

“Medical Overview”

Nick Lennox

**Down Syndrome NSW’s Health Conference on Friday 27
September 2019**

Venue: Novotel Central Hotel, Sydney CBD

Audience: Parents, family members and carers of adults with Down syndrome



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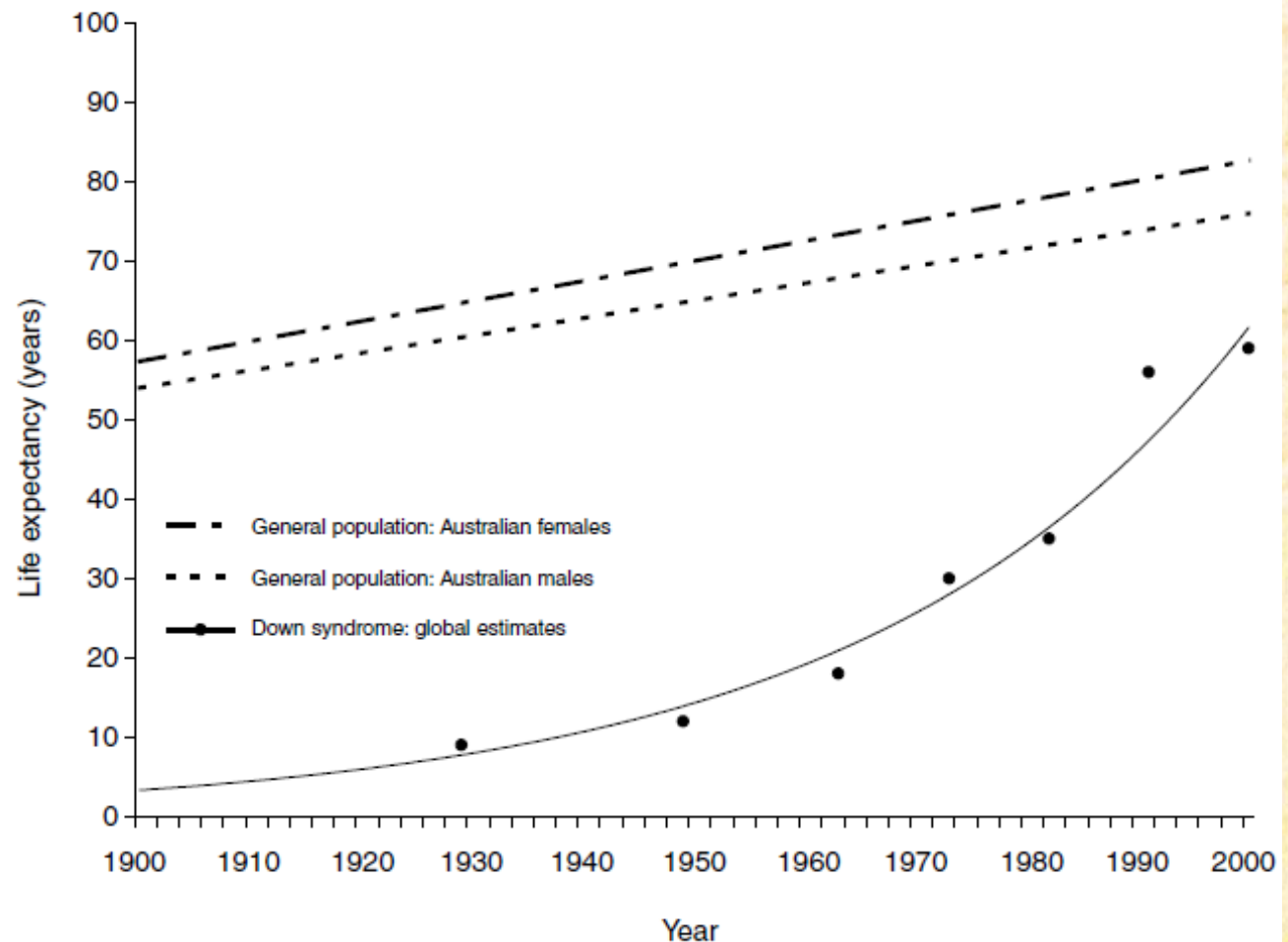
Overview

- Why does ageing lead to more health issues for people with Down syndrome?
- What are the range of different health issues that people with Down syndrome experience as they age?
- How common are age related issues in people with Down syndrome?
- Can some age-related health issues be prevented? What are the preventable health steps?
- How can we get doctors to take health concerns of people with Down syndrome seriously?



