



UNIVERSITY OF
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Self-injurious behaviour in genetic disorders and autism spectrum disorder

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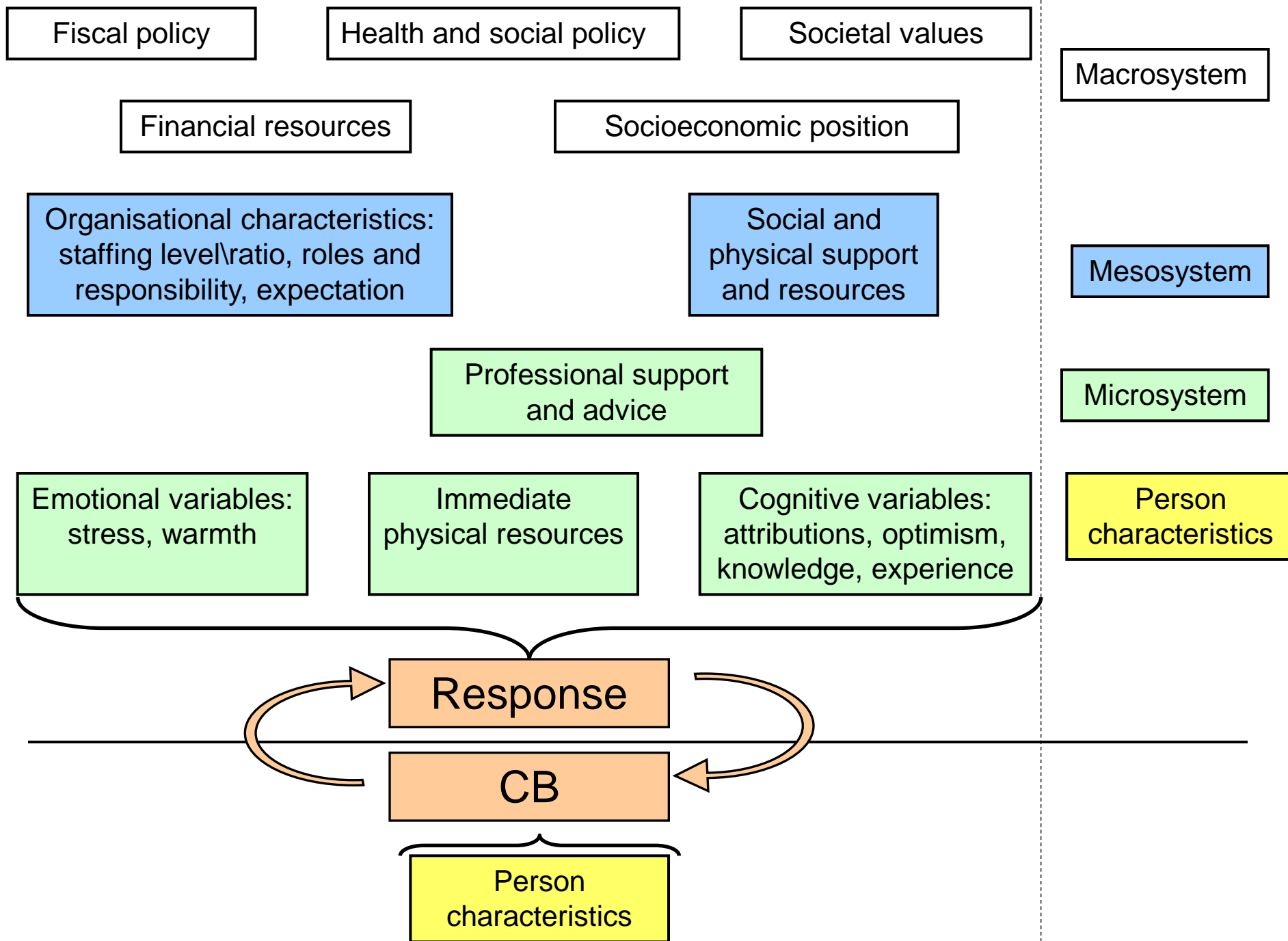
Self-injurious Behaviour and Severe Intellectual Disability

- Prevalence: 10% - 35%
- Can be chronic and resistant to intervention (c. 80% persistence over 18 years)
- Human and economic costs: pain and discomfort, family and carer stress, relationship breakdown, medication effects, compromised quality of life and accomplishment, placement breakdown.
- Economic costs are likely to be significantly underestimated
- Lack of appropriate 'clinical' intervention. (SIB, 1987, psychological 2%, medication 40%)

Environmental influences on self-injurious behaviour (shedding more heat than light?)

“There is strong evidence that many behavior disorders, including SIB, are learned. As such, these behaviours are acquired through an individual’s history of interaction with the environment.....”

Brian Iwata



A robust and sustained operant learning theory literature

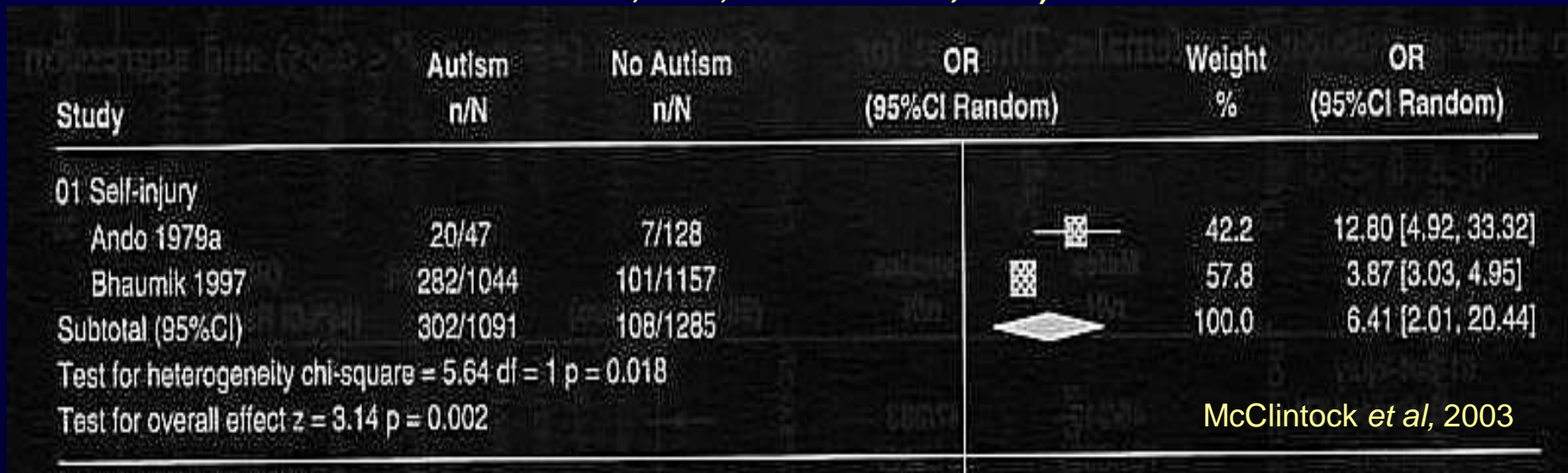
- c. 6,000 to 7,000 published papers
- The rise of functional analysis
- Translation into Positive Behavioural Support (e.g. FCT)
- Emerging evidence from RCT's
- But.....
 - is it a complete account?

