

CdLS Foundation

Neurodevelopmental Disorders and Behavioral Management in Cornelia de Lange Syndrome (CdLS)

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Outline

- Background
- Language Disorders
- Attention Deficit Hyperactivity Disorder
- Anxiety/Depression
- Aggression/Self-Injury
- Conclusions



Warnings

- Non-medication interventions can be effective too, but I will try to focus on medications for this talk
- The information presented here is a combination of anecdotal and observational data from individuals with CdLS, and extrapolated data from other neurodevelopmental disorders
- In general, there is a great need for **evidence-based** research on neurobehavioral interventions for CdLS

Background

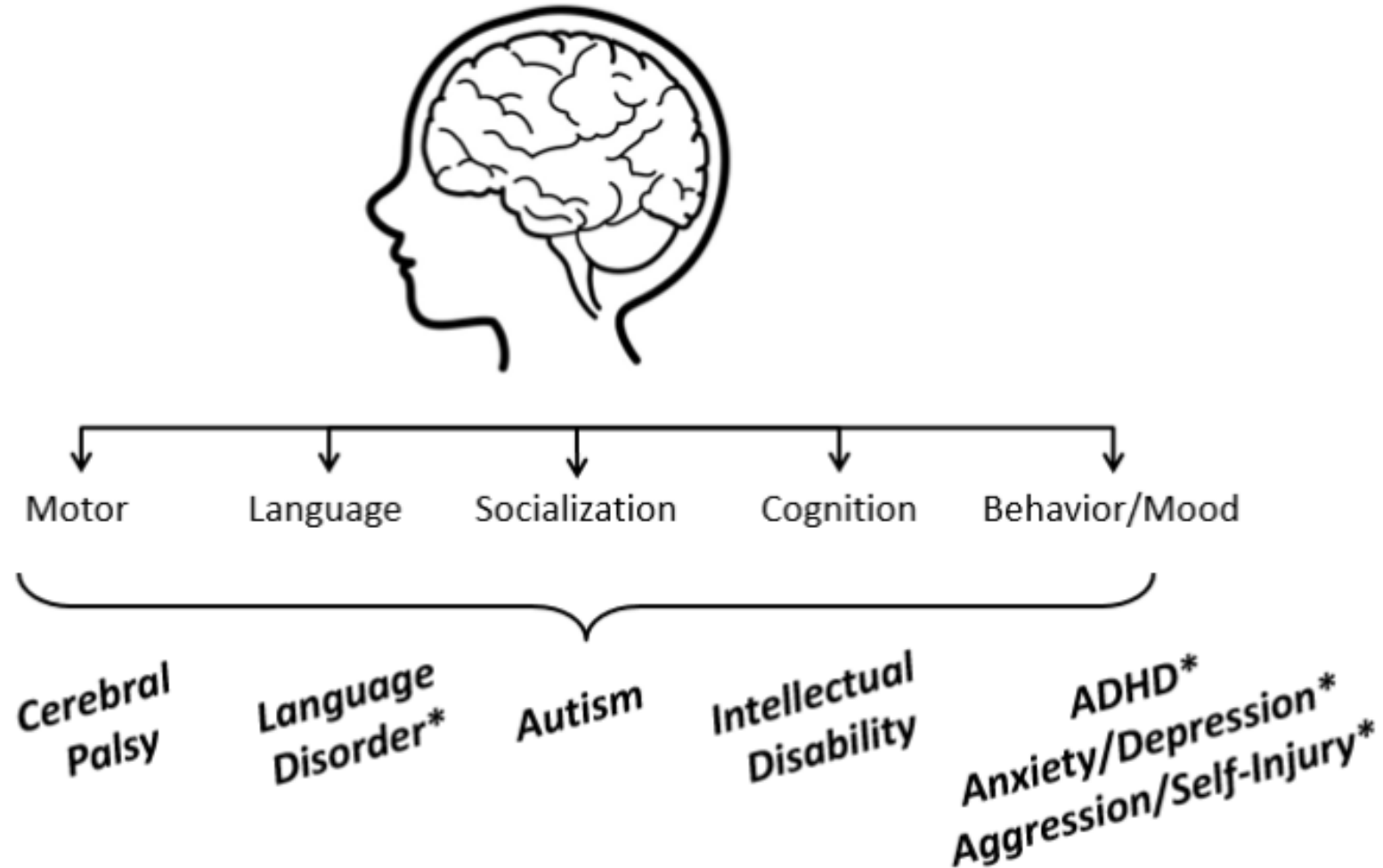
CdLS

- CdLS is associated with distinct facial features, intellectual disability, autism spectrum disorder, and multiple systemic features
- Behavioral features such as hyperactivity, aggression, and anxiety are common

Significance

- Challenging behaviors and developmental concerns can affect the **quality of life** for individuals with CdLS and their families
- Addressing these concerns early on – through medications, therapies, or other interventions – may help optimize long-term neurodevelopmental outcomes

Development



Language

Language affects our ability to interface with the rest of the world (e.g., socialize, communicate)

