Our starting point

- In many, many ways children and adults with CdLS are just the same as everyone else and have the same needs, wants and rights as everyone else.
- Everyone is unique and each child and adult with CdLS is unique.
- In some ways children and adults with CdLS differ from people who do not have CdLS.
- We shall look at these differences but we will not forget the similarities or that everyone is unique.
• Prevalence: 1 in 40,000. (thought to be higher)
• 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.

Remember. CdLS is a syndrome.
So there are a number of individual problems that need to be solved

syn·drome (sĭn'drōm')

1. A group of symptoms that collectively indicate or characterise a disease, psychological disorder, or other abnormal condition.
The key issues

- Physical difference and disorder (health problems)
- Self-injury
- Poor expressive communication
- Autistic spectrum like behaviours
- Specific cognitive impairments
- Changes with age
The ear canal is very narrow and the nerves in the ear may not form correctly. This effects hearing and, in turn, speech.
Table 1: Percentage of individuals with Cornelia de Lange Syndrome (CdLS) showing specific health problems in published studies

<table>
<thead>
<tr>
<th>CdLS Studies</th>
<th>n</th>
<th>Lamb abnormalities</th>
<th>Gastrointestinal</th>
<th>Heart</th>
<th>Dental</th>
<th>Genitalia</th>
<th>Genito-urinary</th>
<th>Eye</th>
<th>Ear</th>
<th>Respiratory</th>
<th>Epilepsy</th>
<th>Skin</th>
<th>Gastrointestinal</th>
<th>Heart</th>
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<th>Genitalia</th>
<th>Genito-urinary</th>
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<th>Respiratory</th>
<th>Epilepsy</th>
<th>Skin</th>
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<td>Howley et al. 1985</td>
<td>64</td>
<td>33</td>
<td>71</td>
<td>38</td>
<td>93</td>
<td>94 (male)</td>
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<td>Gaudette 1990</td>
<td>138</td>
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The key issues

• Physical difference and disorder (health problems)
• Self-injury
• Poor expressive communication
• Autistic spectrum like behaviours
• Specific cognitive impairments
• Changes with age

C.Oliver@Bham.ac.uk
Understanding and Changing Challenging Behaviour in Cornelia de Lange Syndrome

Written by: Chris Oliver, Jo Moss, Penny Tunnichiffe, Jane Petty, Gemma Griffith, Richard Hastings, Pat Rowlin, Leah Bult, Dolores Villa and Michael Vip

A Brief Introduction

Chris Oliver will find information that will help you understand and cope with challenging behaviour in children and adults who have Cornelia de Lange Syndrome. The information is based on the latest research and best practice guidelines. The learning disabilities of children and adults with Cornelia de Lange Syndrome will be discussed in detail. This will help you to understand their experiences and to gain insights into their thoughts and feelings. However, there are situations where you may need to do more to cope with challenging behaviour. The information provided in this book will help you to develop strategies to help you cope with these difficult situations.
Prevalence of Self-Injury and Physical Aggression in Syndromes

15% of all children bang their head between the ages of 1 and 4. 50% of these children have a middle ear infection.
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

(p=.004) (p=.001) (p=.001)
Pain gate theory

The experience of pain is influenced by many factors and there is no simple pathway from sensation to experience. Three types of nerve fibres are important.
Chronic and sharp pain travel along the C fibres and A-delta fibres respectively. Messages from the fibres to the brain can be blocked by stimulating A-beta fibres. (Rub it better!)