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Figure 1: Global trends in life expectancy estimates for people with Down syndrome compared with those for the general Australian population, 1900–2000. Sources: Australian data, Australian Bureau of Statistics;^{77,78} Down syndrome data drawn from various developed countries, see Table I.



In past 50 years the life expectancy for people with Down syndrome has increased by an average of 0.94 life years per calendar year



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Limitations

- Aging a relatively new phenomenon
- Clinical convenience samples
- Longitudinal cohort studies difficult to maintain



Ref: Co-occurring medical conditions in adults with Down syndrome: A systematic review toward the development of health care guidelines [George T. Capone](#)
AMJA [Volume 176, Issue 1](#) January 2018 Pages 116-133



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Why does ageing lead to more
health issues for people with
Down syndrome?



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WHY

- Underlying “cause” COMPLEX & UNCERTAIN
- Certainly biologically multiple AND plus psychosocial



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Biological

- Genetic - Trisomy 21
 - Heart, blood vessels & brain
 - Structure and function
 - E.g. Micro bleeds
 - Excess deposition of Beta Amyloid protein
 - Other potential processes
 - Inflammation, oxidative stress, mitochondria dysfunction.....
- Compounded by
 - Obesity, sleep apnoea, dementia etc.



Ref: D.M.Wilcock et al. / Biochimica et Biophysica Acta 1862 (2016) 909–914



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What are the range of different health issues that people with Down syndrome experience as they age?

AND

How common are age related issues in people with Down syndrome?



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Jane Tracy

Australians with Down syndrome

Health matters

Table 1. Implications of trisomy 21 for body systems

Organ/system affected	Resulting conditions
Heart	<ul style="list-style-type: none"> • Congenital heart disease (50% neonates) • Mitral valve prolapse (50% adults)
Gastrointestinal tract (GI)	<ul style="list-style-type: none"> • Congenital GI anomalies (12%) including: oesophageal or duodenal web/atresia, tracheoesophageal fistula, pyloric stenosis, Meckel diverticulum, imperforate anus, Hirschsprung disease (1%) • Chronic constipation: relates to hypotonia, diet, exercise • GORD (common): relates to congenital GI dysmotility, hypotonia, weight, acquired dysmotility in Alzheimer disease (AD), <i>Helicobacter pylori</i> infection • Coeliac disease (5–15%)
Neurological	<ul style="list-style-type: none"> • Intellectual disability/learning difficulties (100%), epilepsy (6% children, 80% in those with advanced AD), AD (% increases as age increases – rare under 40 years, ~50% at 60 years) • Cord compression from subluxation/dislocation of atlanto-axial joint (~2%)
Sensory impairment	<ul style="list-style-type: none"> • Hearing loss (75%): sensorineural and/or conductive (otitis media, glue ear) • Vision: refractive errors (50%), strabismus (44%), nystagmus (20%), conjunctivitis, tearing from small/blocked nasolacrimal ducts, congenital (3%) and acquired (50%) cataracts, keratoconus (adults)
Respiratory	<ul style="list-style-type: none"> • Vulnerability to respiratory tract infections including URTI, otitis media, pneumonia (12-fold risk related to immunodeficiency, aspiration, hypotonia) • Sleep apnoea (50–75%): relates to hypotonia, anatomy of mid-face, weight
Musculoskeletal	<ul style="list-style-type: none"> • Characteristic physical features: small stature, single palmer crease (20%), brachycephaly, microcephaly • Hypotonia: contributes to feeding difficulties, sleep apnoea, hyperflexibility, joint subluxation/dislocation, eg. hips (6%), patella • Hypoplasia of midface: contributes to otitis media, mouth breathing • Atlanto-axial instability (14%, symptoms/signs in ~2%)



