Emotion regulation difficulties in rare genetic conditions associated with visual impairment: new insights for clinical practice

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Rare genetic syndromes associated with intellectual disability

- Lowe
- Bardet-Biedl
- Alstrom
Background – Lowe Syndrome

Lowe Syndrome (LS) is an X-linked recessive, multi-system, metabolic disorder

1 in 500,000

Mutations in the OCRL-1 gene.

Intellectual disability (ID): 50-65% reported to be in the severe to profound range.

Corrected visual acuity is rarely better than 20/100.

(Lewis, Nussbaum and Brewer, 2012)
Temper Outbursts

Characterised by a cluster of behaviours including shouting, crying, screaming, arguing, facial flushing, stamping and destruction

Prevalence in ID populations

- **Down syndrome**: 30 - 35%
- **Cri du chat**: 67%
- **Lowe**: 74 - 80%
- **Prader-Willi**: 88%

(Dyckens et al., 2005; Kutsch et al., in prep; Smith et al., 1996)
Clinically significant outbursts:

A combination of unusual frequency, duration or severity (based on aggressive or destructive behaviours) indicated clinically significant emotional dysregulation requiring professional intervention in pre-school children.

Wakschlag et al. (2007)
Boys with Lowe syndrome: Ages ranged from eight to 37 years ($M = 18.29$ years; $n = 9$ under 18 years; $n = 8$ adults 18 years or over).

Temper outburst interview (Tunnicliffe et al., 2014)

<table>
<thead>
<tr>
<th>Response</th>
<th>Frequency</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Timing of the next outburst:</strong></td>
<td></td>
</tr>
<tr>
<td>Within the next 15 min</td>
<td>2</td>
</tr>
<tr>
<td>Within the next hour</td>
<td>6</td>
</tr>
<tr>
<td>By this time tomorrow</td>
<td>6</td>
</tr>
<tr>
<td>By this time next week</td>
<td>2</td>
</tr>
<tr>
<td>By this time next month</td>
<td>1</td>
</tr>
<tr>
<td><strong>Duration of longest outburst in the last month:</strong></td>
<td></td>
</tr>
<tr>
<td>Less than a minute</td>
<td>0</td>
</tr>
<tr>
<td>Less than 5 minutes</td>
<td>4</td>
</tr>
<tr>
<td>Less than 15 minutes</td>
<td>4</td>
</tr>
<tr>
<td>Less than an hour</td>
<td>5</td>
</tr>
<tr>
<td>More than an hour</td>
<td>4</td>
</tr>
</tbody>
</table>

Cressey, 2017
<table>
<thead>
<tr>
<th></th>
<th>Lowe Syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Physical aggression</td>
<td>15/17 (88%)</td>
</tr>
<tr>
<td>Any aggression</td>
<td>16/17 (94%)</td>
</tr>
<tr>
<td>Self injury</td>
<td>6/17 (35%)</td>
</tr>
</tbody>
</table>

*Cressey, 2017*
Pain and Discomfort
Does the prevalence of self-injury and aggression vary across syndromes?

Conducting A FLACC

Face
Legs
Activity
Crying
Consolability

1 to 2 minutes

No association between pain, health conditions and temper outbursts in Lowe

No associations with visual ability
Consequence

Antecedent

Behaviour

Consequence

(Carr & Durand, 1985)
Social Communicative Function of Challenging Behaviour: Positive Reinforcement

- Need for others to do or give something
- Increase in chance of CB
- REWARD
  - Positive Reinforcement
- ENGAGE
  - Comfort
  - Reprimand
  - Offer
  - Restrain
  - Occupy
  - Distract
- ACTION
- AVERSIVE!
  - Concern
  - Frustration
  - Anxiety
  - Confusion
  - Distress

Need for others to stop something

Increase in chance of CB

REWARD

Negative Reinforcement

DISENGAGE

Comfort
Reprimand
Offer
Restrain
Occupy
Distract

CONCERN
Frustration
Anxiety
Confusion
Distress

AVERSIVE!

ACTION
Pathways from genes to behaviour: temper outbursts

Consequence
Behaviour is operantly reinforced
(Carr & Durand, 1985)

Antecedent

Temper Outburst
Aggression
Self-Injury

Kutsch et al., in prep
Behavioural interventions?

- Teaching a sign/word to request an object (give every time initially when sign is used)

- Not delivering the object in response to aggression/outbursts (break the association)

- Ensuring the object is given when behaviour is not present.
Primary trigger

This included frustrated goals \((n = 1)\), delayed gratification \((n = 1)\), “not getting what he wants” or “not getting his own way” \((n = 6)\), “not being able to do something he wants to do” \((n = 2)\).
Emotional Regulation Difficulties

Failure of the brain centres such as the ventromedial frontal cortex to dampen activation of the amygdala.

These regions are important for learning and extinguishing fears.
Consequence

Behaviour is operantly reinforced (Carr & Durand, 1985)

Pathways from genes to behaviour: temper outbursts

Antecedent (thwarted)

Genetic change

Consequence

Emotional Regulation Difficulty
Participants

26 boys and men with Lowe syndrome aged 6-34 years (Mage VABS Language Composite (Receptive/Expressive) ranged 2 yrs 6 m to 10.25 months (Mean: 5 yrs 10 m).
Questionnaires/Rating Scales

Behaviour Rating Inventory of Executive Function - Preschooler Version (BRIEF-P; Gioia et al. 2001).

Adapted Challenging Behaviour Questionnaire (CBQ; Hyman, 2002).

Vineland Adaptive Behavior Scales-II (VABS-II; Sparrow et al, 2005).

Current Health Questionnaire & Non-Communicating Child Pain Checklist (Breau et al., 2002).
Computerised Tests

Teddy Task (Sonuga-Barke, 2002)

SwIFT (Carroll & Fitzgibbon, 2015)

Go No-Go Task (Simpson et al., 2006)
<table>
<thead>
<tr>
<th></th>
<th>Daily Living Composite</th>
<th>Outburst Score</th>
<th>Delay of Gratification</th>
<th>Shifting</th>
<th>Inhibition</th>
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</thead>
<tbody>
<tr>
<td>Language Composite</td>
<td>0.83**</td>
<td>-0.69**</td>
<td>0.31</td>
<td>0.43</td>
<td>0.78**</td>
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<tr>
<td>Daily Living</td>
<td></td>
<td>-0.64*</td>
<td></td>
<td>0.46</td>
<td>0.65*</td>
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<tr>
<td>Outburst Score</td>
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<td></td>
<td>-0.51*</td>
<td>-0.24</td>
<td>0.44</td>
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<tr>
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<td></td>
<td></td>
<td>0.13</td>
<td>-0.45</td>
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<tr>
<td>Shifting</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td>-0.71*</td>
</tr>
</tbody>
</table>

**p < .005; * p < .01
Spearman’s Rho Correlations
Conclusions

1. Evaluation of how well a child regulates their emotion may be important to consider, as well as traditional behavioural interventions.

2. Implications for other genetic syndromes associated with visual impairment (BBS; bilateral optic nerve hypoplasia)
Points for practice – TD individuals

*Emotional understanding* might act as a mediator for some forms of emotional regulation

*Parental scaffolding* to help a child try out new emotional regulation strategies (prompting and instruction in problem solving and cognitive reappraisal)

*Stop-think-relax* and *mindfulness based interventions* with older individuals
Points for practice – LD individuals

1. Self-monitoring interventions
2. Relaxation (counting and breathing)

Rose et al. (2000) Anger management (adults)

- Role play (video feedback)
- Daily review with a trusted person
- Identification of triggers
- Thought-stopping
- Positive self statements