Does diagnosis and/or cause of
intellectual disability matter?

Prevalence of self-injurious behaviour in ASD = 40 – 50%

OR = 6.41

(Baghdadli, Pascal, Grisi & Aussilloux, 2003; Billstedt, Gillberg & Gillberg, 2005; Murphy, Healy & Leader, 2009; Shattuck *et al.*, 2007)



How common and persistent is self-injury in autism spectrum disorder?

1 in 2

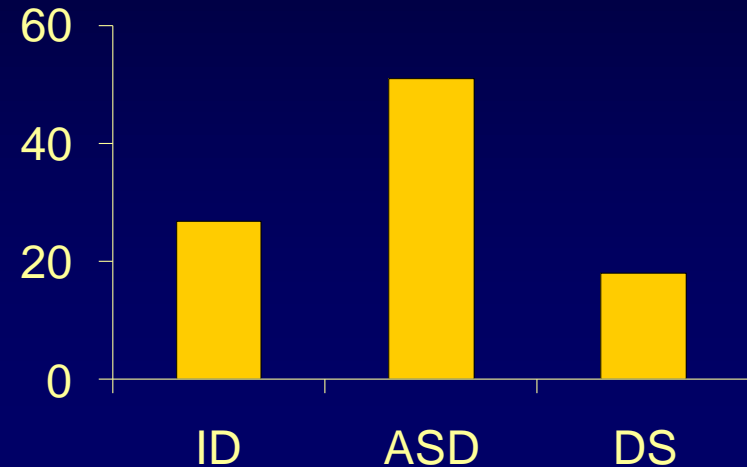
- Two surveys
- Across causes of ID

80%

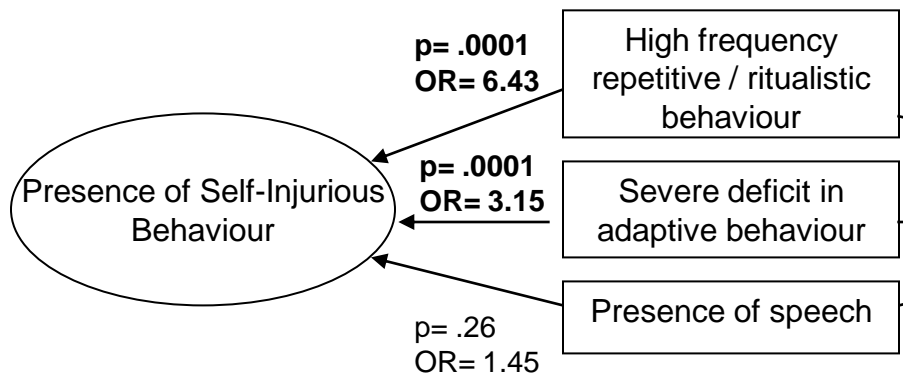
- Persistence over 3 years

Risk

- Degree of ID
- Behavioural markers



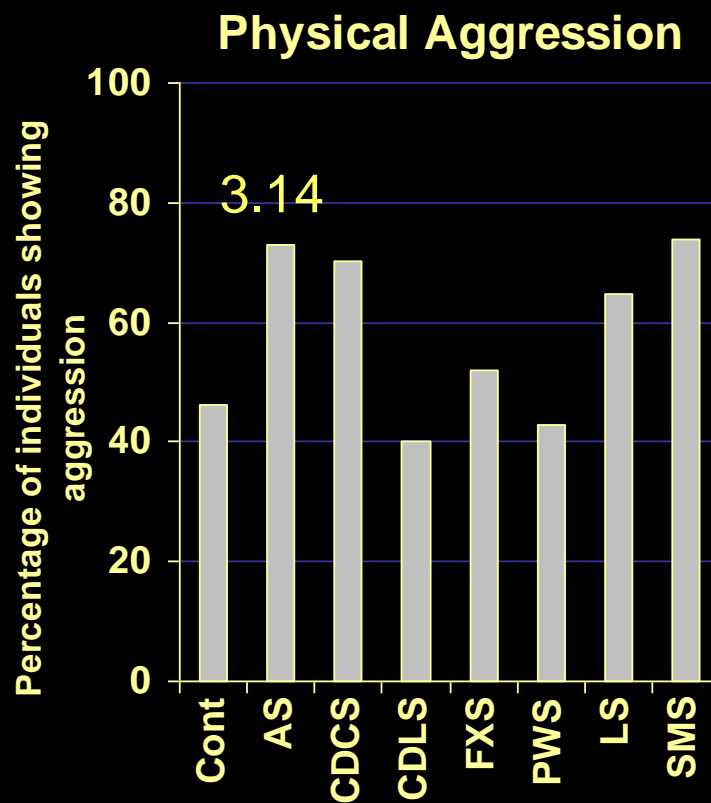
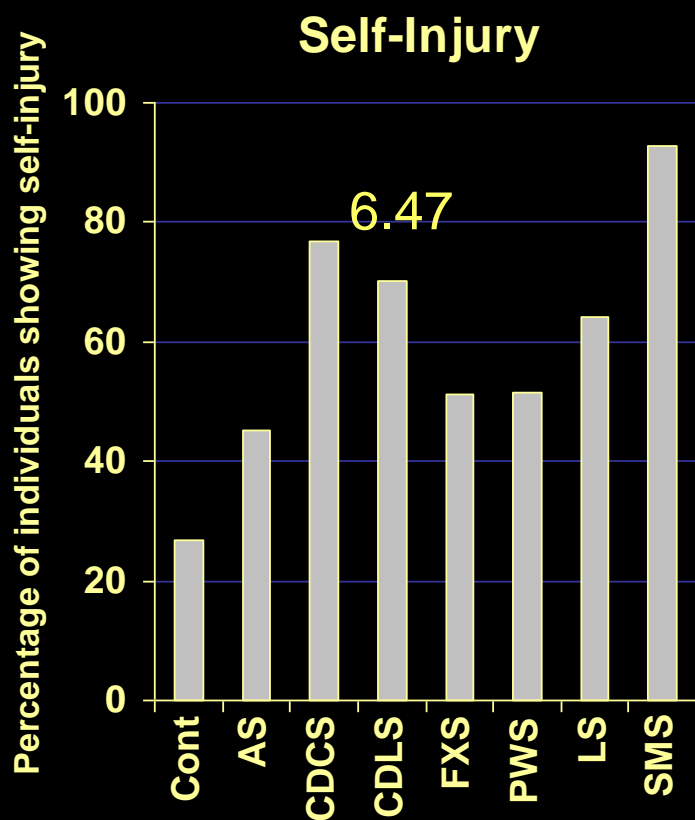
Presence



c. 1,000 children with SLD

One behaviour strongly associated with ASD is an important predictor

Does the prevalence of self-injury and aggression vary across genetic syndromes?



