



UNIVERSITY OF
BIRMINGHAM



The importance of sleep for children with rare genetic syndromes: DYRK1A syndrome

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Today's talk

What is sleep?

Sleep in rare genetic syndromes.

What maintains sleep problems?

What can we do to improve children's sleep?



What is sleep?

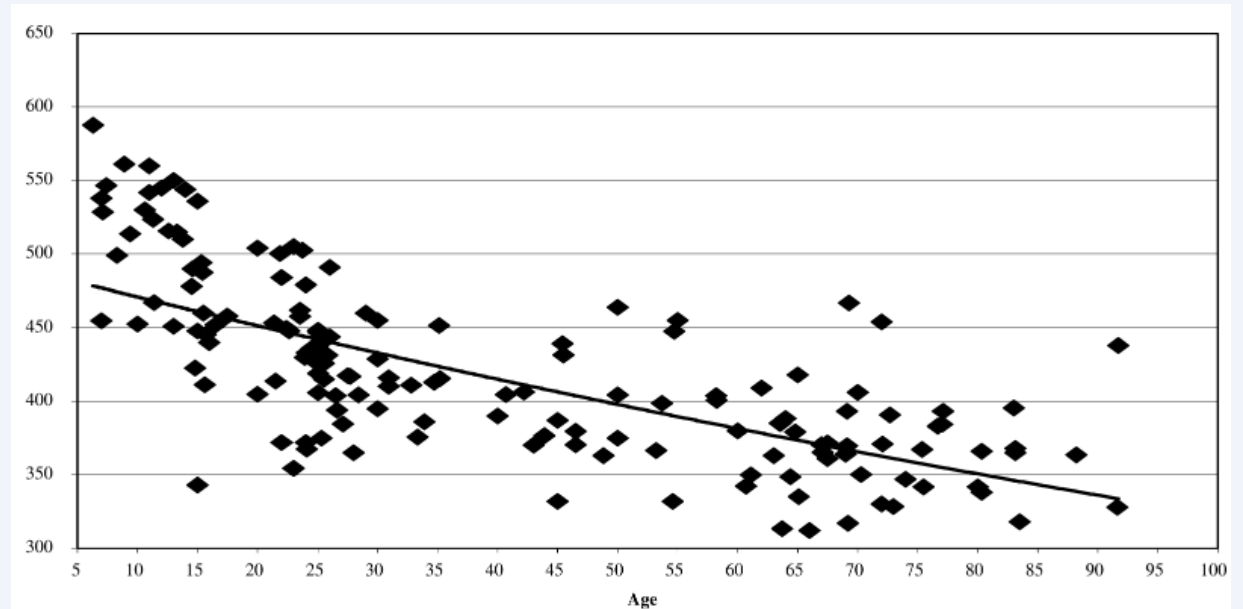
Sleep is essential.



Sleep is integral for all aspects of human functioning:

- behaviour regulation
- learning, memory and attention
- mood
- physical health

Sleep changes throughout life. Childhood seems to be when we need the most sleep.



Sleep difficulties are common in rare genetic syndromes

| | |
|----------------------------|--------|
| Angelman syndrome | 48-96% |
| Fragile X syndrome | 40-84% |
| Prader-Willi syndrome | 53-70% |
| Rett syndrome | 71-86% |
| Tuberous sclerosis complex | 43-59% |
| DYRK1A syndrome | ~50% |