**Expressive
Language**



**Ability to
communicate**

**Receptive
Language**

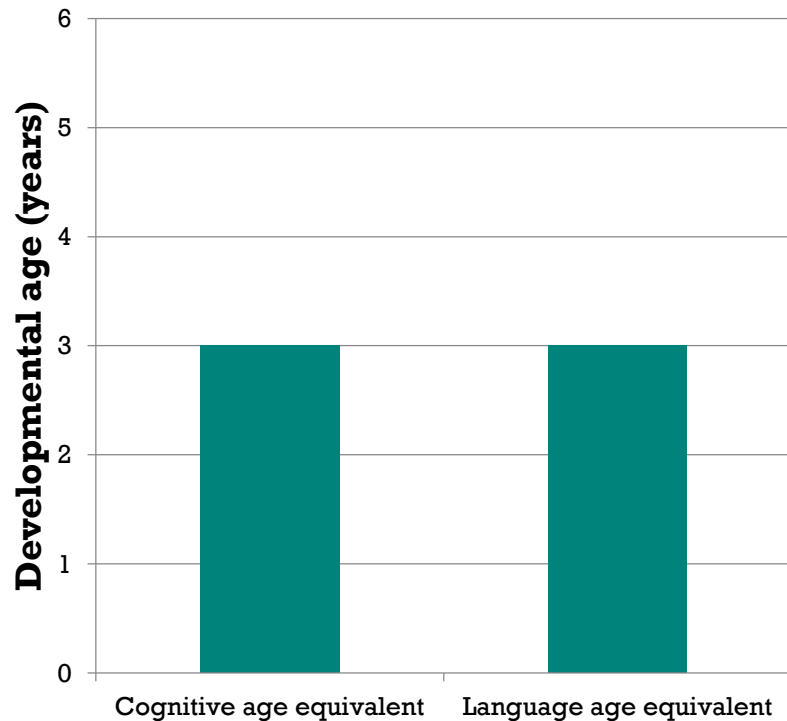


**Ability to
understand**

Language and Cognition

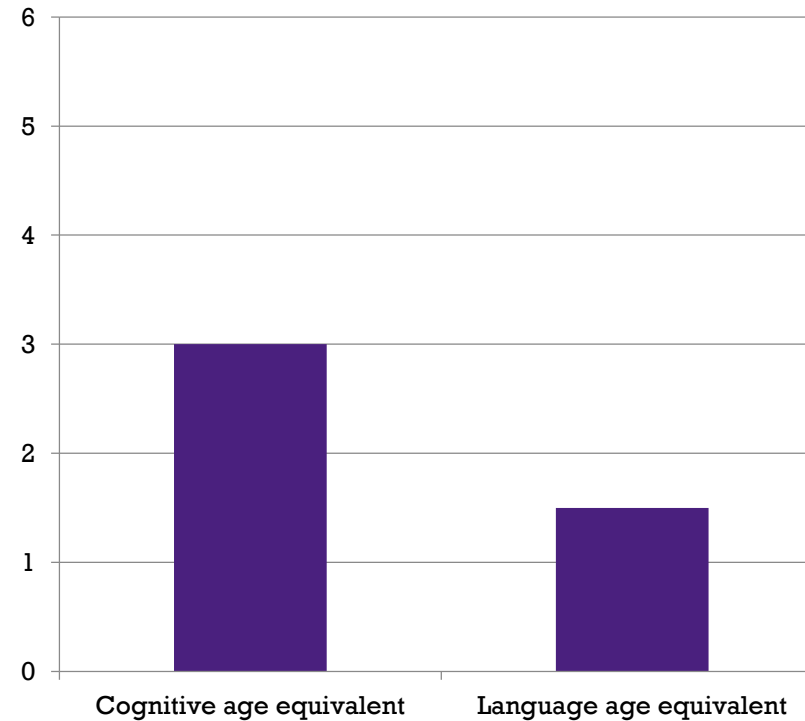
Intellectual Disability

6 year old, IQ = 50



Intellectual Disability PLUS Language Disorder

6 year old, IQ = 50



Language Disorders

Language Disorders Treatment



Individual Speech/Language Therapy



Play/Social Groups



Augmentative and Alternative
Communication (AAC)

AAC

- AAC refers to the use of special devices that allow children to communicate even in the absence of spoken speech
- These devices can be high tech or low tech
- AAC is especially useful for children who are non-verbal