Important individual characteristics

1. Physical health

Cornelia de Lange syndrome



- Prevalence: 1 in 10,000 to 40,000
- Deletions on chromosomes 5, 10 and X
- Main features: mild/moderate to severe ID, small stature, upper limb abnormalities, distinctive facial features, gastroesophageal reflux, limited speech, hirsute, SIB.

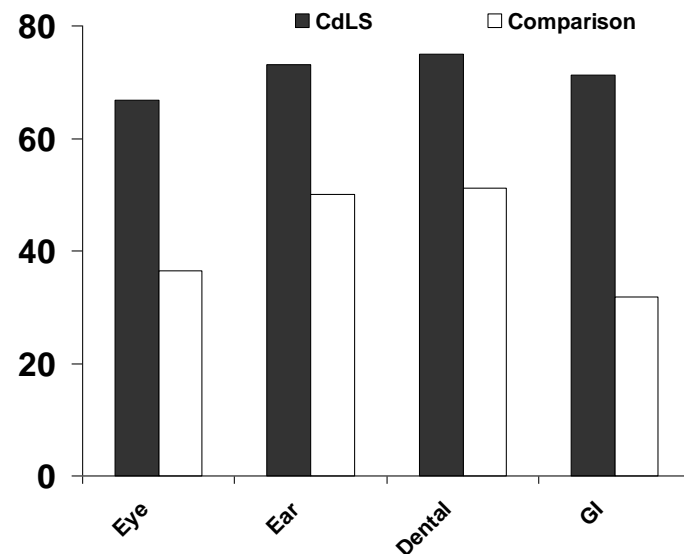
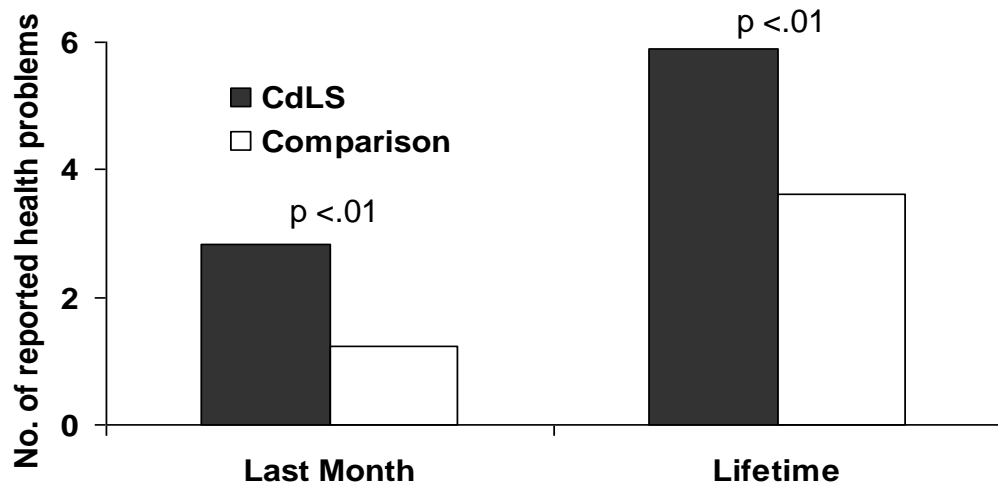


de Lange, C. (1933). Sur un Type nouvea de degeneration (typus amstelodamensis) Arch.med. enf. 36. 713 - 719.

Brachmann,W. (1916). Ein Fall von symmetrischer Monodaktylie durch Ulnadefekt, mit symmetrischer Flughautbildung in den Ellenbeugen, sowie anderen Abnormalitäten (Zwerghaftigkeit, Halsrippen, Behaarung). Jb. Kinderheilk., 84, 225-35.

Table 1 Percentage of individuals with Cornelia de Lange Syndrome (CdLS) showing specific health problems in published studies

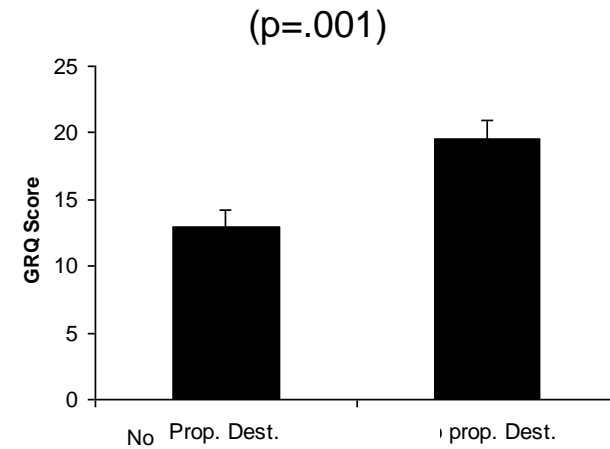
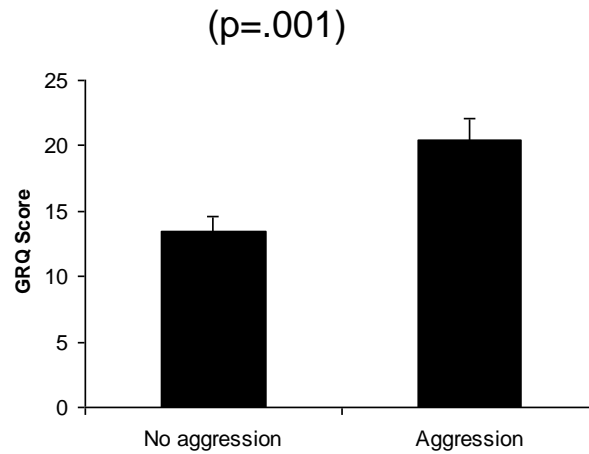
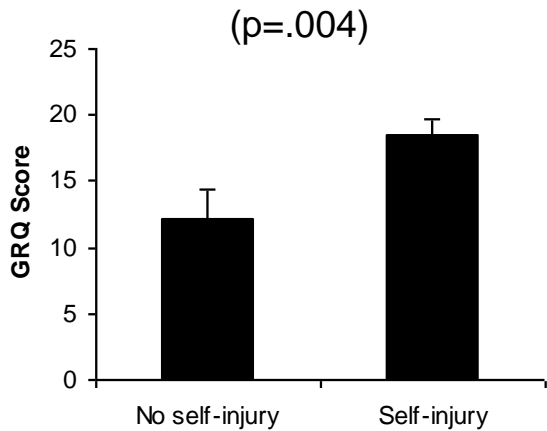
CdLS Studies	n	% of individuals experiencing health problems											
		Limb abnormalities	Gastrointestinal	Heart	Dental	Genitalia	Genito-urinary	Eye	Ear	Respiratory	Epilepsy	Skin	Cleft palate
Hawley et al. 1985	64	33	71	28	93	94 (males)	-	-	-	-	14	-	-
Gualtieri 1990	138	-	41	15	-	-	8	-	-	18	25	-	-
Sataloff et al. 1990	45	-	-	-	-	-	-	-	-	-	-	-	59
Ireland et al. 1993	20	80	10	15	-	-	-	-	-	-	5	-	10
Jackson et al. 1993	310	-	48	25	-	73 (males)	12	50	60	25	23	16	-
Sommer, 1993	17	-	76	-	-	-	-	-	-	-	-	-	-
Kousseff et al. 1994	37	56	49	14	-	-	33	38	-	-	38	-	21
Berney et al. 1999	49	-	67	-	-	-	-	-	-	-	29	-	-
Tsukahara et al. 1998	50	-	-	26	-	-	-	-	-	-	-	-	-
Luzzani et al. 2003	43	-	65	-	-	-	-	-	-	-	-	-	-



