Psychiatric Manifestations in Individuals with Trisomy 21 (T21)

7. Obsessive-Compulsive Disorder (OCD) and Related Disorders

Outline

1. Overview
2. Obsessive-Compulsive Disorder (OCD)
    - 2.1. Application of Diagnostic Criteria in Individuals with ID
    - 2.2. Repetitive Behaviors and Restricted Interests in Individuals with Trisomy 21: One Way of Managing Their World?
    - 2.3. Obsessional Slowness
    - 2.4. Epidemiology
    - 2.5. Diagnosis According to the Degree of ID (DM-ID-2)
    - 2.6. Treatment
3. Excoriation Disorder
4. Key points for clinical practice

1. Overview

In the DSM-5, obsessive-compulsive disorder (OCD) was removed from the chapter on anxiety disorders, and a new specific category, "Obsessive-Compulsive and Related Disorders," was created in order to highlight the common characteristics of these disorders, differentiating them from other anxiety disorders (APA, DSM-5, 2013).

- The related disorders include:
  a) New disorders and b) disorders that were classified in other categories in DSM-IV.

a) The new related disorders are:
   - Hoarding Disorder: this is now a diagnosis in its own right, whereas the DSM-IV listed hoarding as a possible symptom of obsessive-compulsive personality disorder or, when in a more severe form, OCD.
   - Excoriation (Skin-Picking) Disorder: Involves repeated and compulsive skin picking and/or scratching.

b) The disorders that have changed categories are:
   - Body Dysmorphic Disorder (BDD), previously classified in the DSM-IV under somatoform disorders.
   - Trichotillomania, the compulsive pulling out of hair or body hair, classified in the DSM-IV as an impulse control disorder, is now included in OCD and related disorders.

2. Obsessive-Compulsive Disorder (OCD)

2-1: Application of Diagnostic Criteria in Individuals with ID

- The diagnosis of OCD in individuals with ID must be based on the DSM-5 criteria (APA, DSM-5, 2013), in addition to the specifics outlined in DM-ID-2 (Fletcher et al., 2016):

1. **Obsessive-Compulsive Disorder (OCD) is characterized by the presence of obsessions, compulsions, or both. Obsessions and compulsions are frequently associated with ID.**

2. Limited expressive language skills make self-assessment and the use of scales or inventories challenging.

3. Sensory impairments may further restrict the range of obsessive-compulsive phenomena.

4. **The presence of anxiety is no longer required for diagnosis.**

5. Eliminating the anxiety criterion facilitates diagnosis in individuals with ID, as they may not previously have experienced their symptoms as anxiety-producing or may not have been able to identify or communicate anxiety symptoms to clinicians.

2. Obsessive-Compulsive Disorder (OCD)

2-1: Application of Diagnostic Criteria in Individuals with ID (continued)

5. In the general population, obsessions are experienced as intrusive or unwanted; this description may not apply to some people with ID, depending on their ability to identify and/or communicate this concept.

6. Compulsions that require abstract thinking (e.g. contamination or safety compulsions) may not be possible, and counting skills may be absent in people with ID.

7. Individuals with ID may be unaware of societal disapproval of their behavior. Because of this lack of awareness, they may not be inclined to reduce their behaviors the way neurotypical individuals are.

8. Aggression can be the presenting complaint in individuals with ID. A thorough history should be taken to determine if the trigger was, for example, an attempt by someone else to move an object or prevent a ritual, in which case the diagnosis of OCD should be considered.

2. Obsessive-Compulsive Disorder (OCD)

2-2: Repetitive Behaviors and Restricted Interests in Individuals with Trisomy 21: One Way of Managing Their World?

- Sheila Glen in her article *"Repetitive Behaviours and Restricted Interests in Individuals with Down Syndrome — One Way of Managing Their World?"* (2017) **suggests that many of the repetitive behaviors and restricted interests exhibited by individuals with T21 are adaptive and should not necessarily be considered pathological**.

- Around 2 years of age, typically developing children tend to become highly compulsive; they may be dependent on particular bedtime routines, expect things to be done “just right“, may have strong food preferences, and repetitive behavior. If thwarted, they are likely to react negatively.

- The ubiquity of these behaviors in early development suggests they have an important function.