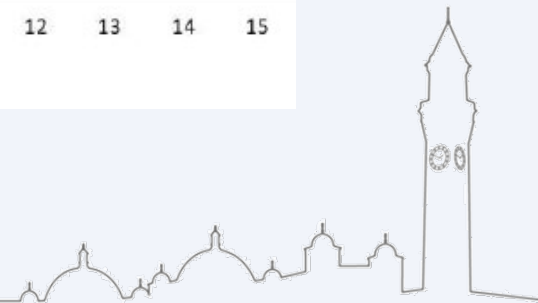
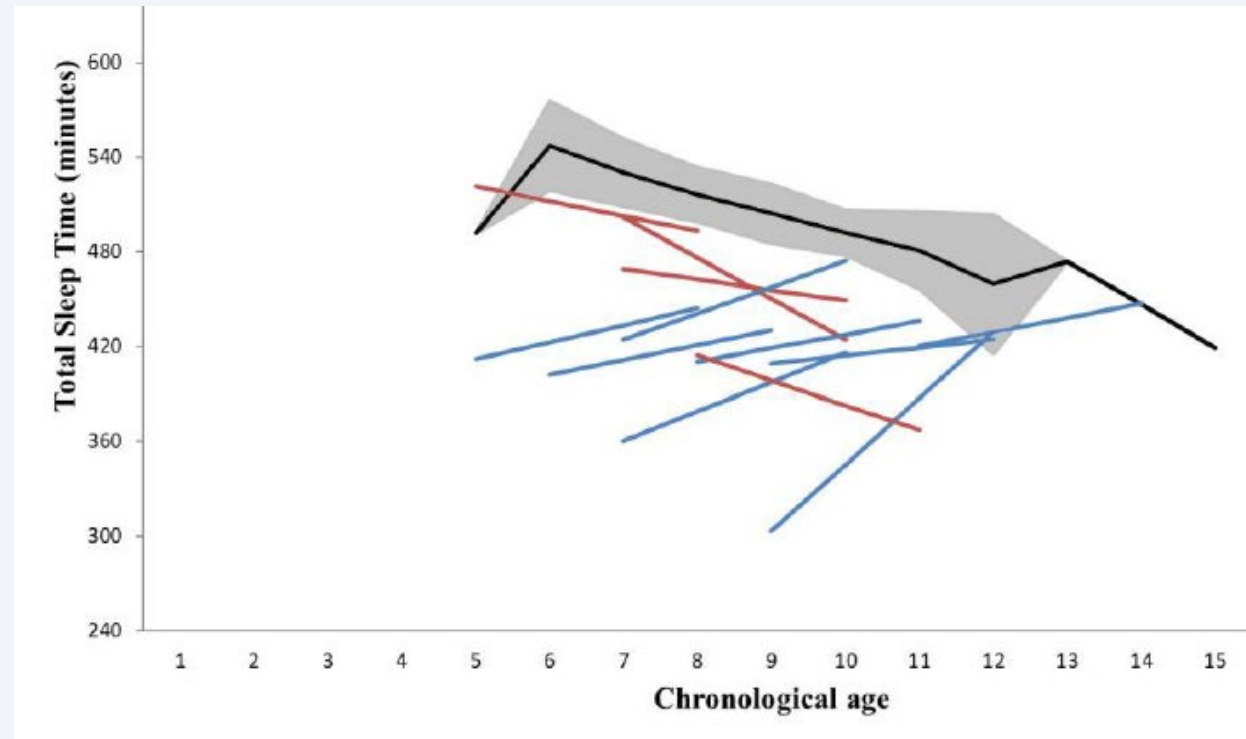
Falling sleep
Staying asleep
Sleep-disordered breathing

Less established findings.
Problems falling asleep and staying asleep.



Sleep difficulties may also be persistent

In several syndromes, sleep difficulties remain consistent as children age.



Effect of sleep difficulties in rare genetic syndromes

Often associated with DYRK1A syndrome

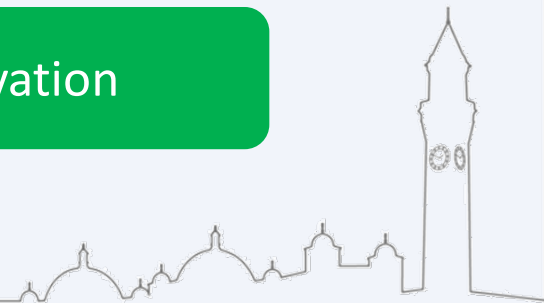
Challenging behaviour (self-injury, aggressive behaviour, hyperactivity, impulsivity)

Sleep difficulties associated with:

Lower mood and greater anxiety

Greater autism-related characteristics

Greater caregiver stress and sleep deprivation



Today's talk

What is sleep?



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What maintains sleep problems?