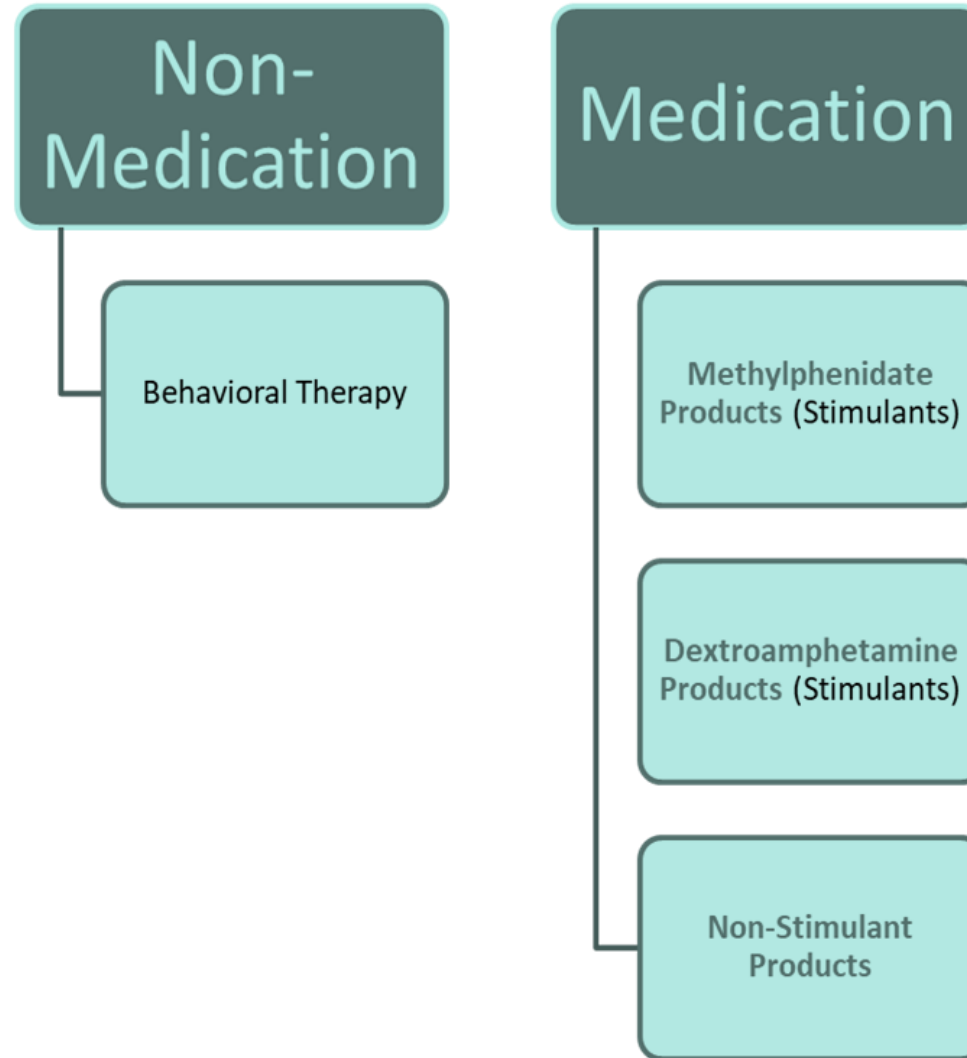


Attention Deficit Hyperactivity Disorder (ADHD)

ADHD

- ADHD is characterized by impairment of attention, distractibility, hyperactivity, and impulsivity
- Symptoms must be present in two or more settings (e.g., home, school)
- Symptoms must cause functional impairments (e.g., social, academic)