- Routinized and compulsive-like behaviors are important for reducing anxiety, for example in bedtime routines. Piaget also emphasized the importance of repetition and "just right" behaviors in learning how to interact with the environment and developing classification skills. In other words, routines (and familiarity) for young children increase feelings of competence, reduce anxiety, and free up cognitive capacity, all of which foster the likelihood that they will explore and learn new ideas and skills.

2. Obsessive-Compulsive Disorder (OCD)

2-2: Repetitive Behaviors and Restricted Interests in Individuals with Trisomy 21: One Way of Managing Their World? (continued)

- Repetitive behaviors and restricted interests tend to decrease in typically developing children by the age of 7. However, some adolescents without psychiatric diagnoses still exhibit repetitive behaviors and restricted interests.

- The average mental age of adults with T21 is around 5-6 years, **so it is likely that the relatively high level of repetitive behaviors and restricted interests is still adaptive at this developmental age**.

- There is also evidence that repetitive behaviors and restricted interests are not unique to individuals with Trisomy 21 but are also found in various other conditions associated with intellectual disability.

2. Obsessive-Compulsive Disorder (OCD)

2-3: Obsessional Slowness

- Obsessional slowness was originally described by S. Rachman in 1974. **These patients had OCD and spent hours performing daily routines such as washing, dressing, and eating.** Although some ritualistic behaviors were observed, **slowness was the most significant problem for these patients**.
- In 1994, R. J. Pary described a small number of individuals with Trisomy 21 who had obsessional slowness. They described slowness as a primary feature, noting that it was rarely associated with manifest anxiety. The author cited regional cerebral blood flow data and other imaging studies showing hypometabolism in the orbital cortex in individuals with Trisomy 21 compared to the hypermetabolism found in individuals with OCD and obsessional slowness.
- In a more recent study, Charlot et al. (2002) described 11 cases of individuals with Trisomy 21 and obsessional slowness. Many of these patients had periods of freezing and difficulty initiating movements. The patients showed low anxiety levels and perfectionist tendencies, similar to the findings of other studies on obsessional slowness.
- Some authors argue that obsessional slowness is a severe form of OCD, while others believe that it should not be considered a distinct disorder and that the diagnosis of severe OCD or a form of catatonia should be considered (*Ganos et al., 2015*).
- In the "Psychiatry" chapter of this module, we addressed these presentations in “5. Regression and Catatonia."

2. Obsessive-Compulsive Disorder (OCD)

2-4: Epidemiology

- Prevalence estimates for OCD in individuals with Trisomy 21 **range from 0.8%** *(Myers & Pueschel, 1991*) **to 4.5%** *(Prasher, 1995*), and **even 7.3%** in the recent study by Rivelli et al., 2022.

- Rivelli et al, 2022, also highlight the greater prevalence of OCD in individuals with Trisomy 21 compared to the general population (7.3% vs. 0.4%), with a significant OR of 20.15 (16.43, 24.71), i.e. $p<0.0001$.

- Ordering and cleaning are the most common behaviors presented (Prasher & Day, 1995).

2. Obsessive-Compulsive Disorder (OCD)

2-5: Diagnosis According to the Degree of ID (DM-ID-2)

DSM-5	Application of criteria in individuals with mild or moderate ID	Application of criteria in individuals with severe ID
A. Presence of obsessions, compulsions, or both.	A. No adaptation.	A. No adaptation.
Obsessions are defined by two criteria: 1. Recurrent and persistent thoughts, urges or images that are experienced, at some time during the disturbance, as intrusive, unwanted, and that in most individuals cause marked anxiety or distress. 2. The individual attempts to ignore or suppress such thoughts, urges, or images, or to neutralize them with some thought or action (i.e., by performing a compulsion)	Obsessions are defined by two criteria: 1. No adaptation. Note: Recurrent and persistent thoughts, urges, or images may not be experienced as intrusive and unwanted depending on the cognitive functioning of the individual. 2. No adaptation. Note: The individual may or may not (due to cognitive deficits) attempt to ignore or suppress such thoughts, urges, or images, or to neutralize them with some other thought or action.	Obsessions are defined by two criteria: 1. No adaptation. Note: Recurrent and persistent thoughts, urges, or images may not be experienced as intrusive and unwanted depending on the cognitive functioning of the individual. 2. No adaptation. Note: The individual may or may not (due to cognitive deficits) attempt to ignore or suppress such thoughts, urges, or images, or to neutralize them with some other thought or action. The individual may be unable to report wanting to ignore, suppress, or neutralize the obsessions.