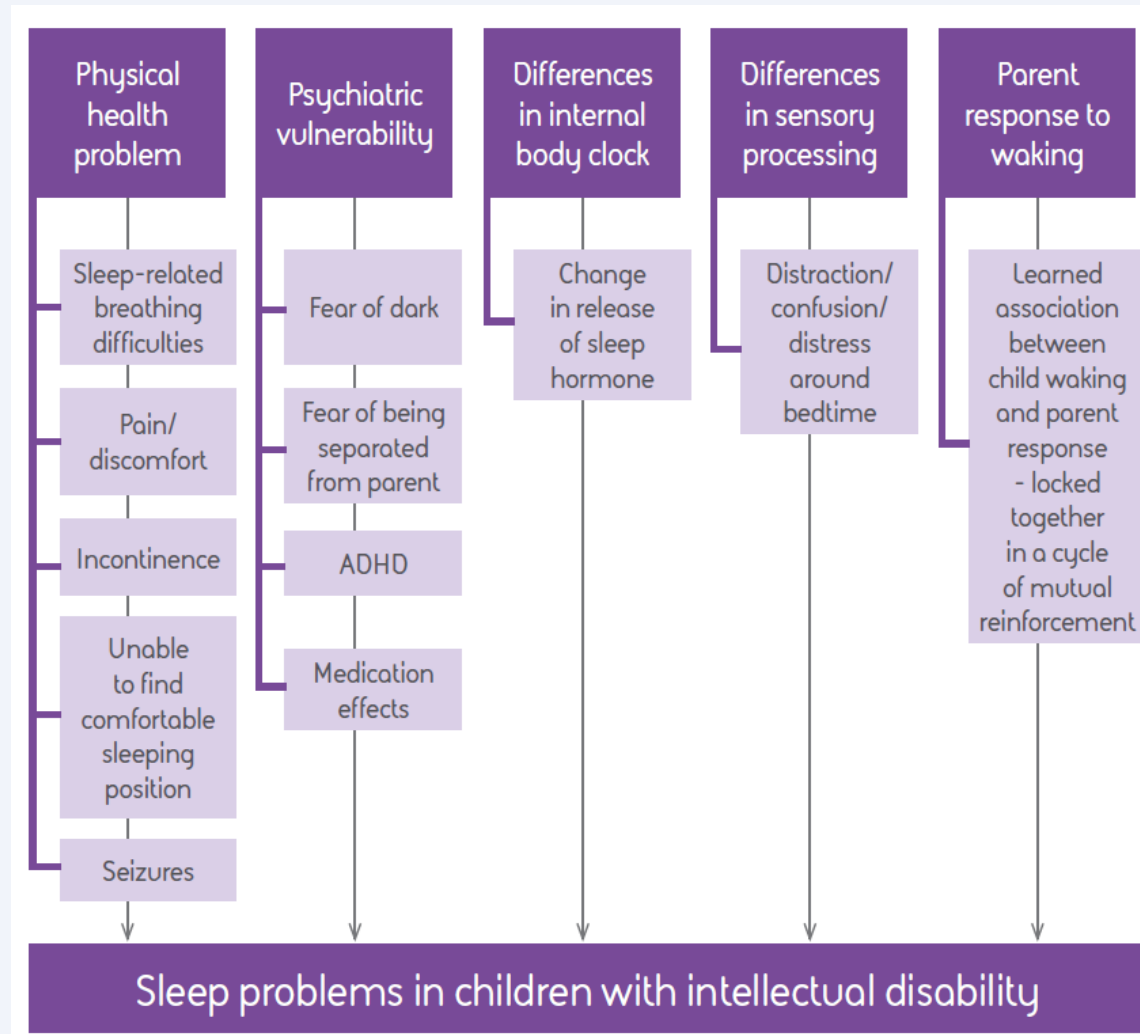
What can we do to improve children's sleep?

Sleep difficulties are common, often persistent, and can have lots of effects in rare genetic syndromes.

What is causing this?