ADHD Treatment



ADHD Medications

Treatment may require a trial-and-error approach

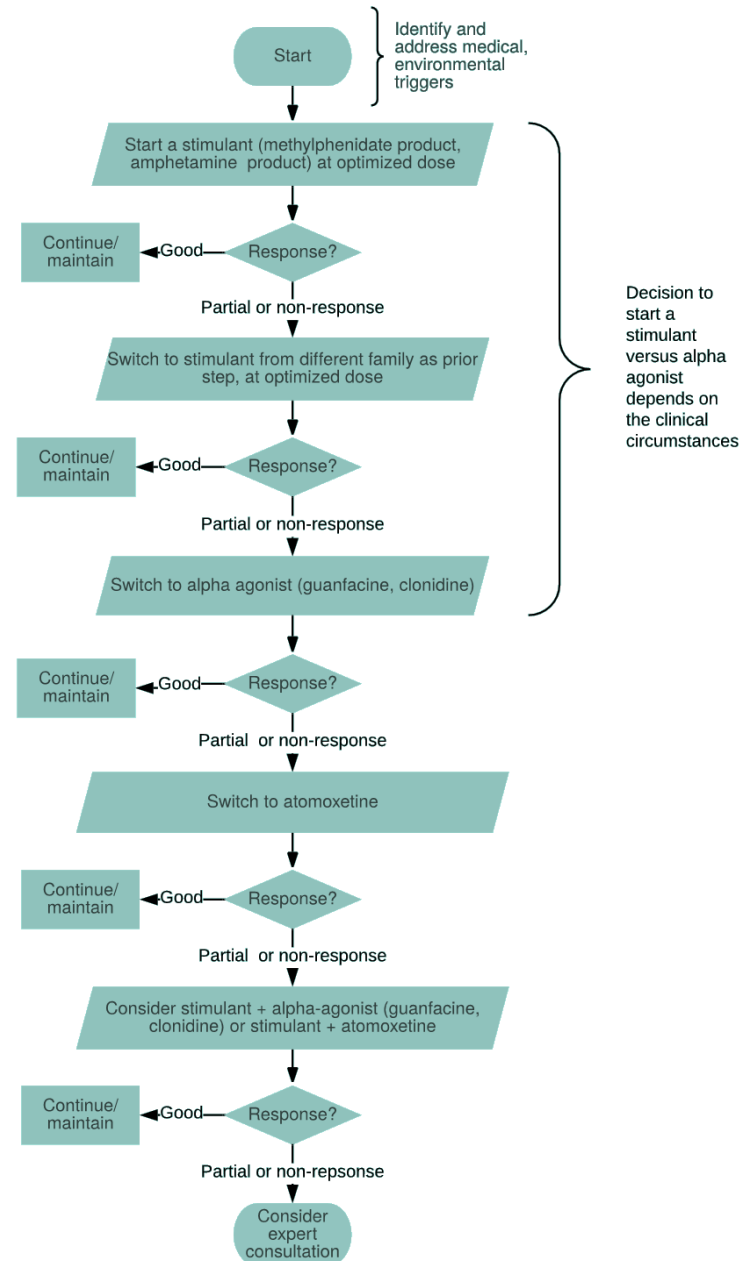
- “Start low, go slow”
- Main side effects are decreased appetite, disrupted sleep
- Wait at least 1 week before deciding whether a new medication or dose change is ineffective

