Conference, Improving the Use of Electromyography in Paediatrics, UK
2004American Association of Clinical Neurophysiologists, New Orleans, USA
2004 Brazilian Paediatric Neurology Society, Sao Paolo Brazil
2003 European Neuromuscular Consortium, Naarden
2002 Harvard Neurology Training Program
2002 Michael J Bresnan Child Neurology Update, Harvard Medical School
2001 Michael J Bresnan Child Neurology Update, Harvard Medical School
2000 Michael J Bresnan Child Neurology Update, Harvard Medical School
2000 European Neuromuscular Consortium, Naarden

National meetings/conferences

2018 Neurodevelopmental and Behavioural Paediatric Society of Australasia
2019 Australasian Neuromuscular Network, Melbourne
2017 ACTT-DMD Sydney
2017 National Metabolic Group, Melbourne Australia
2016 Australasian Neuromuscular Network
2015 RCH research week
2014 RCH clinical trial update
2013 Murdoch Children's Research Institute, Melbourne Australia
2013 RCH Clinical Trial Update
2011-2013 Victorian Palliative Care Service
2011 Murdoch Children's Research Institute, Melbourne Australia
2011 Grand Rounds, Royal Children's Hospital, Melbourne Australia
2011 St Vincent's Hospital Neurology Grand Rounds, Melbourne Australia
2011 Awakening Australia to Rare Diseases, Perth Australia
2011 Australian Neuromuscular Network, Perth Australia
2010 MD2010, Perth
2009 Parent Project Australia, Sydney
2009 Orthopaedic Trainee Course, Royal Australasian College of Surgeons
2009 Clinical Neurophysiology Workshop, Australia and New Zealand Association of Neurologists
2009 Royal Australian College of Physicians Congress
2008 Australasian Sleep Association, Adelaide
2006-2008Australia and New Zealand Association of Neurologists Trainee Update
2007 Austin Neurosciences Grand Rounds, Heidelberg Victoria
2007 Australia and New Zealand Association of Neurologists Neuromuscular Update
2006 Parent Project Australia 'Turning the Tide for Muscular Dystrophy' Brisbane
2005 Australia and New Zealand Association of Neurologists Trainee Update
2005 Susan Ryan Seminar, Parramatta NSW

2004 Muscular Dystrophy Association of NSW, Haberfield
2003-2006 Charcot-Marie-Tooth Association of NSW, Concord NSW
2004 Blacktown-Mt Druitt Health Service Paediatric Update, Blacktown NSW
2004 Children's Hospital at Westmead Postgraduate Weekend for General Practitioners
2004 Children's Hospital at Westmead Paediatric Update

BOARDS

2010-2013, 2015- Executive Board, Cooperative International Neuromuscular Research Group
2010- Executive, Australasian Neuromuscular Network
2003-2006 Executive Board, Institute for Neuromuscular Research
2003-2008 Board, Parent Project Australia

TEACHING/MEDICAL EDUCATION

Undergraduate

2010- Theme Champion, Muscular Dystrophy, 'Molecules to Malady' University of Melbourne Bachelor of Biomedical Science.

Post-graduate

2011- Annual lectures to the Specialist Certificate in Palliative Care, University of Melbourne
2007- Annual lecture series to Victorian paediatric FRACP candidates
2007- Annual bedside and clinical teaching sessions to Part II FRACP candidates
2009, 2014 Orthopaedic Trainee Course, Royal Australasian College of Surgeons
2005, 2008 Australian Association of Neurologists Trainee Update, Canberra Australia
2003-2006 Deputy Head, Post-Graduate Research, Discipline of Paediatrics and Child Health, Faculty of Medicine, University of Sydney.

ADDITIONAL TRAINING

2010 AMA4 Impairment Assessment Training Program: Core (Stream 1)

MEDIA

3.2019 Newshub (NZ): Spinraza licensing for SMA
9.2018 7.30 Report, ABC TV: Duchenne musucular dystrophy
7.2018 7.30 Report, ABC TV: Spinraza for SMA
8.2017 The Age: New treatments for SMA
3.2017 Sydney Morning Herald: New treatments for SMA type 1
5.2016 The Australian: Technology and brain development
4.2016 The Age, The Herald Sun, ABC News, Channel 7 news: Medicinal cannabis
2.2015 The Age, the Herald-Sun, Red Symons 774: Enzyme replacement for Pompe disease
2.2014 3RRR, Melbourne
2012 Catalyst, ABC: Vitamin C for CMT1A