Lots of possible causes of poor sleep



Dr Georgie Agar



Possible causes of poor sleep in DYRK1A syndrome

Possible causes

Seizures

Painful health conditions

(e.g. gastrointestinal reflux, constipation, ophthalmalopia)

Autism

Behavioural insomnia

Prevalence

33 – 72%

29 – 79%

43 – 88%



Seizures

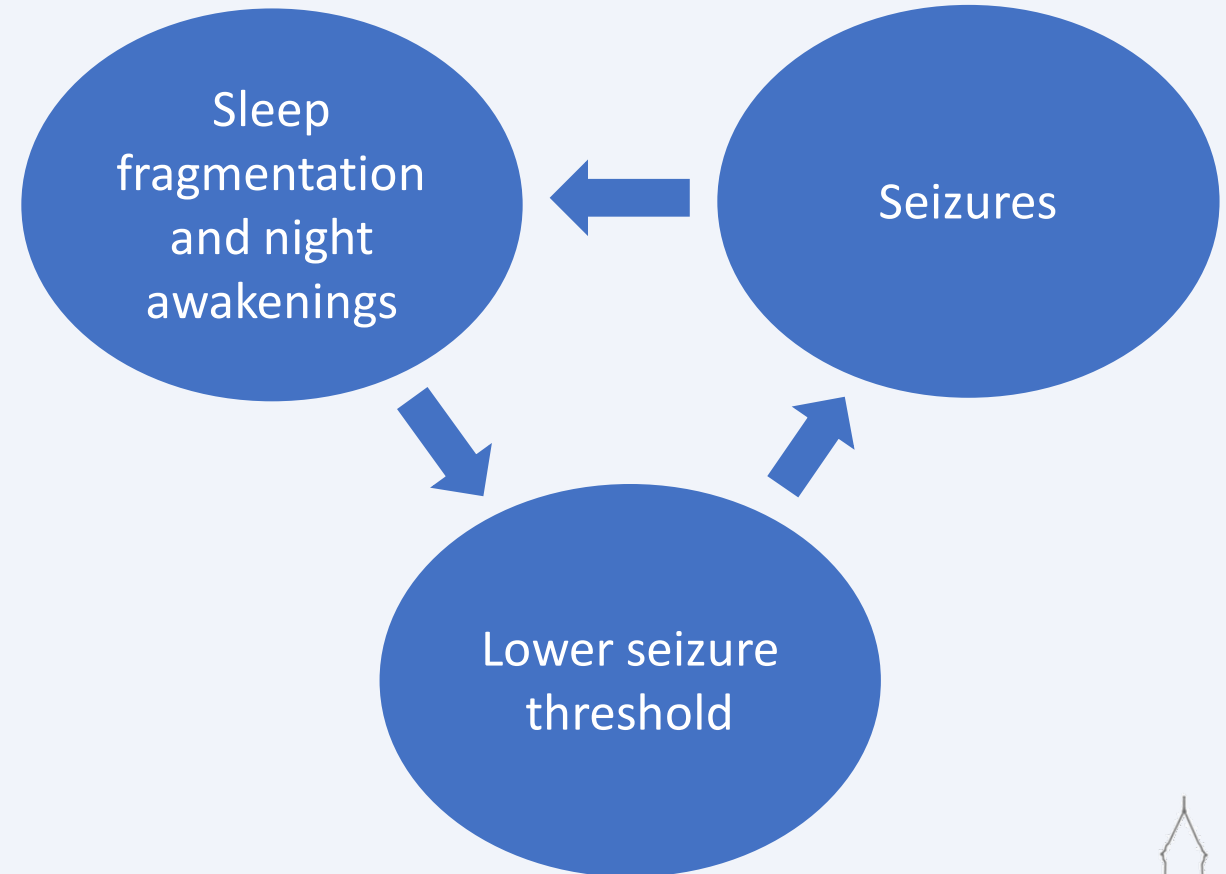
33 – 72% in DYRK1A

Seizures associated with greater sleep problems in genetic syndromes, and intellectual disability.

Sleep fragmentation

Shortened sleep duration

Daytime sleepiness



Anti-epileptic drugs – effect on sleep

AEDs control seizures. May improve nocturnal sleep.

However, some AEDs affect REM and daytime sleepiness.