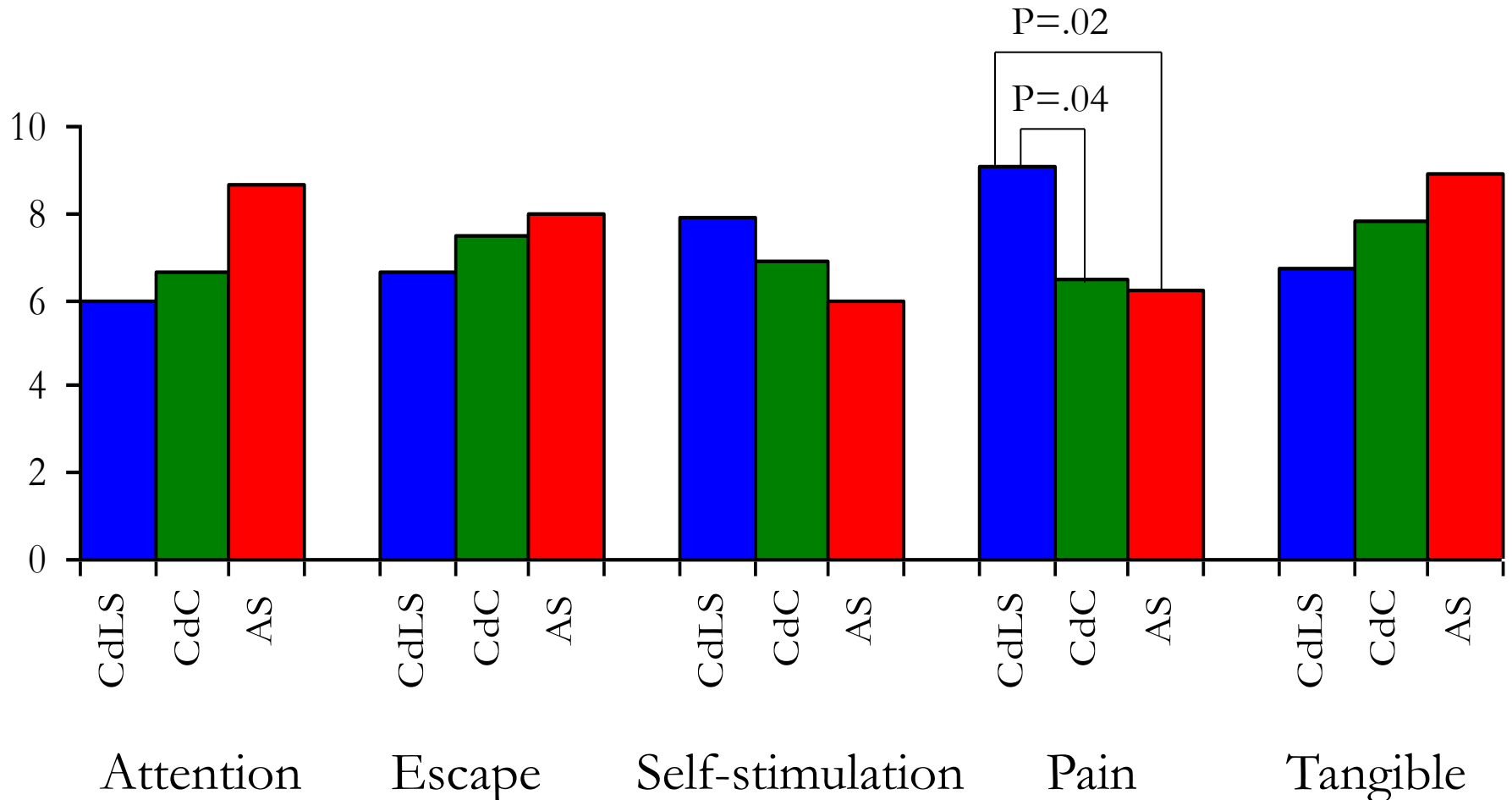
Hall, S., Arron, K., Sloneem, J. and Oliver, C. (2008). Health and sleep problems in Cornelia de Lange Syndrome: A case control study. *Journal of Intellectual Disability Research*, **52**, 458-468.

Arch his/her back
 Lie over object on stomach
 Salivate excessively
 Fidget/wriggle
 Fingers in mouth
 Chew clothes
 Grind teeth
 Scratch/rub chest/throat
 Drink excessively

Cough/gag/regurgitate
 Discomfort
 Refuse food
 Indecisive about food
 Wake during the night
 Sleep sitting or propped up
 Bad Breath
 Respiratory tract infections



Comparison of causes of challenging behaviour in Cornelia de Lange, Angelman and Cri du Chat syndromes