Pain gate theory and learning to self-injure
Disordered pain perception
Electrode stimulates the nerve with a mild electrical impulse. Stimulus duration 0.1ms, frequency around 1Hz. Latency is time from the stimulus to the first positive peak of sensory nerve action potential (SNAP). Median (arm), medial plantar and sural (leg) nerves. Stable velocity at about 5 years old; median nerve mean = 67.5 m/s (SD 4.4)


Physical disorder
Gate closes
PSN decreases efficacy
Self-injury increases to compensate

Chronic Pain
Transient acute pain from self-injury

Pain gate theory and learning to self-injure
The pain cycle and self-injury

1. Chronic or sharp pain is caused by a medical condition or trauma
2. The child blocks the pain by rubbing, scratching or hitting.
3. The fibres carrying the blocking signal work very slowly so the child rubs, scratches or hits more and harder.
4. The hard rubbing, scratching and hitting leads to tissue damage and chronic or sharp pain.
5. Go to 1.

Development and Learning

• Once a behaviour has occurred a number of times it can acquire a function
• The function can be to gain pleasant sensory stimulation (spinning) or to stop unpleasant sensory stimulation (scratch-itch)
• The function can become social or communicative. The behaviour can function to ‘say’:
  – “Come here and pay attention to me”
  – “Give me…..! I want…!”
  – “Stop! No! I don’t like that!”
• When behaviour has a social or communicative function it can get worse over time
The key issues

- Physical difference and disorder (health problems)
- Self-injury
- Poor expressive communication
- Autistic spectrum like behaviours
- Specific cognitive impairments
- Changes with age
The key issues

- Physical difference and disorder (health problems)
- Self-injury
- Poor expressive communication
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- Changes with age

**Percentage of group scoring above cut-offs for Autism and Autism Spectrum Disorder on ASQ**

- Samples range from 26 to c. 250 (* < 50); Age range 4 to 54
Comparative studies show that the heightened prevalence in CdLS is not solely accounted for by degree of intellectual disability.

Table 3  Percentage of participants in each category of autism (as defined by the Childhood Autism Rating Scale) broken down by group

<table>
<thead>
<tr>
<th>Group</th>
<th>Non-autistic</th>
<th>Mild to moderate autism</th>
<th>Severe autism</th>
<th>$\chi^2$</th>
<th>$p$</th>
</tr>
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<tbody>
<tr>
<td>Cornelia de Lange syndrome group</td>
<td>32.8</td>
<td>15.1</td>
<td>32.1</td>
<td>8.77</td>
<td>0.012</td>
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<tr>
<td>Comparison group</td>
<td>71.4</td>
<td>21.4</td>
<td>7.1</td>
<td></td>
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</table>
Presentation of ASD characteristics in CdLS
Social anxiety in CdLS

- High levels of anxiety associated with social situations, particularly with unfamiliar people and unfamiliar situations.

- Motivation for social contact appears to be intact.

- Strong preference to observe rather than participate.

- Increased withdrawal when social demands become heightened.

Moss et al., 2008. AJMR 113, 278-291; Collis et al., In preparation; Reid, Nelson, Moss & Oliver, In preparation

"He can hold conversations but he is quieter when it comes to strangers".

"He is content to just sit there until he's asked a question".

"He tells you he has no friends...it will put him in a mood for about an hour".

"When you ask him a question...it takes a bit to register...and you have got to give him time to answer".
Signs of social anxiety in Cornelia de Lange Syndrome

- Skin picking
- Fidgeting during interaction
- Asking repetitive questions
- Avoidance/delay behaviours
- Challenging behaviours
- Selective mutism

Prevalence of Selective Mutism

Problems with expressive communication and anxiety (especially social anxiety)
The key issues

- Physical difference and disorder (health problems)
- Self-injury
- Poor expressive communication
- Autistic spectrum like behaviours
- Specific cognitive impairments
- Changes with age
GROWING UP WITH CdLS
CHANGES IN ADOLESCENCE AND YOUNG ADULTHOOD

By Dr. Anna Wellman, Dr. Ian Moss and Professor Chris Oller
Centre for Neurodevelopmental Disorders, University of Birmingham
Some important differences in how the mind works

- Executive function (the manager of the mind)
  - Working memory (remember and use)
  - Inhibiting (don’t do that!)
  - Shifting (attending to one thing and then moving to another)
Are there specific cognitive impairments in CdLS?