Individuals with intellectual disability: not 100% sure how AEDs affect sleep.

Evidence that behavioural sleep strategies can improve sleep in children with epilepsy.



Possible causes of poor sleep in DYRK1A syndrome

| Possible causes | Prevalence | |
|---|------------|---|
| Seizures | 33 – 72% | ✓ |
| Painful health conditions (e.g. gastrointestinal reflux, constipation, ophthalmalopia) | 29 – 79% | |
| Autism | 43 – 88% | |
| Behavioural insomnia | | |



Painful health conditions

Pain increases difficulty of falling asleep and staying asleep

These difficulties are common amongst individuals with chronic pain

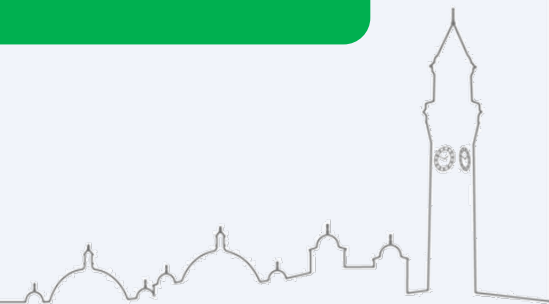
Rare genetic syndromes: Sleep difficulties associated with painful health conditions

Notably gastrointestinal reflux symptoms

DYRK1A syndrome - 29 – 79% experience painful/discomforting health conditions

Such as gastrointestinal reflux, constipation, and ophthalmalgia

Should consider the effect of these health conditions on sleep



Detecting pain

Health conditions need to be identified before they are treated.

In the general population, pain is identified via self-report.

Delayed/absent speech common in DYRK1A syndrome (62-100%)

The FLACC Pain Scale

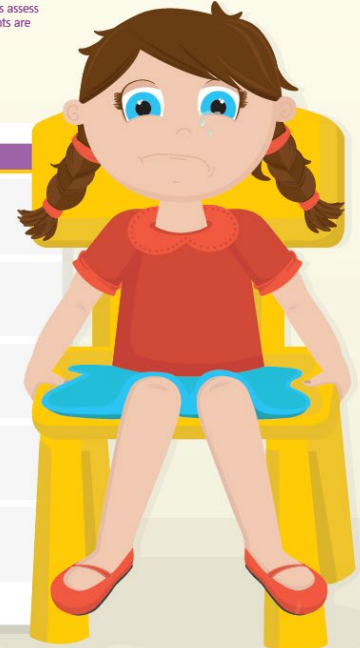
Sometimes it is difficult to assess pain in children who are non-verbal. The FLACC Pain Scale is a system that can help parents and professionals assess pain levels in children who have limited or no expressive communication. The diagram shows the categories for scoring. Zero, one or two points are given to each of the five categories: Face, Legs, Activity, Cry and Consolability.

Interpreting the Behaviour Score
Each category is scored on the 0-2 scale, which results in a total score of 0-10

- 0 relaxed and comfortable
- 1 mild discomfort
- 2 moderate pain
- 3 severe discomfort or pain or both

| Categories | Score Zero | Score One | Score Two |
|------------------------|--|--|--|
| F Face | No particular expression or smile | Occasional grimace or frown, withdrawn, disinterested. | Frequent to constant quivering chin, clenched jaw. |
| L Legs | Normal position or relaxed | Uneasy, restless, tense. | Kicking, or legs drawn up. |
| A Activity | Lying quietly, normal position moves easily. | Squirming, shifting back and forth, tense. | Arched, rigid or jerking. |
| C Cry | No crying (awake or asleep) | Moans or whimpers; occasional complaint | Crying steadily, screams or sobs, frequent complaints. |
| C Consolability | Content, relaxed. | Reassured by occasional touching hugging or being talked to, distractable. | Difficulty to console or comfort |

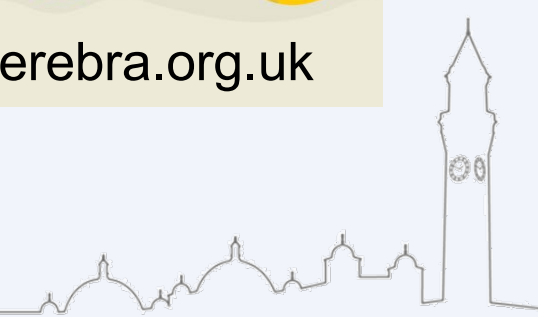
If a child is showing these behaviours, it doesn't necessarily mean that they are in pain, as some of the behaviours measured by the FLACC scale can happen for other reasons. However, parents are advised to follow up high scores with a professional.