Important individual characteristics

2. Accentuated motivation

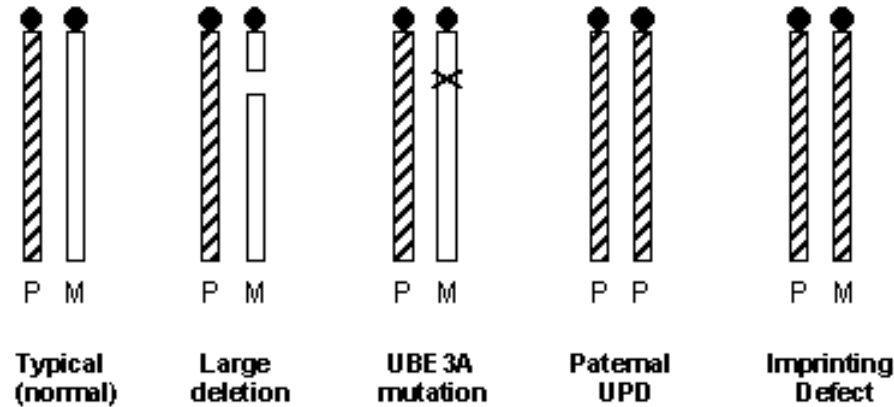
Genetic Mechanisms Leading to AS

Prevalence 1:10,000

AS/PW region: 15q11 – q13

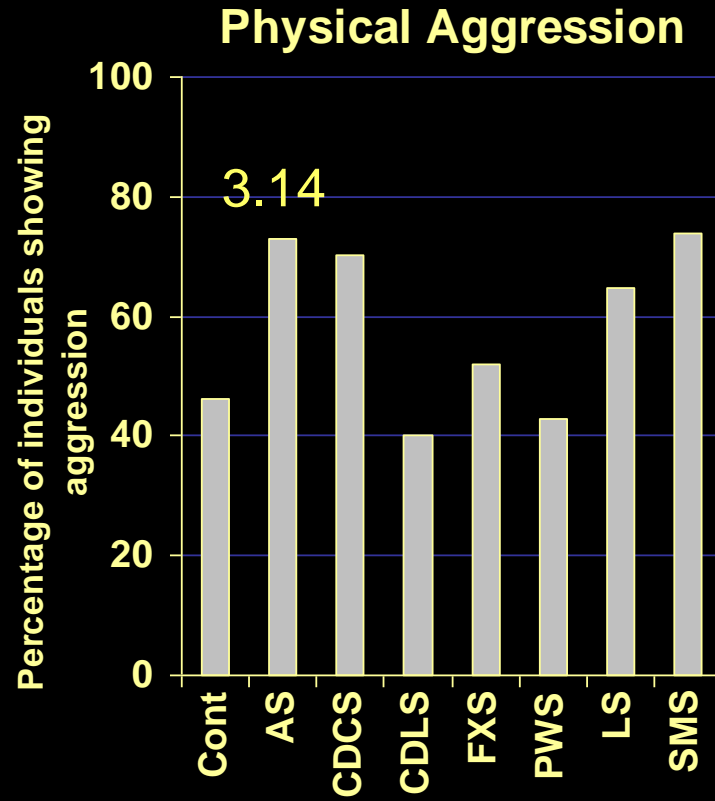
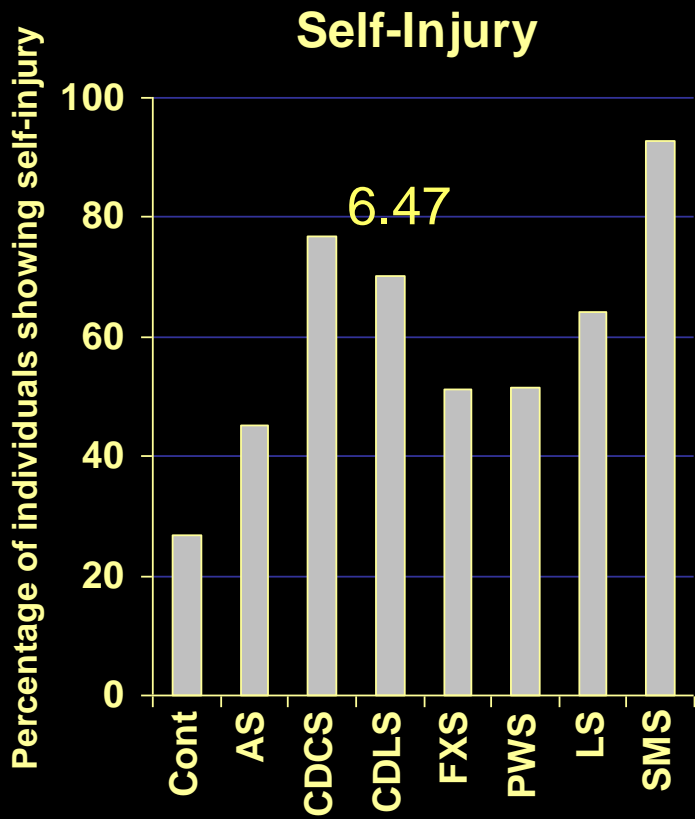
AS from loss of expression of region on maternal chromosome; PW from loss of expression of region on paternal chromosome

Phenotypic difference indicates different expression of normally functioning 15q11-q13 (genomic imprinting)



Angelman: severe intellectual disability, minimal or absent speech, hyperactivity, sleep disturbance, gait ataxia, jerky movements, **heightened sociability**, “happy disposition with frequent inappropriate outbursts of laughter”.