The Cards to be Sorted

Number correct

Maximum length

p<.01
Some important differences in how the mind works

- Executive function (the manager of the mind)
  - Working memory (remember and use)
  - Inhibiting (don’t do that!)
  - Shifting (attending to one thing and then moving to another)

- ... is related to social anxiety\less speech in social interactions (see how you do when you talk to someone new!)

Signs of social anxiety in Cornelia de Lange Syndrome

- Skin picking
- Fidgeting during interaction
- Asking repetitive questions
- Avoidance/delay behaviours
- Challenging behaviours
- Selective mutism

Prevalence of Selective Mutism

- Moss et al., 2008. AJMR 113, 278-291;
- Collie et al., In preparation;
- Reid, Nelson, Moss & Oliver, In preparation
Age group MIPQ-S Interest and Pleasure Subscale Score

11 yrs and under 12-15 yrs 16-18 yrs 19-22 yrs 23-28 yrs 29 yrs and above

Increased cognitive demand
Prevalence of selective mutism

<table>
<thead>
<tr>
<th></th>
<th>Inhibit subscale</th>
<th>Shift subscale</th>
<th>Emotional control subscale</th>
<th>Working memory subscale</th>
<th>Plan/Organise subscale</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cornelia de Lange Syndrome</td>
<td>-.41*</td>
<td>-.26</td>
<td>-.31</td>
<td>-.57**</td>
<td>-.62**</td>
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<tr>
<td>Down syndrome</td>
<td>-.24</td>
<td>.07</td>
<td>-.12</td>
<td>.10</td>
<td>-.07</td>
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</tbody>
</table>

Correlations between participant speech and executive function

No problem with word finding, grammar, vocabulary, content of speech.

Significantly more pauses and blocks
Mean number of words spoken by each participant group during the 3 minutes of coded conversation.

Rate of Gestures produced per one-hundred words spoken by each participant group.

Sotaro Kita and Eva Nielsen
Adaptive Behaviour Receptive Language

Time: \(F_{1,45} = 7.79; p = .008\)

Group: \(F_{1,45} = 15.67; p < .001\)

Time x Group: \(F_{1,45} = 5.69; p = .02\)

Group: \(F_{1,45} = 8.72; p = .005\)

Play/Imagination

Repetitive Behaviour

Group: \(F_{1,45} = 8.72; p = .005\)
The take home messages

- Health and pain
  - Know
  - Act
  - Advocate
- Early hearing problems and speech
- Autism Spectrum Disorder
  - Does the diagnosis help me and my child
- Thinking differently
  - Predictability and routine (softly, softly)
  - Imbalance in ability
  - Support for what if’s
- The future
  - Planning for change
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PhD and MPhil in CdLS funded by Big Lottery and MRC (1999-2003)
PhD in CdLS funded by Big Lottery (2008-2009)
PhD in CdLS funded by Cerebra (2009-2012)
PhD in CdLS funded by UK and Ireland group (2011-2014)
Debbie Allen, Jane Appleby, Ian Apperly, Sarah Beaumont, Sarah Beck, Lisa Collis, Fay Cook, Louise Davies, Kate Eden, Ruth Fishwick, Christina Goredema, Sarah Gomiak, Glyn Humphreys, Abby Marr, Jonathan Martin, Anna Mitchell, Chris Oliver, Jan Oyebode, Jane Petty, Laurie Powis, Barzan Rahman, Donna Reid, Caroline Richards, Kristina Stockdale-Juhberg, Penny TunesiCillie, Lucy Wilde, Kate Woodcock.

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Cerebra

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Birmingham Children's Hospital
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Newlife
National Autistic Society
ESRC
Tuberous Sclerosis Alliance

www.birmingham.ac.uk/cnnd
C.Oliver@Bham.ac.uk
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