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REFERENCE Merkel, S. I., Voepel-Lewis, T., Shayevitz, J. R., & Malviya, S. (1997). The FLACC: A behavioral scale for scoring postoperative pain in young children. *Pediatric Nursing*, 23(3), 293-297.

www.cerebra.org.uk



Possible causes of poor sleep in DYRK1A syndrome

| Possible causes | Prevalence | |
|---|------------|---|
| Seizures | 33 – 72% | ✓ |
| Painful health conditions (e.g. gastrointestinal reflux, constipation, ophthalmalopia) | 29 – 79% | ✓ |
| Autism | 43 – 88% | |
| Behavioural insomnia | | |



Autism/autism-related behaviours

May impact sleep in many ways.

Difficulty transitioning between activities

Preferences for routines/Insistence on sameness

Communication differences

Other behaviours too...

Difficulties going to bed and trying to fall asleep

Breaking bedtime routines or changing sleep environments may cause distress

Difficulty understanding bedtime instructions and routines



Possible causes of poor sleep in DYRK1A syndrome

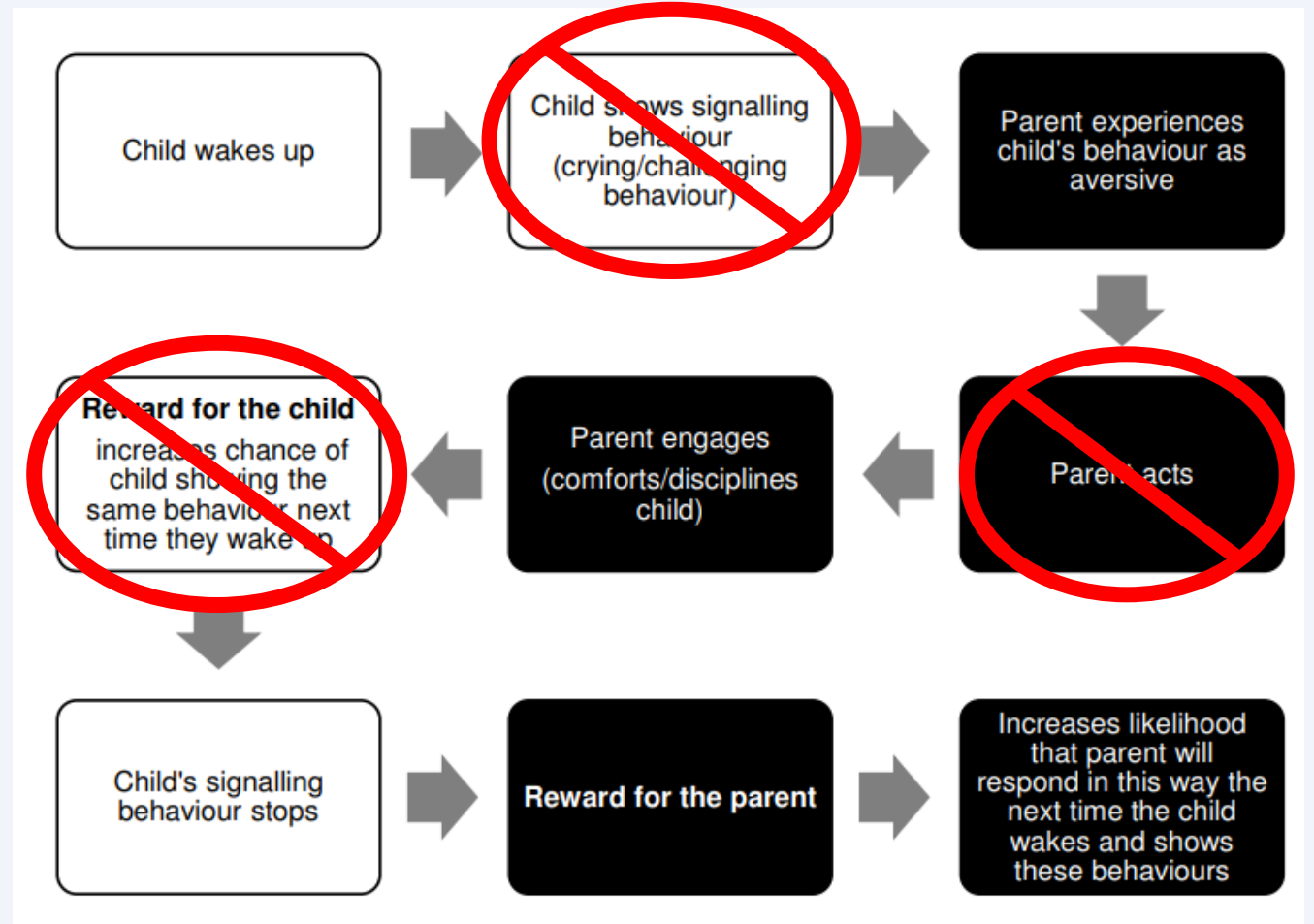
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Behavioural insomnia