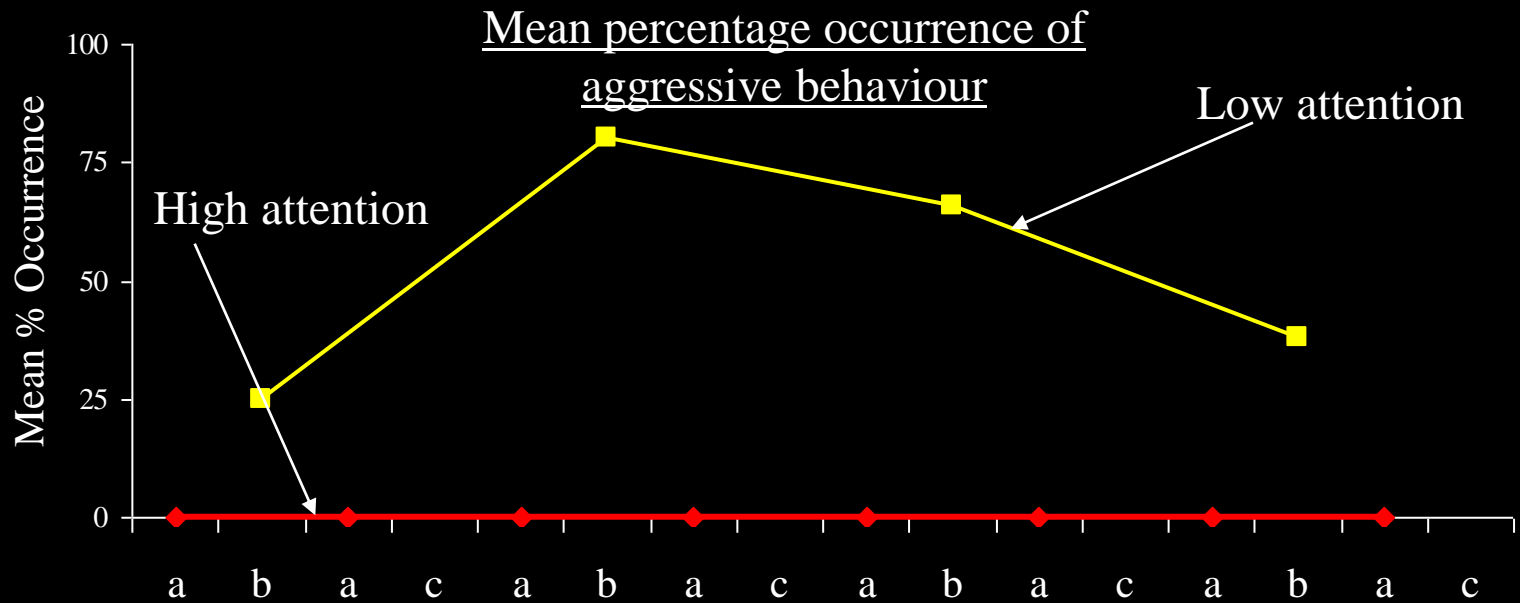


Condition A: High attention

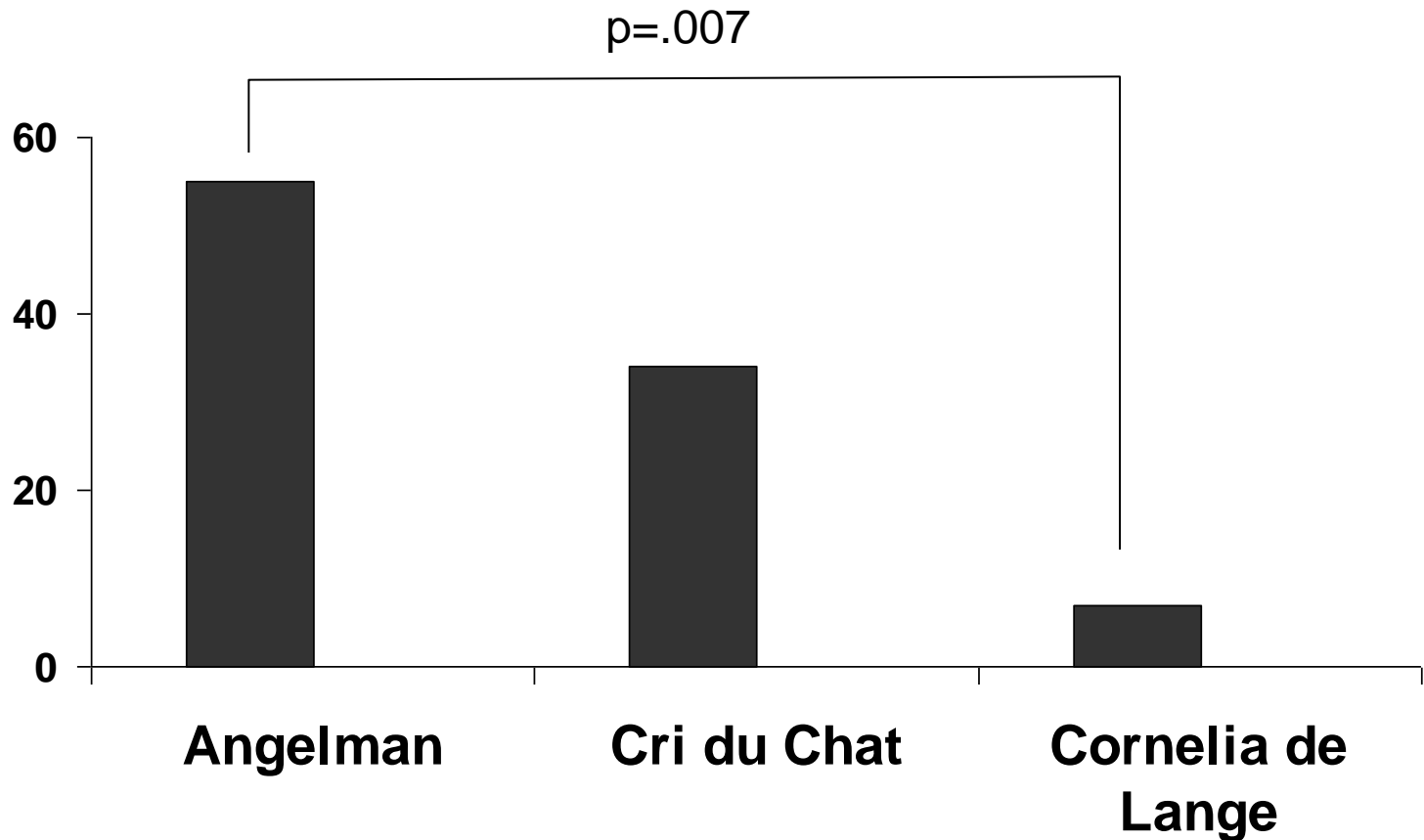
Condition B: Low attention



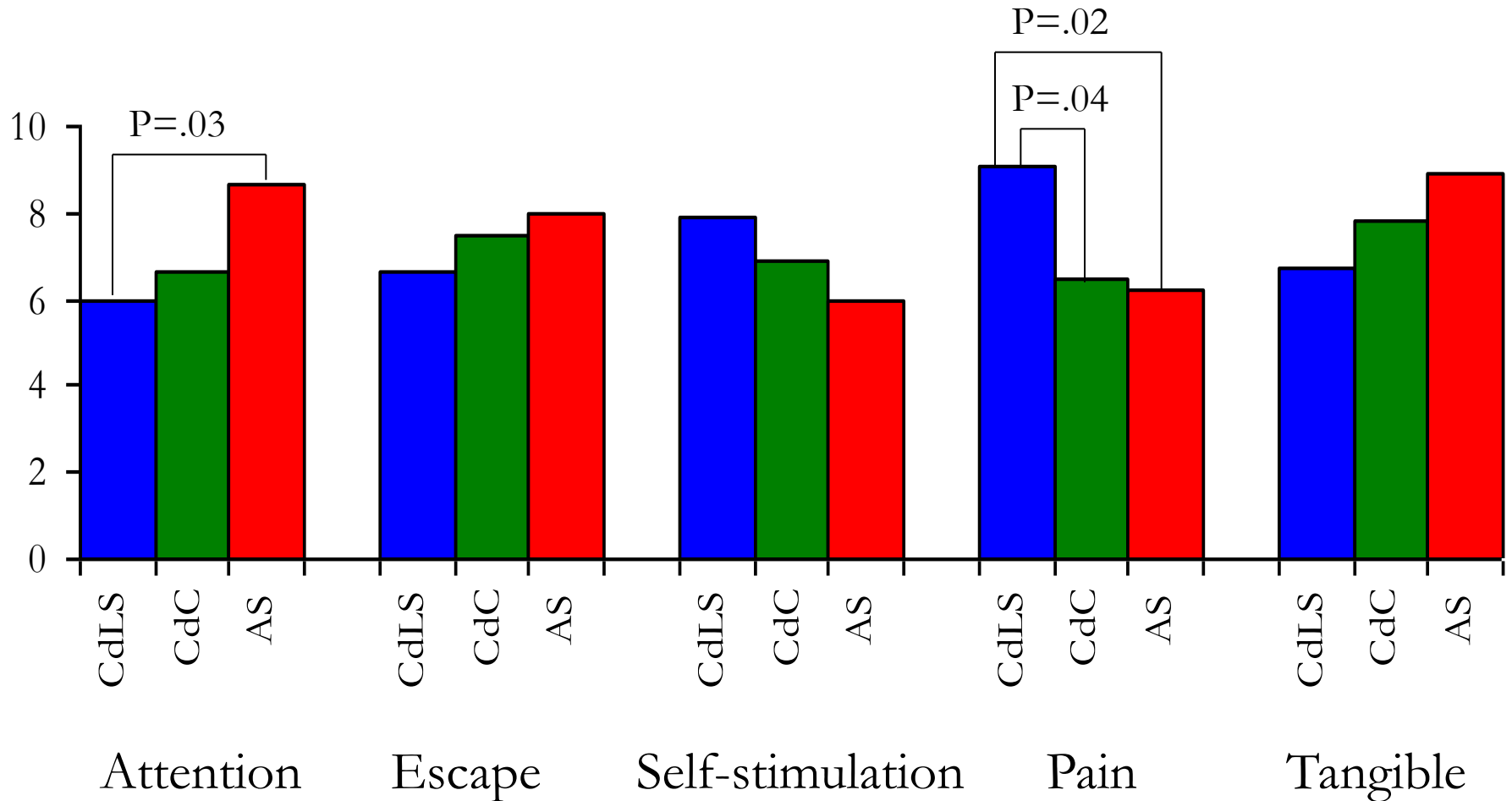
Comparison



Percentage of children showing aggression in 'low attention' conditions



Comparison of causes of challenging behaviour in Cornelia de Lange, Angelman and Cri du Chat syndromes

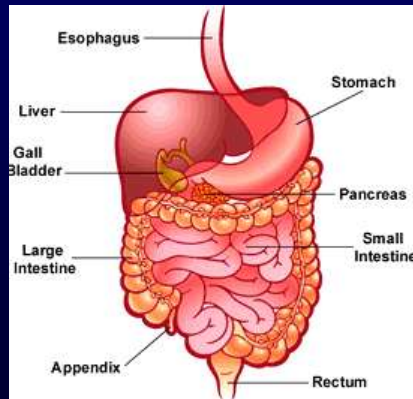


.... but is this relevant to ASD?

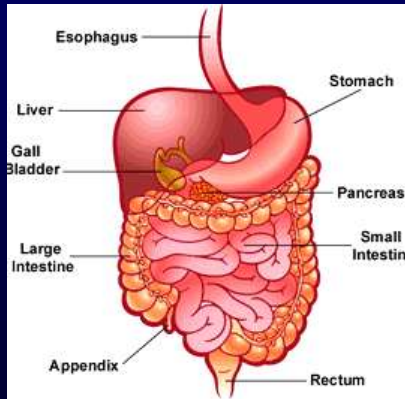