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Health matters

Immune system	<ul style="list-style-type: none">• Recurrent respiratory tract infections including pneumonia• Folliculitis, blepharitis, conjunctivitis, periodontal disease, aphthous ulcers• Autoimmune disease: alopecia, hypothyroidism, diabetes, coeliac disease
Dermatological	<ul style="list-style-type: none">• Dry skin, infections, eg. folliculitis, atopic dermatitis, seborrhoea, blepharitis, psoriasis, alopecia (~8%)
Endocrine	<ul style="list-style-type: none">• Hypothyroidism (3–54%, increases with age)• Failure to thrive, obesity, diabetes• Women: early menopause (~44 years), decreased fertility• Men: hypogonadism, undescended testes, decreased fertility• Osteoporosis: increased risk related to diet, anticonvulsants, sun exposure, weight bearing exercise, hypogonadism, early menopause
Genitourinary	<ul style="list-style-type: none">• Congenital renal tract anomalies, hypospadias, cryptorchidism
Haematological	<ul style="list-style-type: none">• Functional defects in white cells related to decreased immunity, macrocytosis, transient leukaemia or thrombocytopenia in neonates. Leukaemia is more common in children/young adults with DS (~1%)
Dental	<ul style="list-style-type: none">• Delayed and abnormal dentition (eg. partial adontia 50%, microdontia 40%), periodontal disease, malocclusion, difficulties with oral hygiene
Mental health	<ul style="list-style-type: none">• Bio-psycho-social risk factors: increased risk of anxiety, depression, obsessive compulsive disorder, AD (rare under 45 years, ~50% by 60 years)

Jane Tracy Australians with Down Syndrome
<https://www.racgp.org.au/afp/2011/april/australians-with-down-syndrome/>



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Health Issues - Adults

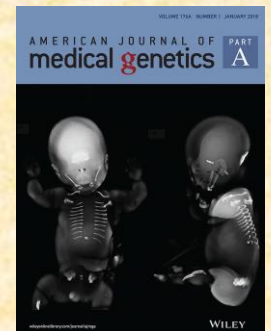


- Age-related dementia
 - rare under 40 but ~ 50% over 60 years
- Hearing impairment 73% (up to 100% age 60)
- Visual impairment (~50%) & disease
 - Ages - Cataracts (~50%) & Keratoconus
- Behaviour- Mental health – OCD, Depression & Anxiety Disorder



Health Issues - Adults

- Obesity & overweight
 - 60-70%
 - Obesity (48% vs. 41%) Women Vs Men
- Sleep Apnoea (85%)
- Osteopenia-osteoporosis
 - > general population & others with ID
 - Male = Female
 - ? Increase fractures but probably



George T. Capone AMJA Volume176, Issue1
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Other Health issues

- Pulmonary disease -12x > risk pneumonia
- Thyroid disease (c.f. 3%) - increases age 27%
- Degenerative change - cervical spine 33–64%
- Atlanto-Axial Instability 2-20% (Children 15-20%)
- Dental & gastrointestinal problems
- A long list

<https://www.racgp.org.au/afp/2011/april/australians-with-down-syndrome/>

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Can some age-related health issues be prevented? What are the preventable health steps?



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Prevented

- Challenging
- Largely screening & identification
- Preventable
 - Depression and anxiety
 - Obesity & sleep apnoea
 - Dementia – a lot of interest

If I was you question – Maximise connection, manage weight & exercise



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Screening

- Eyes & ears
- Thyroid screen
- Sleep apnoea
- Osteoporosis
- Congenital heart disease – echocardiographs
- Lipids & diabetes testing
- Annual Health assessment



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Summary

- Connection & a good active life
- Weight control - diet management & exercise
- Osteoporosis prevention
- Screening
 - Thyroid, Sleep apnoea
 - Vision and hearing testing & screening
 - Echocardiogram



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Summary

- ✓ Connection & a good active life
- ✓ Weight control - diet management & exercise
- ✓ Osteoporosis prevention
- ✓ Active screening



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How can we get doctors to take
health concerns of people with
Down syndrome seriously?