ADHD Medications

	Pill (can't crush/chew)	Capsule (can open up and sprinkle onto food)	Chewable Pill (can chew)	Liquid	Patch
METHYLPHENIDATE PRODUCTS					
Short Acting	Focalin Ritalin		Methylin Chewable	Methylin Solution	
Long Acting	Concerta Ritalin SR	Focalin XR Ritalin LA Metadate CD		Quillivant XR	Daytrana
AMPHETAMINE PRODUCTS					
Short Acting	Adderall			ProCentra	
Long Acting	Vyvanse Dexedrine	Adderall XR			
NON-STIMULANT PRODUCTS					
Short Acting	Clonidine Guanfacine				
Long Acting	Gunfacine ER Clonidine ER Atomoxetine				Clonidine patch



General Approach to Treatment of ADHD in CdLS

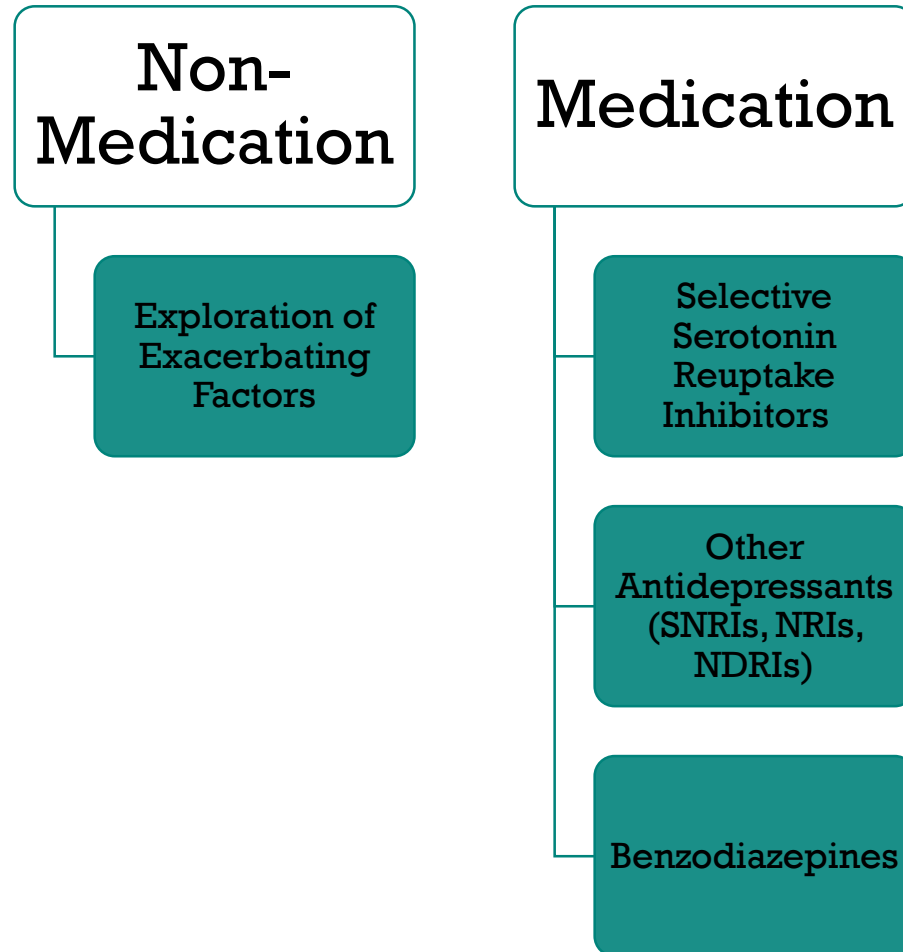


Anxiety and Depression