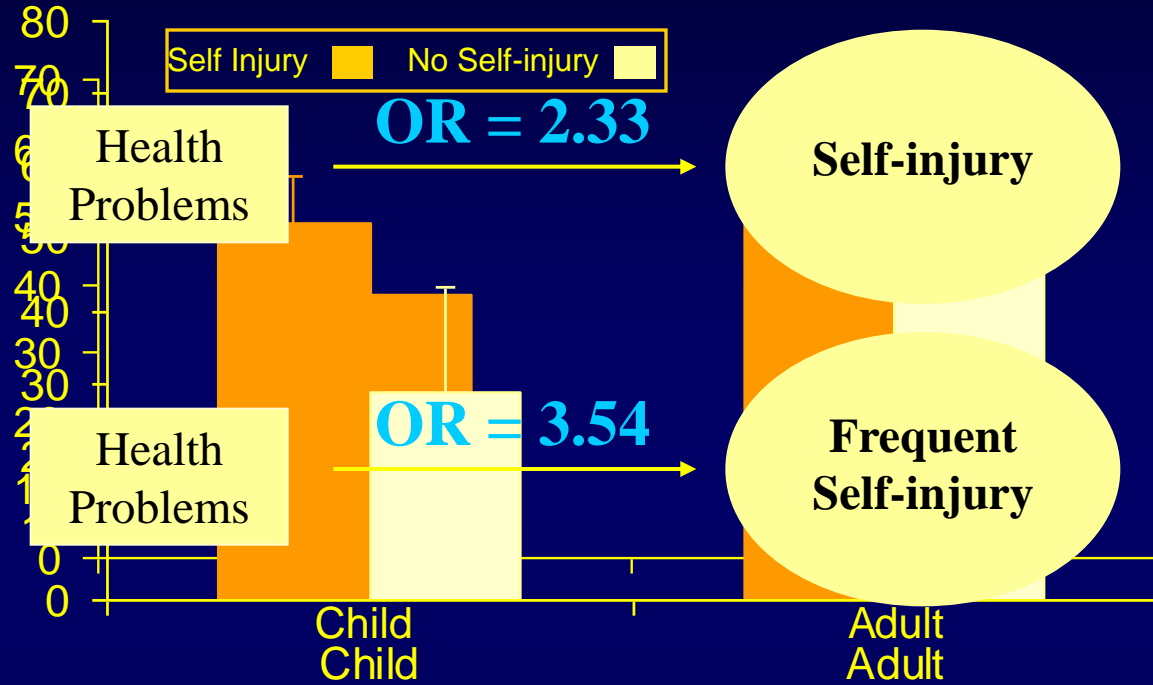
Caroline Richards

PhD Student funded by Research Autism

Painful Health Conditions?



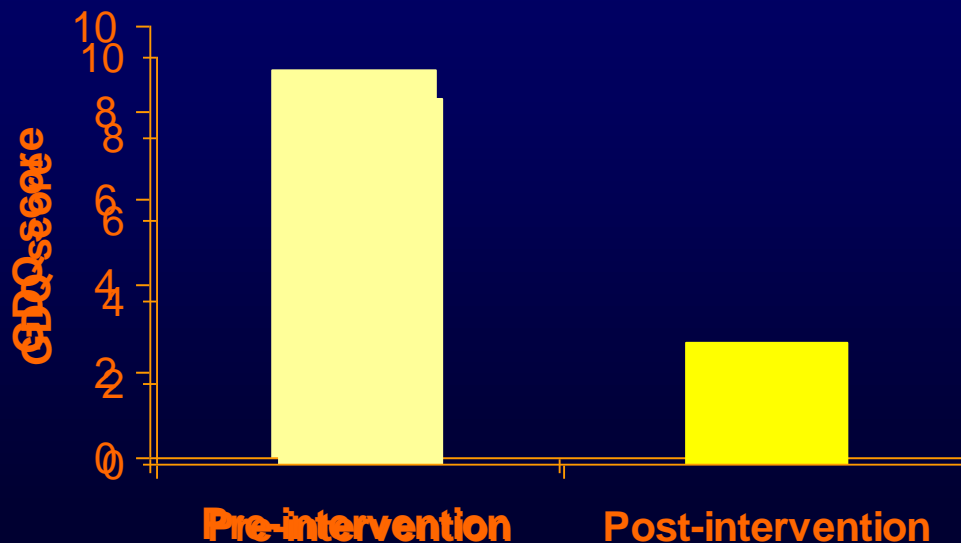
Painful Health Conditions





Case Study

- Thomas – 9 years old
- Parent's report of self-injury
 - hitting his body, head banging hitting himself with hard objects, biting his hand, biting hard objects, scratching his face
- Observations
 - Severity; stopping observations; out of nowhere
- Intervention