Table adapted from DM-ID2: Fletcher RJ, Barnhill J, Cooper F, 2016.

2. Obsessive-Compulsive Disorder (OCD)

2-5: Diagnosis According to the Degree of ID (DM-ID-2)

DSM-5	Application of criteria in individuals with mild or moderate ID	Application of criteria in individuals with severe ID
Compulsions are defined by two criteria: 1.Repetitive behaviors (e.g., hand washing, ordering, checking) or mental acts (e.g., praying, counting, repeating words silently) that the individual feels driven to perform in response to an obsession or according to rules that must be applied rigidly. 2. The behaviors or mental acts are aimed at preventing or reducing anxiety or distress, or preventing some dreaded event or situation; however, these behaviors or mental acts are not connected in a realistic way with what they are designed to neutralize or prevent, or are clearly excessive. Note: Young children may not be able to articulate the aims of these behaviors or mental acts.	Compulsions are defined by two criteria: 1.No adaptation. Note: Repetitive behaviors or mental acts may be difficult to elicit due to cognitive deficits and limited expressive language skills. Consider behaviors such as ordering, telling, asking or repetitive physical acts (e.g., rubbing) as compulsions. 2. No adaptation. Note: The function of the compulsive behavior may not be ascertainable due to cognitive deficits and limited expressive language skills, recognition of excessiveness or intent of the behaviors may not be present.	Compulsions are defined by two criteria: 1.No adaptation. Note: Absence of compulsions that require abstract thinking does not rule out OCD; observe individuals for compulsions requiring simple thinking, such as fixed sequences or arrangements, excessive ordering, and filling/emptying. 2. No adaptation. Note: The function of the compulsive behavior may not be ascertainable due to cognitive deficits and limited expressive language skills. The criteria regarding intent of the behavior do not apply to children and do not apply to individuals with severe/profound intellectual disabilities.

Table adapted from DM-ID2: Fletcher RJ, Barnhill J, Cooper F, 2016.

2. Obsessive-Compulsive Disorder (OCD)

2-5: Diagnosis According to the Degree of ID (DM-ID-2)

DSM-5	Application of criteria in individuals with mild to moderate ID	Application of criteria in individuals with severe ID
B. The obsessions or compulsions are time-consuming (e.g., take more than 1 hour per day) or cause clinically significant distress or impairment in social, occupational, or other important areas of functioning.	B. No adaptation. Note: Distress may not occur and/or may not be ascertainable. Intense preoccupation may be observed or drive to perform the compulsion may be observed. Challenging behavior, especially aggression, and self-injurious behavior, may occur if the individual is prevented from completing the compulsion.	B. No adaptation. Note: Distress may not occur and/or may not be ascertainable. Intense preoccupation, strong urges to engage in compulsive activity may be observed. Aggression, especially directed towards caregivers who impede the completion of the compulsion, may be seen.
C. The obsessive-compulsive symptoms are not attributable to the physiological effects of a substance (e.g., a drug of abuse or medication) or another medical condition.	C. No adaptation	C. No adaptation

Table adapted from DM-ID2: Fletcher RJ, Barnhill J, Cooper F, 2016.

2. Obsessive-Compulsive Disorder (OCD)

2-5: Diagnosis According to the Degree of ID (DM-ID-2)

DSM-5	Application of criteria in individuals with mild or moderate ID	Application of criteria in individuals with severe ID
D. The disturbance is not better explained by the symptoms of another mental disorder (e.g., excessive worries, as in generalized anxiety disorder; preoccupation with appearance, as in body dysmorphic disorder; difficulty discarding or parting with possession, as in hoarding disorder; hair pulling, as in trichotillomania [hair-pulling disorder]; skin picking, as in excoriation [skin-picking] disorder); stereotypies, as in stereotypic movement disorder; ritualized eating behavior, as in eating disorders; preoccupation with substances or gambling, as in substance-related and addictive disorders; sexual urges or fantasies, as in paraphilic disorders; impulses, as in disruptive, impulse-control, and conduct disorders; guilty ruminations, as in major depressive disorder; thought insertion or delusional preoccupations, as in schizophrenia spectrum and other psychotic disorders; or repetitive patterns of behavior, as in autism spectrum disorder).	D. No adaptation	D. No adaptation

Table adapted from DM-ID2: Fletcher RJ, Barnhill J, Cooper F, 2016.

