

Appendix D. Baseline Assessment: Indications for the Primary Outcome

Each customized DSC2U *Caregiver Checklist* and *PCP Plan* was the amalgamation of hundreds of rules acting on the caregiver's unique set of responses. The rules that supported the Indications for each of our primary outcomes is detailed below in plain-language.

## **Appendix D: Baseline Assessment: Indications for the Primary Outcomes**

### **Primary outcome: Audiology screening**

An **audiogram** was recommended when participants reported the person with Down syndrome:

- was under 21 and had not had a formal hearing exam in the past 12 months.
- OR was under 21 and the participant was unsure whether they had a formal hearing exam in the past 12 months.
- OR was 21 or older and had not had a formal hearing exam in the past 2 years.
- OR was 21 or older and the participant was unsure whether they had a formal hearing exam in the past 2 years.

### **Primary outcome: Vision screening**

An **eye exam** was recommended when participants reported the person with Down syndrome:

- was under 5 and had not seen an ophthalmologist for a formal eye exam in the past 12 months.
- OR was under 5 and the participant was unsure whether they had seen an ophthalmologist for a formal eye exam in the past 12 months.
- OR was between the ages of 5 and 12 (inclusive) and had not seen an ophthalmologist for a formal eye exam in the past 2 years.
- OR was between the ages of 5 and 12 (inclusive) and the participant was unsure whether they had seen an ophthalmologist for a formal eye exam in the past 2 years.
- OR was between the ages of 13 and 20 (inclusive) and had not seen an ophthalmologist for a formal eye exam in the past 3 years.
- OR was between the ages of 13 and 20 (inclusive) and the participant was unsure whether they had seen an ophthalmologist for a formal eye exam in the past 3 years.
- OR was 21 or older and had not seen an ophthalmologist for a formal eye exam in the past 2 years.
- OR was 21 or older and the participant was unsure whether they had seen an ophthalmologist for a formal eye exam in the past 2 years.

### **Primary outcome: Screening/evaluation of celiac disease**

**Celiac screening** was recommended when participants reported the person with Down syndrome:

- had never had blood work done for celiac disease and showed symptoms of celiac disease (as defined below).
- OR showed symptoms of celiac disease (as defined below) and the participant was unsure whether they had ever had blood work done for celiac disease.
- OR had blood work done for celiac disease but not within the last 12 months, did not have a prior diagnosis of celiac disease, and showed symptoms of celiac disease (as defined below).
- OR had blood work done for celiac disease but not within the last 12 months, had a prior diagnosis of celiac disease, was currently on a gluten-free diet, and showed symptoms of celiac disease (as defined below).

The person with Down syndrome was considered to show **symptoms of celiac disease** when participants reported that they:

- experienced at least one of the following symptoms within the last month:
  - unexplained difficulty gaining weight
  - constipation that is hard to treat
  - frequent diarrhea
  - frequent vomiting
  - Nausea
  - bulky or foul-smelling stools
  - new accidents with stool
  - passing excessive gas
  - bloating
- OR experienced at least two of the following symptoms within the last 6 months:
  - overactive, restless, unable to sit still
  - inattention
  - impulsive, acts without thinking
  - easily distracted
  - uncooperative, disobeys
  - temper tantrums or outburst or meltdowns
  - throws or breaks objects
  - hits self
  - bites self
  - hurts herself on purpose (e.g. hitting head, biting hands)
  - bangs head
  - Irritability
  - kicks or hits others
  - mood changes rapidly for no reason
  - Aggression
  - property destruction (e.g. Breaks things, hits walls, throws things)
  - throws or breaks objects
  - cries easily for no reason
  - loss of previously learned skills

**Primary outcome: Screening/evaluation of thyroid dysfunction**

**Thyroid testing** was recommended when participants reported the person with Down syndrome:

- had not had thyroid function tests done within the last year.
- OR was unsure whether they had thyroid function tests done within the last year.
- OR had thyroid tests done within the last year, did not have a prior diagnosis of Hashimoto's thyroiditis or hypothyroidism, and showed symptoms of hypothyroidism (as defined below).
- OR had thyroid function tests done within the last year, had a prior diagnosis of Hashimoto's thyroiditis for which they were taking medication, and showed symptoms of hypothyroidism (as defined below).
- OR had thyroid function tests done within the last year, had a prior diagnosis of hypothyroidism for which they were taking medication, and showed symptoms of hypothyroidism (as defined below).
- OR had thyroid tests done within the last year, did not have a prior diagnosis of Graves disease, Hashimoto's thyroiditis, hypothyroidism, or hyperthyroidism, and showed symptoms of hyperthyroidism (as defined below).

