

Symptoms of Dup15q Syndrome - Clinician Global Impression

(Dup15q-CGI)

Rating guidance

When completing the **Dup15q-CGI-Severity** please rate your **overall impression** for each of your patient's Dup15q symptoms **during the past week (7 days)**, considering the following aspects for each symptom: severity, frequency, and impact on ability to undertake day-to-day activities. Base your rating on your total clinical experience with the Dup15q population (consider maternal duplications only), and your interactions and conversations with the patient and/or their caregiver(s) during this visit.

When rating, please consider the whole spectrum of Dup15q severity levels from very mild or mild (e.g., mild developmental phenotype, mild cognitive impairment, some expressive language) to severe or very severe (e.g., intractable epilepsy, nonverbal, severe intellectual disability, severe motor impairment). Please assume that "moderate" reflects the average severity in all Dup15q patients of a given age for that symptom. Hence, any patients who are less severe than average Dup15q patients of their age should be rated in the "mild" and "very mild" categories, even though their impairment may be significant in the context of typical development.

If the patient is consistent with peers without any development issues of the same age for a particular symptom, then rate "none" on that item. For example, if a 2-year-old Dup15q patient has the same difficulty with fine motor skills as expected for a 2-year-old without any developmental issues, then rate that patient as "none" for fine motor skills impairment.

When completing the **Dup15q-CGI-Change**, please rate your **overall impression of change** for each of your patient's Dup15q symptoms **during the past week (7 days) relative to their baseline severity**, considering the following aspects of each symptom: severity, frequency, and impact on day-to-day activities.

The information provided below for each symptom are examples but are not an exhaustive list.

1. Seizures

Please think about the different types of seizures that can occur (including non-epileptic myoclonus) and the different aspects of seizure severity given as examples below when answering question 1:

- Frequency of seizures;
- Duration of seizures;
- Ability to control seizures using medications;
- Type of medications used to control seizures (can include both chronic therapies and emergency/rescue medications);
- Hospitalizations due to seizures;
- Ability to perform everyday activities uninterrupted by seizures;
- Impact on everyday activities due to side effects from medications;
- Level of alterations in the home setting to accommodate seizures.

Rate “none” if they have had no seizures, **and** their ability to perform everyday activities was not affected by adaptations due to the risk of seizures or treatments associated with seizures.

2. Expressive communication difficulties

People with Dup15q may communicate in many different ways. To make this rating, please consider the totality of their expressive communication approaches, including the examples listed below when answering question 2:

- Eye movements;
- Pointing or gesturing;
- Sign language – either the individual’s own or international standard sign language;
- Speaking whole words;
- Verbal approximations – for example, grunting, babbling, or non-verbal resonating;
- Printed picture boards or other technology devices;
- Other augmented and alternative communication approaches, if used.

Please consider the extent to which words/gestures/sounds are used consistently and used to convey their usual meaning.

Rate “none” if their expressive communication ability is consistent with expectations for a typically developing child of their age.

3. Fine motor skills impairment

Please think about the fine motor skill examples listed below when answering question 3:

- Ability to hold or manipulate items with their hands – for example, holding a block or a crayon, zipping up a zipper, or picking up a small object;
- Difficulty with overall coordination impairment of hand movements – for example, due to tremors or ataxia.

Rate “none” if their fine motor skills are consistent with expectations for a typically developing child of their age.

4. Gross motor skills impairment

Please think about the gross motor skill examples listed below when answering question 4:

- Consider walking difficulties due to:
 - Hypotonia;
 - Incoordination;
 - Loss of balance, trips, or falls.
- Need for mobility aids and assistance with walking;
- For children who are not at an age where walking would be expected, please consider movement that would be expected for their age such as:
 - Coordinating limb movements;
 - Sitting up;
 - Crawling;
 - Standing.

Rate “none” if their gross motor skills are consistent with expectations for a typically developing child of their age.

5. Cognitive/intellectual impairment

Please think about both deficits due to developmental delay, as well as specific cognitive deficits that might be present such as the examples listed below when answering question 5:

- Maintaining attention on a task;
- Learning;
- Memory;
- Executive function;
- Problem solving;
- Receptive language;
- Ability to follow directions/instructions.

Rate “none” if their cognitive/intellectual abilities are consistent with expectations for a typically developing child of their age.

6. Impairment in activities of daily living/self-care

Please think about the daily living and self-care examples listed below when answering question 6:

- Feeding/eating;
- Using the toilet;
- Dressing/undressing;
- Washing/bathing;
- Brushing teeth.

Rate “none” if their abilities are consistent with expectations for a typically developing child of their age.