2. Obsessive-Compulsive Disorder (OCD)

2-5: Diagnosis according to DM-ID2

DSM-5	Application of criteria in individuals with mild or moderate ID	Application of criteria in individuals with severe ID
Specify if: - With good or fair insight - With poor insight - With absent insight/delusional beliefs Specify if: - Tic-related: The individual has a current or past history of a tic disorder.	The various specifiers assessing the patient's awareness of beliefs related to the disorder (e.g., good, fair, poor, absent/delusional beliefs) should be applied in the context of the individual's cognitive and developmental functioning.	

Table adapted from DM-ID2: Fletcher RJ, Barnhill J, Cooper F, 2016.

2. Obsessive-Compulsive Disorder (OCD)

2-6: Treatment

- Current knowledge about treating OCD in the general population is summarized in the article by Swierkosz-Lenart et al. (2023): "Therapies for obsessive-compulsive disorder: Current state of the art and perspectives for approaching treatment-resistant patients".

- There are few studies on the treatment of OCD in individuals with Trisomy 21, but **overall, the recommendations do not differ from those for the general population**. These studies include:
  - O'Dwyer et al., 1992, reported two cases of OCD, one of whom showed a partial response to fluvoxamine 200 mg, and another patient who did not respond to successive therapeutic trials with clomipramine 150 mg and fluoxetine 40 mg.
  - Raitasuo et al., 1998, reported the case of a T21 patient with anorexia nervosa, major depression, and OCD who showed a decrease in OCD symptoms when treated with citalopram 40 mg.
  - Sutor et al., 2006, reported a case series of four adults with T21 and OCD treated with SSRIs alone or in combination with risperidone. All patients showed significant improvement in OCD symptoms, and none experienced extrapyramidal symptoms or tardive dyskinesia.

3. Excoriation (Skin Picking) Disorder

Excoriation disorder, also known as dermatillomania or skin picking disorder, is an OCD-related disorder involving repetitive self-injurious behavior such as pulling, scratching, picking, or digging into one's own skin with fingers or tools such as paper clips or tweezers in the absence of a dermatological condition.

- In the general population, Hayes et al., 2009, found that up to 63% of participants had a form of excoriation disorder without clinical significance, while 5.4% had forms causing clinically significant distress.

- Direct associations were found between excoriation disorder and depressive, anxious, and obsessive-compulsive symptoms, supporting the emotional regulation model of excoriation disorder.

- **Studies on the prevalence of excoriation disorder in people with intellectual disabilities are limited. However, higher prevalence has been found in specific genetic syndromes, including Prader-Willi, fragile X, and Smith-Magenis.**

- Although there are no specific studies on excoriation disorder in individuals with T21, clinicians regularly report it, indicating a need for further investigation.

- Considering the reduced emotional regulation abilities of individuals with intellectual disabilities, skin picking may be a strategy for coping with stress.

4. Key Points for Clinical Practice

- Obsessive-Compulsive Disorder (OCD) is characterized by the presence of obsessions, compulsions, or both. Obsessions and compulsions are frequently associated with ID.
- Prevalence estimates for OCD in individuals with T21 range from 0.8% to 7.3%.
- When applying diagnostic criteria, it is important to consider the characteristics of ID as well as the specific phenotypic characteristics of individuals with T21.
- Compulsions requiring abstract thinking or counting, which may be limited or absent in those with ID, may not be possible.
- Individuals with ID may be unaware of societal disapproval of their behavior. Because of this lack of awareness, they may not be inclined to reduce their behaviors.
- Aggression can manifest as an expression of OCD in people with ID, triggered if someone else attempts to move an object or prevent a ritual, for example.
- In Trisomy 21, a great many repetitive behaviors and restricted interests remain adaptive and part of the behavioral phenotype, serving as a "way of managing their world.”
- Obsessional slowness described in adolescents and young adults with T21 should not be considered a distinct disorder. Instead, the diagnosis of a severe form of OCD or a form of catatonia should be considered.
- There are few studies on the treatment of OCD in individuals in T21. The cases reported in the literature have been managed using the standard treatment approaches typically recommended for OCD in the general population: antidepressant treatment, occasionally supplemented by a neuroleptic.