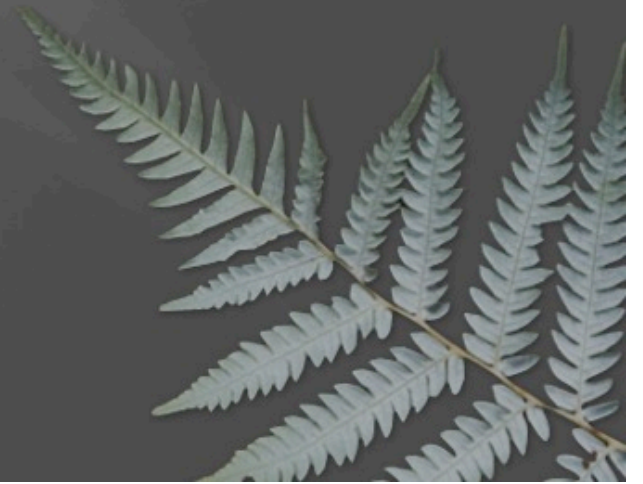


# GEORGE WILLIAMS

CHARGE Syndrome  
Lessons over 25 years



# LESSONS IN CHARGE SYNDROME

26 YEAR'S JOURNEY

PAEDIATRICIAN / PERINATOLOGY

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SYDNEY

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# biography

Born 21/4/47 Worcester RSA

3rd of 8 children (4 boys & 4 girls)

children born in 12 years

adhd cd bright but unhappy 5-12 yrs

went to school at 4 yrs

medicine at UCT at 17 yrs (1963 - 69)

internship NSH - 1970

paed training LH - 1971 to 1975

emigrated to Oz in 1975 - political activist

# Oz and Canada

- Paed registrar at CH (Camperdown) -1975 to 1980
- Perinatal fellowship at McMaster U -1980 to 1982
- Neonatologist at CWH in Sydney
- Director of Nicu at CHC till 1988
- Baby with Charge died 1982 - first experience !!!!
- Sophia C was born on 17/6/83 - met Rob Last
- My Charge journey begins
- Private practice in 1988, visited North Rocks School
- Met several families - the Baptism of Fire !!!!

# OVERVIEW

- STORIES
- LESSONS
- AFFIRM
- EMPOWER
- ADVICE
- QUOTE

# JASMIN F - 28/8/87

- My care at 9 1/2 years - 12/3/97
- BW 8 lbs, St Mag H, floppy and blue, heart murmur
- Craniostenosis, Charge diag at 6 wks Prof Sillence
- Intussusception at 8 mths - surgery
- Attending normal year 4 - no help with learning!!!
- Neurosurgery x 4, PS, PDA, PFO
- Nocturnal enuresis - p/b alarm, balance problem
- Live in Tasmania - July 94 - Dec 96
- Ear infections, grommets, kidney reflux repair

# Jasmin

- Sat 12m, crawled 18m, walked 2 yrs, talked at 2 yrs
- Talks loudly, developmental delay
- Coloboma L>R, L smaller eye, profound HL on L
- L choanal stenosis, no sccs, tandem walk difficult
- Renee, younger sister, has CHARGE
- Spont L Retinal detachment in 99, prosthesis fitted
- Psychiatrist - improved mental health state
- Parents are normal - charge free
- CHD 7 testing in Holland were negative
- 64.2 kgs, 168.4 cms, Hrt, thyroid N, Pituitary N
- Working !!!!!

# ALEXANDRIA M - 23/6/97

- Came into my care 5/9/97 - Gosford / RNSH / CHW
- E's 2nd child, 36 weeks, bw 2520, Apgars 3 & 4
- Choanal atresia, L iris & chorioretinal coloboma
- VSD, PDA, evolving CoA, misshapen ears, poor growth with vomiting, sliding hiatus hernia on Ba
- GORD - fundoplication and G and pyloromyotomy
- dysfunctional SSB, chewing, swallowing/choking
- small labiae, Ca Mg Ph, PTH and T cells N
- Recurrent chest infections

# Alexandria M

- Chronic lung disease
- SOA - nocturnal CPAP
- Hearing - N on R, severe SNL on L
- AEP on brainstem - no response on L
- CT: brain structure N, N cochleas, hypoplastic vestibule, absence of sccs, abnormal ossicles, small iam, L>R, Rob Last involved early !!!
- Good Charger - verbal, learns well, cheeky, good roller skate
- 6/08 - wt 42 kgs (70p), L 141 cms, Hc 52 cms (40p)

# Niall C - dob: 18/8/94

- My care since 17 months
- Last reviewed July 2008 - foster care
- Preg & Del N, BW 3520 g, diagnosed at birth
- PDA repair at 2 months
- Typical CHARGE face and ears
- SOA Bipap and CPAP, Fundo and gastrostomy
- Coloboma, Mod to sev HL - hearing aids, glue ear and grommets, smaller R eye - glasses for VI
- Significant developmental delay - early intervention
- Repeated lung aspiration and pneumonia/ asthma