Process of rewarding behaviours that extend bedtime settling and night awakenings.

We want to stop rewarding these behaviours, and therefore reduce settling difficulties and night awakenings.



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What maintains sleep problems?



What can we do to improve children's sleep?

There are several possible causes of sleep disruptions in children with DYRK1A.

What can we do to improve children's sleep?



What can we do to improve children's sleep?

First and foremost: resolving painful/discomforting health conditions, and seizures.



What can we do to improve children's sleep?

Cerebra Sleep Guide - behavioural strategies

Sleep problems we may expect to see in children with rare genetic syndromes.

Context-specific strategies for improving children's sleep.

Range from behavioural strategies to sleep hygiene.



Dr Georgie Agar



<https://cerebra.org.uk/download/sleep-a-guide-for-parents/>



What can we do to improve children's sleep?

Cerebra Sleep Advice Service


Complete online form and sleep diary for two weeks

Telephone consultation with sleep practitioner

HOME > GET ADVICE AND SUPPORT > SLEEP ADVICE SERVICE > CONTACT THE SLEEP TEAM

Contact the Sleep Team

Please fill in our referral form to let us know a little bit about your child's sleep problem. Then we'll email you a sleep pack to fill in. When we receive your completed sleep application and diary you will be assigned a sleep practitioner. They'll then arrange a telephone consultation with you and will give you support over the phone.



<https://cerebra.org.uk/get-advice-support/sleep-advice-service/contact-the-sleep-team/>



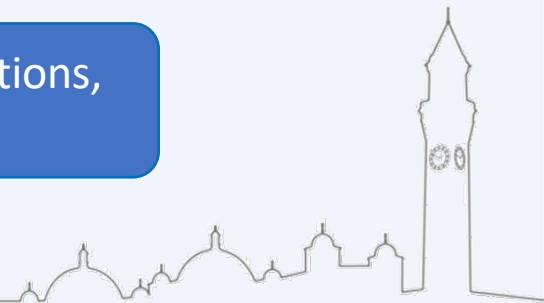
What can we do to improve children's sleep?

Pharmaceutical options

Most researched and implemented is melatonin.

Most effective for falling asleep. Not for night awakenings.

Recommended that melatonin is administered following unsuccessful behavioural interventions, or alongside behavioural interventions.



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What can we do to improve children's sleep?



How are we addressing this?



How are we addressing this?



How are we addressing this?



Survey – completed online or posted to you

Measuring several aspects of living with a rare genetic syndrome:

Behaviour
Sleep
Physical health
Mental health
Experiences with health services
Caregiver wellbeing

Personalised feedback reports provided after completing the survey

Access online survey/request
postal pack here!



<https://www.cerebranetwork.com/beond>



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