Anxiety and Depression

- Symptoms can include irritability, excessive worry, perseveration, social avoidance
- Anxiety and depression can be difficult to recognize, especially with increasing levels of intellectual disability associated with CdLS

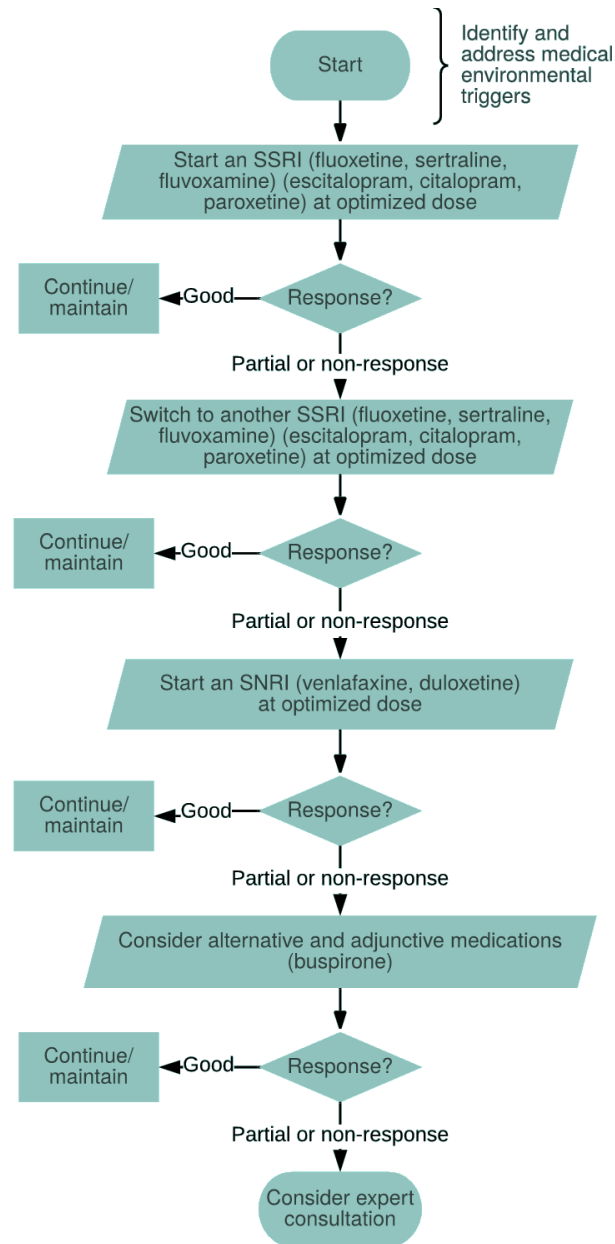
Anxiety and Depression Treatments



Anxiety and Depression Medication

- A **selective serotonin reuptake inhibitors (SSRI)** may help anxiety and mood issues in CdLS
- **CAVEAT:** SSRIs can lead to **behavioral activation** and **worsening agitation** in some individuals 
- It may be prudent to avoid benzodiazepines for the management of anxiety due to risk of dependency and paradoxical agitation