# Niall C

- Poor growth - growth hormone replacement
- Possible seizures - EEG N
- Challenging behaviour - Ritalin & Risperdal
- Child psychiatrists and behavioral therapists
- Variety of medications
- North Rocks school
- Problems respite and parent support
- DOCS and Minister placed him care
- Mum is a lawyer and has regular contact
- LD, ADDH, R F palsy, prepubertal, 33 kgs, 140 cms

# Abigail C - dob: 24/7/02

- Tracey 3rd pregnancy & met her 5 days before birth
- Polyhydramnios, cleft lip/palate, Doppler and Amniocentesis N, Steroids
- SoL at 33 weeks, Apgar's 3 and 6, bw 2705 grams
- Clefts, Mucus +, RDS, 60 % O<sup>2</sup> - TOF+OA, low BS
- T/F to RCH in Randwick, diagnosed with CHARGE
- Tetralogy of Fallot (complex) - HF, GORD, Coloboma large, R choanal atresia and stenosis x 2
- Stricture of O, chylothorax, multiple surgeries, bleeding, BE, genetic studies N, CPAP, UTI's,

# Abigail C

- Discharged after 14 months !!! Died ?
- 10 volumes of medical notes
- 9 major and 4 minor anaesthetics
- 70 X ray procedures, 3 central lines, 30 ECG's
- 4 LBO, 5 balloon dilatations
- 3 major heart operations
- Many infections
- Respiratory arrest

# Abigail C

- Denver 11 at 25 months - personal / social skills ~ 8 mths, fine motor / adaptive skills ~ 12 mths, language 15 months & gross motor ~ 7 mths
- Last review July 08 - 17.4 kgs, 106 cms, hearing aids, kindergarten with itinerant support (H)
- Dumping syndrome, Duplex R kidney with Uti's
- Frail, easy fatigue, tachycardia, poor balance, little muscle, gastrostomy in place, toilet timed, walking and verbal but difficult to understand
- IM intellectual functioning and for IQ assessment

# Abigail C - lessons

- George missed the diagnosis
- non - acceptance of DD agency individual hearing and visual abilities
- Communication starts at birth
- Multiple sensory deficit compounds - interdependent
- CHARGE S is nasty, complex and pervasive
- Are there limits to care?

## Julia P - dob 3/3/83

- First seen Dec 1998 - 15 yrs & 9 months
- GORD on Losec, Premarin, GH from 5 to 12 years
- Choanal atresia, facial palsy, no coloboma, LD dextrocardia, VSD, PS, abnormal sccs with poor balance, winging of scapulae, short gut from NEC resection - loose stools, Wt 30 kgs L 144 cms Hc 44
- 17 hospitalisations, 15 anaesthetics, 26 operations
- Diagnosed with DiGeorge - is this true ?
- 22q11.2 negative, Calcium, Phosphate, PTH - N
- T cells normal

# Ashleigh W - dob: 22/4/01

- Enrolled in APSU study- born at 34 weeks, bw 2465
- Apgar's 4 & 8, apnoea at 20 minutes, intubated at birth - choanal atresia - after failed nasal intubation
- Grade 3 to 4 VERR - antibiotics
- PDA, R coloboma, "coloboma" behind the L eye
- Ent registrar did choanal repair day 2 - drilled deeply and difficulty- csf leak - CT confirmed bleeding, air bubbles and a tract - neuro- surgeons had to fix leak
- Developmental delay, L proptosis, hemiparesis
- Sharon Barrey-Grassick supporting

# Ashleigh W

- Suck, swallow and feeding issues
- Severe HI - hearing aids
- SOA - nocturnal cpap
- GORD - GI and fundoplication
- Asymmetry with better limb on R side -Jan V Dijk
- Left hemiparesis
- Large L retrobulbar lymphangioma (?coloboma)
- Visited to Sydney - many specialists
- I/V radiology 14/7/05 - Injected sclerosing agent
- Lawsuite - Kim Blake to be invited as expert witness

# ADVICE

- THE JOURNEY IS TOUGH
- RELENTLESS AND UNPREDICTABLE
- IS IT WORTH IT ?
- SEEK RELIABLE INFORMATION
- SURROUND YOURSELF WITH PEOPLE WHO CARE AND YOU CAN TRUST
- DEVELOPMENT A NETWORK
- KEEP GOOD RECORDS
- BE AN ADVOCATE
- DISABILITY vs LIMITS vs WORTH

# INCREASED SELF-EMPOWERMENT

## Herbert 1988

- KNOWLEDGE - child, behaviour and temperament
- SKILLS - communication, problem solve, build social relationships and enhance academic skills
- VALUES - strategic thinking, life goals, objectives, ideologies, rules, roles, relationships, emotional barriers, attributions and prejudices

# Learning and Loving in a CHARGED Life

- The adventure of a charged life is to learn
- the purpose of a charged life is to grow
- the nature of a charged life is to change
- the challenge of a charged life is to overcome

William Ward (modified)

Photos of Abbey C

Video clips of Ashleigh W