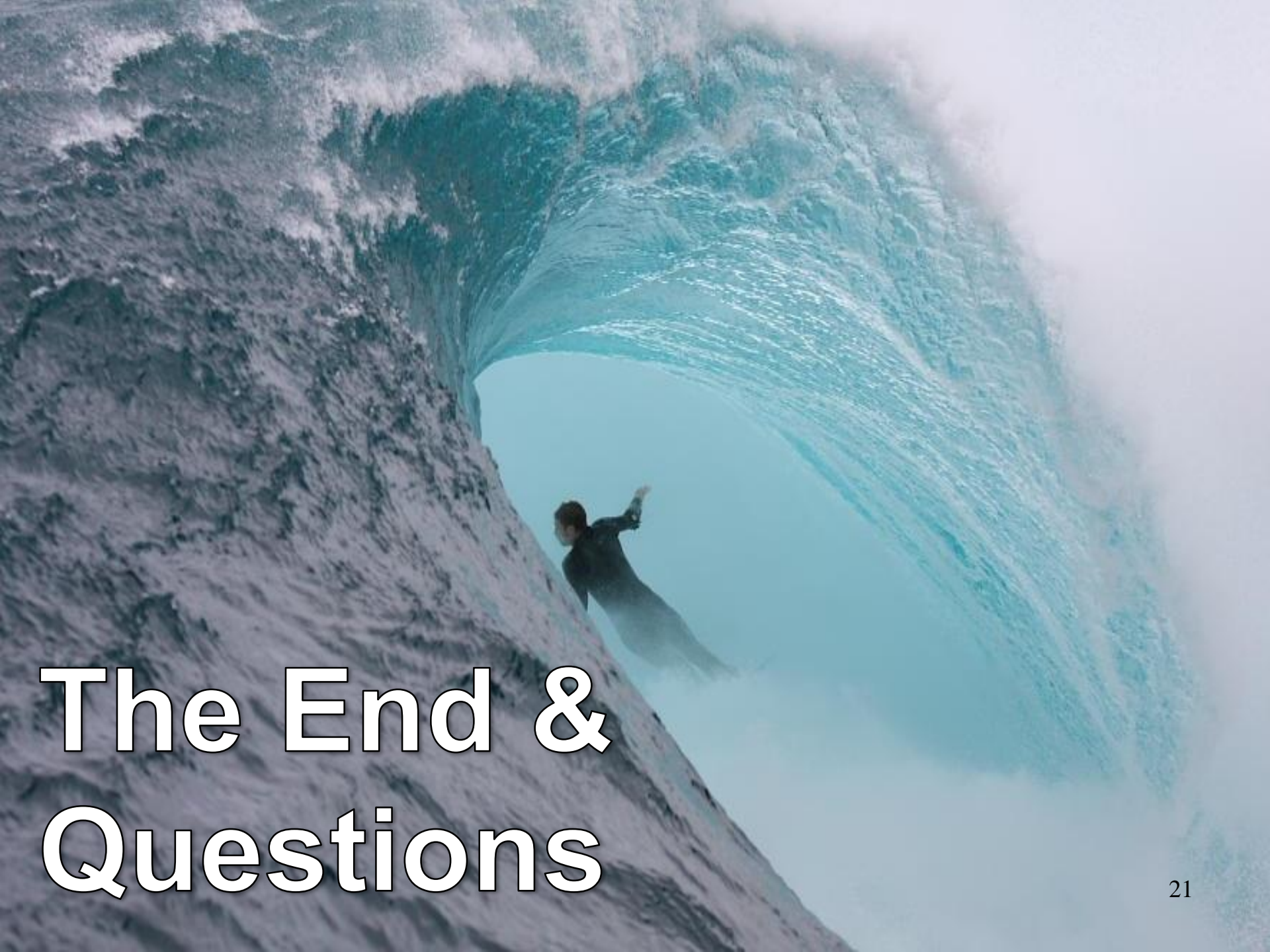
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Doctors

- Respectful advocacy
 - “You never really understand a person until you stand in their shoes” Atticus Finch
- Continuity of care
- Know your child’s risks and advocate
- Provide information?
- Annual health checks
 - CHAP plus AFP article (Conflict of Interest)



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The End & Questions