General Approach to Treatment of Anxiety and Depression in CdLS



Aggression/Self-Injury

Aggression/Self-Injury

- Aggressive/self-injurious behaviors include head-banging, hand-biting, and excessive scratching
- They can be a source of significant parental distress for families affected by CdLS

Aggression/Self-Injury Treatment

- Aggressive, self-injurious behaviors require a full behavioral assessment in order to identify triggers
- Sometimes undiagnosed medical problems – such as reflux, dental issues – can worsen aggression/self-injury

Aggression/Self-Injury Medications

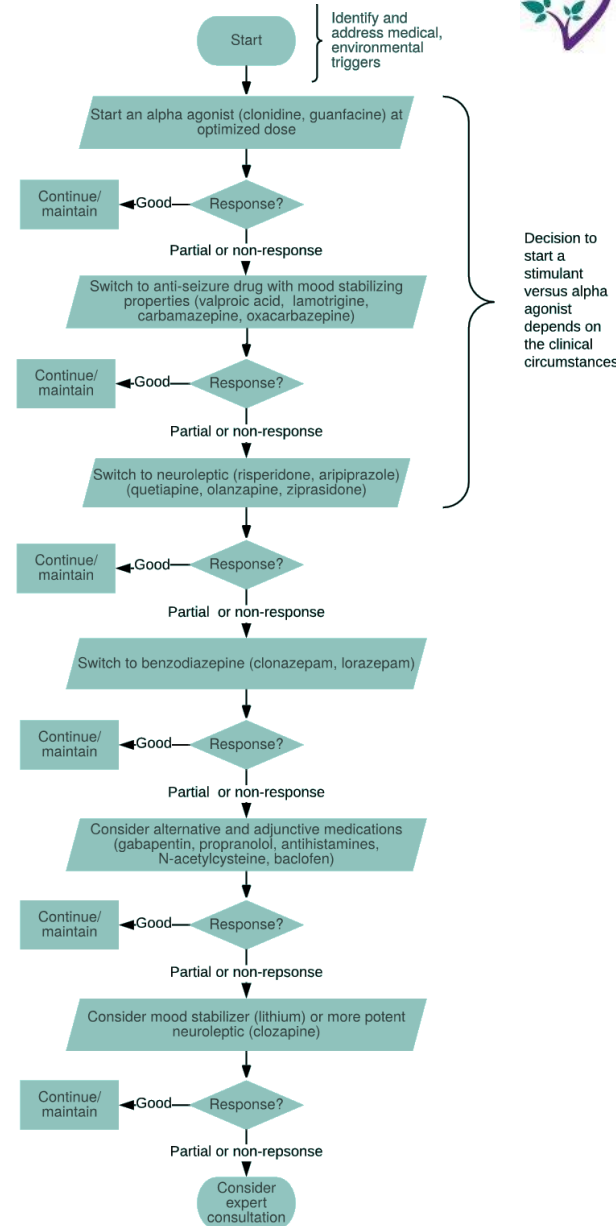
- **Second-generation neuroleptics (such as risperidone)** may be an option for CdLS
- Other second-generation neuroleptics to consider include aripiprazole, quetiapine, and olanzapine
- In some individuals, a mood stabilizer (valproate, lithium) may be beneficial, though the data is limited

Aggression/Self-Injury Medications

- **CAVEAT:** Neuroleptics are potent medications, with potential for serious side effects, such as **metabolic syndrome** and **weight gain**



General Approach to Treatment of Aggression in CdLS



Conclusions

- There is a spectrum of neurodevelopmental disorders associated with CdLS
 - Intellectual Disability
 - Autism
 - Language Disorder
 - Cerebral Palsy
- Identifying the target behavioral symptom(s) can help tailor medication management:
 - Hyperactivity
 - Anxiety/Depression
 - Aggression/Self-Injury
- There is a great need to study what medications work and don't work for individuals with CdLS

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THANK YOU