10 children with ASD and reported SIB

'Standard' experimental functional analyses

No Function

10 participants

Function

0 participants

ASD 'Modified' experimental functional analyses

No Function

4 participants

Function

6 participants

Other

1 participants

Sensory Escape

2 participants

Access to Repetitive Behaviour

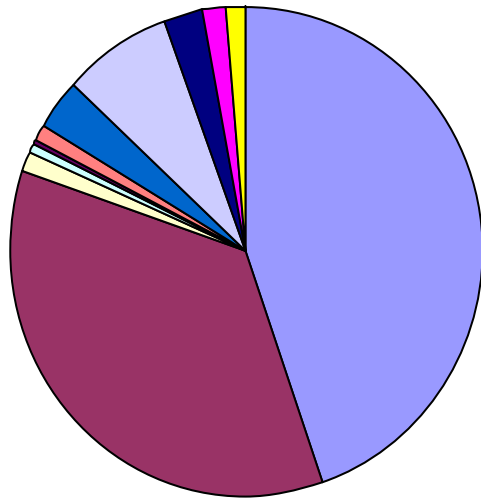
3 participants

6 in 10

• Function for self-injury

Some closing thoughts

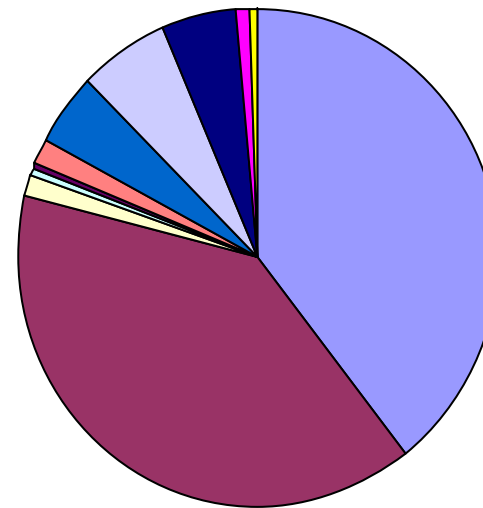
- Self-injury in ASD: a common, persistent and disabling problem
- a common, persistent and disabling problem that is treatable:
 - Pain and discomfort: overlooked and underestimated
 - Take a walk in my shoes: interactions between person characteristics and operant learning
- Research and clinical delivery: some take home figures.....



2001 - 2005

5,271 publications

(Self-injury 1.25%)



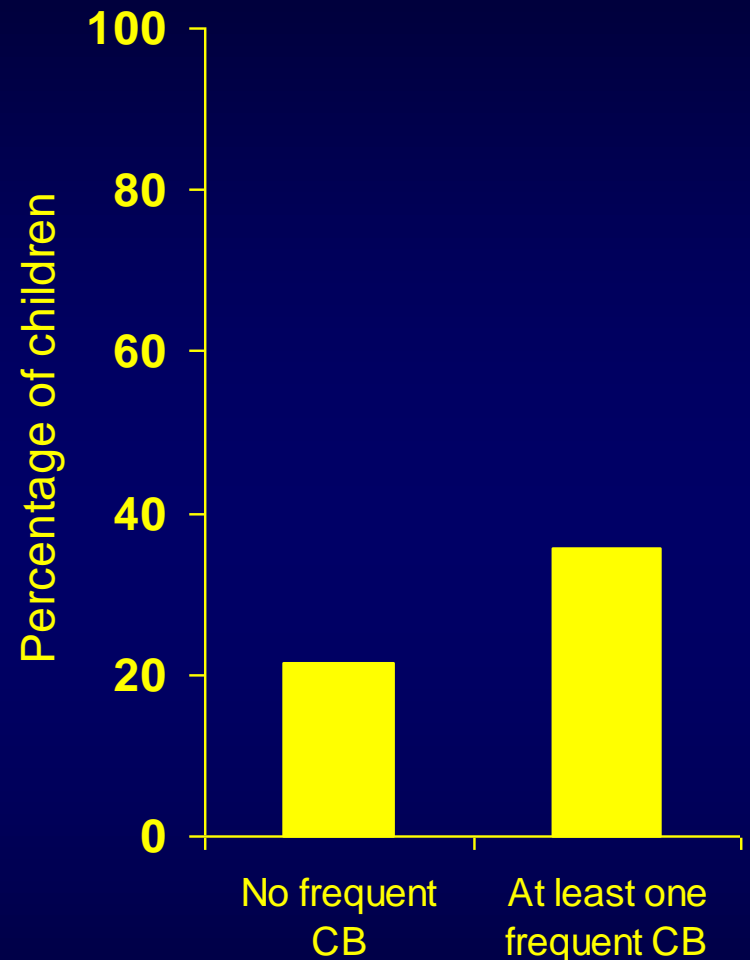
2006 - to date

13,222 publications

(Self-injury 0.086%)

- Causes
- Brain abnormality
- Prevalence
- Diet
- Sleep
- Diagnosis
- Mental health
- Cognitive function
- Interventions
- Self-injurious behaviour
- Challenging behaviour

Of those children showing frequent and severe challenging behaviour, what percentage have had at least one contact with a relevant professional in the last month?



God grant me the serenity to accept
the things I cannot change, courage to
change the things I can and the
wisdom to know the difference.

(Reinhold Niebuhr)



Core Funding

Cerebra

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Birmingham Children's Hospital
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ESRC

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Debbie Allen, Jane Appleby, Ian Apperly, Sarah Beaumont, Sarah Beck, Lisa Collis, Fay Cook, Louise Davies, Kate Eden, Ruth Fishwick, Christina Goredema, Sarah Gorniak, Glyn Humphreys, Abby Marr, Jonathan Martin, Anna Mitchell, Chris Oliver, Jan Oyebode, Jane Petty, Laurie Powis, Barzan Rahman, Donna Reid, Caroline Richards, Kristina Stockdale-Juhlberg, Penny Tunnicliffe, Lucy Wilde, Kate Woodcock.

Gastro-intestinal distress and SIB in ASD

Arch his/her back
Lie over object on stomach
Salivate excessively
Fidget/wriggle
Fingers in mouth
Chew clothes
Grind teeth
Scratch/rub chest/throat
Drink excessively
Cough/gag/regurgitate
Discomfort
Refuse food
Indecisive about food
Wake during the night
Sleep sitting or propped up
Bad Breath
Respiratory tract infections