7. Impairment in social interaction

Please think about the social interaction examples listed below when answering question 7:

- Poor nonverbal communication – for example lack of eye contact, gestures, facial expressions (e.g. social smiling);
- Shared enjoyment in interaction and/or play – for example showing and directing attention to favorite objects or toys, offering to share;
- Lack of interest in interacting with others or reacting to / initiating social bids;
- Peer relationships – depending on age, interest in and responding to other children, engage in reciprocal or pretend play, friendships

Rate “none” if their social interaction is consistent with expectations for a typically developing child of their age.

8. Maladaptive behaviors

Please think about the maladaptive behavior examples listed below when answering question 8:

- Self-harming or aggressive outbursts – for example, hitting, biting, pushing, hair pulling, grabbing, pinching, kicking, shouting;
- Hyperactive, excitable, or impulsive behavior;
- Putting objects in their mouth;
- Refusing to participate in activities – for example, school, social, or domestic activities.

Rate “none” if behaviors are normal for a typically developing child of their age, for example, occasional shouting and grabbing for young children.

9. Sleep problems

Please think about the difficulties with sleep and quality of sleep examples listed below when answering question 9:

- Difficulties falling asleep;
- Disturbing nighttime awakenings;
- Early morning awakenings;
- Short total sleep time;
- Patterns of sleep problems;
- Interruption in ability to perform everyday activities due to sleepiness;
- **Do not** consider obstructive sleep apnea and seizures during sleep.

Rate “none” if their sleep is consistent with expectations for a typically developing child of their age.

10. Overall severity of Dup15q syndrome

Please think about the following when rating the overall severity of the patient’s Dup15q symptoms: the totality of the patient’s condition **during the past week (7 days)**, your answers to questions 1–9, and your total clinical experience with this population.

Symptoms of Dup15q Syndrome - Clinician Global Impression of Severity Questionnaire

Instructions for questions 1–10

Rate your **overall impression** of the severity of your patients’ Dup15q symptoms **during the past week (7 days)**.

- Base your rating on your total clinical experience with the Dup15q population and your interactions and conversations with the patient and/or their caregiver(s) during this visit;
- When making your rating consider **the severity of the symptom, the frequency with which it occurs, and the impact on functioning**;
- As a general guide, consider assigning a “moderate” severity rating when the symptom is average for a person with Dup15q of that age;
- An area for notes is included for you to document your decision-making when choosing a given severity level;
- This measure should be completed in conjunction with the rating guidance.

	None (1)	Very mild (2)	Mild (3)	Moderate (4)	Severe (5)	Very severe (6)	Notes to explain your rating
1. Seizures	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
2. Expressive communication difficulties	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
3. Fine motor skills impairment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
4. Gross motor skills impairment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
5. Cognitive/intellectual impairment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
6. Impairment in activities of daily living/self-care	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
7. Impairment in social interaction	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
8. Maladaptive behaviors	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
9. Sleep problems	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
10. Overall severity of Dup15q symptoms		<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

Symptoms of Dup15q Syndrome - Clinician Global Impression of Change Questionnaire

Instructions for questions 1–10

Rate your **overall impression** of change of your patients' Dup15q symptoms **during the past week** relative to their baseline severity.

- Base your rating on your total clinical experience with the Dup15q population and your interactions and conversations with the patient and/or their caregiver(s) during this visit as well as any relevant information such as medical records;
- When making your rating consider **the severity of the symptom, the frequency with which it occurs, and the impact on functioning**;
- All changes should be indicated, irrespective of the possible cause of the change;
- As a general guide, rate a patient's level of change in relation to what is typical for Dup15q patients of that age for that symptom;
- An area for notes is included for you to document your decision-making when choosing a given change level;
- This measure should be completed in conjunction with the rating guidance.

	Very much improved (1)	Much improved (2)	Minimally improved (3)	No change (4)	Minimally worse (5)	Much worse (6)	Very much worse (7)	Notes to explain your rating
1. Seizures	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
2. Expressive communication difficulties	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
3. Fine motor skills impairment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
4. Gross motor skills impairment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
5. Cognitive/intellectual impairment	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
6. Impairment in activities of daily living/self-care	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
7. Impairment in social interaction	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
8. Maladaptive behaviors	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	
9. Sleep problems	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

10. Overall change in Dup15q symptoms

A) Considering the responses given for the above questions, how would you rate any change in the overall symptoms of the patient's Dup15q and the impact on the patient's functional ability?

Very much improved (1)	Much improved (2)	Minimally improved (3)	No change (4)	Minimally worse (5)	Much worse (6)	Very much worse (7)	Notes to explain your rating
<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	<input type="checkbox"/>	

B) Would you consider this change in overall Dup15q symptoms to be meaningful?

<input type="checkbox"/>	Yes , there has been a meaningful improvement or a meaningful worsening in Dup15q symptom severity
<input type="checkbox"/>	No , there has not been a meaningful improvement or a meaningful worsening in their Dup15q symptom severity
<input type="checkbox"/>	Not applicable – No change in their overall Dup15q symptom severity
Notes to explain your rating	