- OR had thyroid tests done within the last year, did not have a prior diagnosis of Hashimoto's thyroiditis or hypothyroidism, had a prior diagnosis of Graves disease for which they were taking medication, and showed symptoms of hyperthyroidism (as defined below).
- OR had thyroid tests done within the last year, did not have a prior diagnosis of Hashimoto's thyroiditis or hypothyroidism, had a prior diagnosis of hyperthyroidism for which they were taking medication, and showed symptoms of hyperthyroidism (as defined below).

The person with Down syndrome was considered to show **symptoms of hypothyroidism** when participants reported that they:

- Experienced at least one of the following symptoms within the last month:
  - increasing fatigue
  - weight gain
  - constipation that is hard to treat
  - dry skin
  - hair loss
  - feeling too cold
- OR when they showed symptoms of depression (as defined below).

The person with Down syndrome was considered to show **symptoms of hyperthyroidism** when participants reported that they:

- Experienced at least one of the following symptoms within the last month:
  - undesired weight loss
  - frequent diarrhea
  - irregular periods
  - feeling too hot
- OR when they showed symptoms of anxiety (as defined below).
- OR when they showed symptoms of obsessive compulsive disorder (as defined below).
- OR when they showed symptoms of attention deficit hyperactivity disorder (as defined below).

### **Primary outcome: Screening/evaluation of sleep apnea**

A **sleep study** was recommended when participants reported the person with Down syndrome:

- was 4 or older, had never had a sleep study performed for obstructive sleep apnea, and did not show symptoms of sleep apnea (as defined below), depression (as defined above), or anxiety (as defined above).
- OR was 4 or older, did not show symptoms of sleep apnea (as defined below), depression (as defined above), or anxiety (as defined above), and the participant was unsure whether they had ever had a sleep study performed for obstructive sleep apnea.
- OR had never had a sleep study performed for obstructive sleep apnea and showed symptoms of sleep apnea (as defined below), depression (as defined above), or anxiety (as defined above).
- OR showed symptoms of sleep apnea (as defined below), depression (as defined above), or anxiety (as defined above), and the participant was uncertain whether they had ever had a sleep study performed for obstructive sleep apnea.
- OR has had a sleep study performed for obstructive sleep apnea but not within the last 12 months, does not have a prior diagnosis of obstructive sleep apnea, and shows symptoms of sleep apnea (as defined below), depression (as defined above), or anxiety (as defined above).

The person with Down syndrome was considered to show **symptoms of sleep apnea** when participants reported that they:

- experienced at least one of the following symptoms within the last month:
  - snoring
  - gasping, snorting, choking during sleep
  - breathing with open mouth during sleep
  - sleeping on multiple pillows
  - not refreshed despite adequate sleep
  - falling asleep in car on short drive
  - falling asleep/napping at school
- OR exhibited symptoms of depression (as defined above).
- OR exhibited symptoms of anxiety (as defined above).

#### **Ancillary Definitions Utilized by One or More of the Primary Outcome Rule Sets**

The person with Down syndrome was considered to show **symptoms of depression** when participants reported that they:

- were over the age of 4 and in the last 6 months had experienced both feeling down, depressed, or hopeless *and* little interest or pleasure in doing things.
- OR were over the age of 8, in the last 6 months had experienced feeling down, depressed, or hopeless, and in the last 6 months had experienced at least one of the following symptoms:
  - cries easily for no reason
  - moves slowly
  - loss of previously learned skills
  - eating too much or eating too little
  - trouble falling or staying asleep
  - tense, anxious, worried
  - distressed about being alone
- OR were over the age of 8, in the last 6 months had experienced little interest or pleasure in doing things, and in the last 6 months had experienced at least one of the following symptoms:
  - cries easily for no reason
  - moves slowly
  - loss of previously learned skills
  - eating too much or eating too little
  - trouble falling or staying asleep
  - tense, anxious, worried
  - distressed about being alone

The person with Down syndrome was considered to show **symptoms of anxiety** when participants reported that they:

- were 8 or older and had experienced at least one of the following symptoms in the last 6 months:
  - irritability
  - fears particular situations
  - chews body parts
  - general agitation

The person with Down syndrome was considered to show **symptoms of obsessive compulsive disorder** when participants reported that they:

- were 6 or older and in the last 6 months had experienced ritualistic compulsive behaviors (e.g. checking things/cleaning/grooming), which they found distressing, or which interfered with daily activities.
- OR were 6 or older and in the last 6 months had experienced getting obsessed with idea or activity, which they found distressing or which interfered with daily activities.

The person with Down syndrome was considered to show **symptoms of attention deficit hyperactivity disorder** when participants reported that they:

- exhibited at least two of the following symptoms within the last 6 months:
  - overactive, restless, unable to sit still
  - inattention
  - impulsive, acts without thinking
  - easily distracted
  - uncooperative, disobeys