COMMUNITY PARTICIPATION

2016 Year 12 Fintona Girls School
2015 Oak Parlour program, Trinity College, University of Melbourne
2010 Duchenne Foundation, Sydney Australia
2010- Annual Australasian Neuromuscular Network family information days
2007 CMT Association of Tasmania
2006- Annual presentations to Muscular Dystrophy Association Australia
2003-2007 Annual lectures Charcot-Marie-Tooth Association of NSW
2006 Parent Project Australia
2005 Susan Ryan Seminar, Parramatta NSW

2005-2008 Annual meetings, Muscular Dystrophy Association of NSW

PREVIOUS EMPLOYMENT HISTORY

2006-2015 Senior Staff Specialist, Department of Neurology, Royal Children's Hospital
2005-2006 Senior Lecturer, Discipline of Paediatrics and Child Health, University of Sydney
Deputy Head, Post-Graduate Research, Paediatrics and Child Health, University of Sydney
2003-2006 Conjoint Senior Lecturer, Discipline of Paediatrics and Child Health, University of Sydney
Staff Specialist, Department of Neurology, The Children's Hospital at Westmead, Sydney
2002-2003 Clinical Associate, Harvard Medical School Boston and Tufts University, Boston
Neurophysiology Fellow, Lahey Clinic and Children's Hospital Boston
1999-2002 Clinical Fellow, Harvard Medical School, Boston
1998-1999 Clinical Associate Lecturer, Paediatrics and Child Health, University of Sydney
1999 Research Fellow, Paediatric Neurology, The Children's Hospital at Westmead, Sydney
1998 Clinical Fellow, Paediatric Neurology, The Children's Hospital at Westmead, Sydney
1996-1998 Paediatric Registrar, The Children's Hospital at Westmead, Sydney
1994-1996 Paediatric Registrar, Sydney Children's Hospital
1993 Paediatric Resident, Royal Children's Hospital, Melbourne
1992 Internship, Austin Hospital, Melbourne

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Email: stuart.gray@cardillograypartners.com.au

12 March 2019

Professor M Ryan
Consultant Neurologist
Royal Children's Hospital Melbourne

Via email only: monique.ryan@rch.org.au

Dear Professor,

Re: Kathleen Folbigg

We advise that we act on behalf of Kathleen Folbigg.

Procedural matters

We draw your attention to the following Court Rules, copies attached:

1. UCPR 31.23 Code of conduct
2. UCPR 31.27 Experts' Reports
3. UCPR Schedule 7 - Expert witness code of conduct

Please note that in order to be admissible at the hearing your report should comply with the following matters:

1. Address your report to this Firm and refer to these instructions;
    a. Your report must state:
    b. Your name and address;
    c. The matters set out in UCPR 31.27 - that as an expert you have specialised knowledge based on your training, study or experience, which is set out in the Report; and sets out the opinion that you hold as an expert, and which is wholly or substantially based on that specialised knowledge.

2. Attach a copy of this letter, the letter of instruction and its attachments (we will arrange for a copy of the attachments to be annexed to avoid such cumbersome exercise having to be undertaken by you) to your report;
3. Complete and attach the Certificate - Expert Report;
4. Complete and attach the Expert Certificate, s177 Evidence Act.

General

1. What is encephalopathy and what are its causes?
2. Does the exclusion of a viral cause exclude an encephalopathic process?
3. What tests, within your knowledge and experience, are available to treating clinicians now to test for the causes of encephalopathy in children that were not available in 1990? Please provide details of and about these tests.
4. Assume the whole genome sequencing has been done on Patrick. Are there any mutations that could produce an encephalopathy that might be identified for such sequencing?
5. Please read the clinical file provided to you with regards to Patrick. In your opinion, is the clinical and radiological presentation recorded in that material consistent with a single hypoxic episode on 18 October 1990? Please provide reasons for your answer.
6. If, in your opinion, the the clinical and radiological presentation recorded in that material in regards to Patrick **is not** consistent with a single hypoxic episode on 18 October 1990, then what is the clinical documentation and records consistent with?

Documents

We ***enclose*** the following documentation.

1. Autopsy Report for Patrick Folbigg

We ask that you also have regard to any relevant documents made available to you via email from Rhanee Rego of Cardillo Gray Partners dated 7 March 2019.

Costs

We undertake to be responsible for your professional fee and look forward to receiving your medico legal report in due course.

Would you please address your tax invoice as follows:

Kathleen Folbigg
CO/ Cardillo Gray Partners
PO Box 409
Newcastle NSW 2300

Should you have any questions or wish to discuss this matter please do not hesitate to contact us on (02) 4910 0677.

Yours faithfully,
CARDILLO GRAY PARTNERS

Stuart Gray
Partner

Encl.: