ID AND PSYCHIATRIC DISORDERS IN THE NEW CLASSIFICATION SYSTEMS

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POLYNOMIC APPROACH

- IDD/ID is a highly complex construct that combines a medical diagnosis (a meta-syndrome) and its personal and social impact in human functioning.

- This complexity indicates that full understanding of the diverse facets requires at least two 'semantically-similar' names which can be used in different contexts:
  - "Intellectual Developmental Disorder" (the medical syndrome)
  - "Intellectual Disability" (the rights, normative, social, and personal aspects of this syndrome).
ID: DISABILITY OR DISORDER?
MR name change: DSM-5 Position

- Intellectual disability (intellectual developmental disorder) as a DSM-5 diagnostic term replaces “mental retardation” used in previous editions of the manuals.

- The parenthetical name “(intellectual developmental disorder)” is included in the text to reflect deficits in cognitive capacity beginning in the developmental period.

- Together, these revisions bring DSM into alignment with terminology used by the WHO International Classification of Diseases, other professional disciplines and organizations.

AAAID Proposed Recommendations for ICD-11 and the Condition Previously Known as Mental Retardation

Vaw J. Truel, Bath Lockstone, and Manjere Nygren

Abstract

The World Health Organization (WHO) is in the process of revising a number of medical and psychiatric classifications, including the diagnosis of intellectual disability as part of the International Classification of Diseases (ICD-11). The American Association on Intellectual and Developmental Disabilities (AAAID) convened a small group of distinguished professionals in the field of intellectual and developmental disabilities to advance revisions to the ICD-11 proposal regarding revisions of intellectual and developmental disabilities. The AAAID Technical Committee, under the leadership of John O. Zinkin, comprised professionals in the field of intellectual and developmental disabilities. The recommendations of the AAAID group were presented to the WHO.

Recommendations

1. Terminology: We recommend that the category name “Disorders of Intellectual Disability” (Disorders of ID) be used rather than “Intellectual Developmental Disorders.”
ID in ICD-11-beta

- 06 Mental and behavioural disorders
  - Neurodevelopmental disorders
    - Disorders of intellectual Development (DID)
      6A00 DID, mild
      6A01 DID, moderate
      6A02 DID, severe
      6A03 DID, profound
      6A04 DID, provisional
      6A0Y Other specified DID
      6A0Z DID, unspecified
  - KA00 Conditions with DID as a relevant clinical feature

WPA-SPID POSITION STATEMENT

- ID is a health condition
- ID is a meta-syndrome characterised by an impairment in cognitive functioning prior to the acquisition of skills through learning
- the intensity of the deficit is such to interfere in a significant way with individual normal functioning as expressed in limitations in activities and restriction in participation (disabilities)

WHO, ICD-11 Beta Draft, 2014

IDD DEFINITION

- A group of developmental conditions characterized by a significant impairment of cognitive functions, which are associated with limitations of learning, adaptive behavior and skills.
- IDD is a life span condition requiring consideration of developmental stages and life transitions.
- Most individuals with IDD continue to acquire skills and competencies, especially with optimal care, training, education and opportunities for learning.
- However IDD is a vulnerable group associated with a higher rate of mental and physical disorders and related unmet care needs as well as an increased risk of abuse and neglect.

Is IQ reduction an useful diagnostic criterion for IDD?

Moving beyond intelligence in the revision of ICD-10: specific cognitive functions in intellectual developmental disorders

A lower level of intelligence, as measured by IQ, has implications from the central nervous system to neural networks. IQ is influenced by a range of factors, including genetic and environmental influences, and by the brain's ability to process information. The current classification system, however, relies solely on IQ to define intellectual disability (ID) in the DSM-5. Similarly, ID is defined by the intellectual functions in traditional terms, without considering the range of cognitive functions that may be involved in the development of ID. This is a limitation, as it fails to capture the diversity of cognitive functions that may be involved in the development of ID.

The revision of the DSM-5 has led to the development of a new classification system that is more focused on specific cognitive functions. This approach is based on a new concept of intellectual disability that takes into account the range of cognitive functions that may be involved in the development of ID. This approach is more comprehensive, as it takes into account the range of cognitive functions that may be involved in the development of ID.

IQ AND COGNITIVE PROFILE IN GS

<table>
<thead>
<tr>
<th>Syndrome</th>
<th>ID Severity</th>
<th>Cognitive Phenotype</th>
</tr>
</thead>
<tbody>
<tr>
<td>Down Syndrome</td>
<td>Mostly mild and moderate</td>
<td>Morphosyntax, short-term memory, explicit memory, long-term memory</td>
</tr>
<tr>
<td>Prader-Willi</td>
<td>Mild and moderate</td>
<td>Phonological loop, emotion modulation, attention switch</td>
</tr>
<tr>
<td>Sindrome di Williams</td>
<td>Mild and moderate</td>
<td>Emotional-relational modulation, concrete speech, visual-spatial construction, selective and sustained attention, memory, planning, inhibition</td>
</tr>
<tr>
<td>Fragile X Syndrome</td>
<td>Mostly mild and moderate</td>
<td>Sequential processing, short-term memory, motor control, coordination, verbal labelling and comprehension, visual-spatial elaboration, writing, calculation</td>
</tr>
</tbody>
</table>
Findings of recent studies of genetics, neuroimaging and neurophysiology identified more correlations with cognitive functions (such as perceptual organization deficit, poor working memory, lexical, visual-spatial and phonological processing) than with IQ scores.  

N = 8450 adults

- around 1/8 of the population has borderline intelligence (12.3% of the sample)
- this people present higher rate of:
  - neurotic disorders
  - substance abuse
  - personality disorders
  - social disability
  - psycho-pharmacological therapies, but not speech therapies
  - health service use, including emergency services


**IQ: A SECONDARY LEVEL**

- full-scale IQ is an outmoded concept and inadequate basis for diagnosing IDD/ID.

- At present, IQ cannot be removed from the criteria for the diagnosis of IDD/ID, but it should be placed at SECONDARY level to full neuropsychological assessment and cognitive profiling.

- This endorses the idea of a broad-based approach to diagnosing IDD/ID, using targeted cognitive and neuropsychological measures, plus developmental observation and assessment and confirmation after a follow-up period of:
  - functioning,
  - learning
  - and skills.

things found very exemplary about the WPA letter:

(a) the strong assertion (based on a Delphic analysis of many studies) is that a consensus has formed that full-scale IQ is an outmoded concept and inadequate basis for diagnosing ID

(b) the endorsement of the idea that a broad-based approach to diagnosing ID, using targeted cognitive and neuropsych measures, plus observation, should be used to understand the person

(c) emphasis on the brain-based science of ID, instead of a just a disability-based number

Stephen Greenspan, letter to the WPA-SPID Past President, March 2014

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IQ and ID/IDD: DSM-5 Position

- DSM-5 emphasizes the need to use both clinical assessment and standardized testing of intelligence when diagnosing ID, with the severity of impairment based on adaptive functioning rather than IQ test scores alone.
- By removing IQ test scores from the diagnostic criteria, but still including them in the text description of intellectual disability, DSM-5 ensures that they are not overemphasized as the defining factor of a person’s overall ability, without adequately considering functioning levels.
- IQ or similar standardized test scores should still be included in an individual’s assessment. In DSM-5, ID is considered to be approximately two standard deviations or more below the population, which equals an IQ score of about 70 or below.
- The assessment of intelligence across three domains (conceptual, social, and practical) will ensure that clinicians base their diagnosis on the impact of the deficit in general mental abilities on functioning needed for everyday life.
- when lower than 60, IQ validity is questionable

APA, Diagnostic and Statistical Manual for Mental Disorders - 5th edition, 2013
DEFINING COGNITIVE IMPAIRMENT IN ID/IDD

LEVEL OF SPECIFIC FUNCTIONING

<table>
<thead>
<tr>
<th>Basic cognitive functions (ICF b163 and b140)</th>
</tr>
</thead>
<tbody>
<tr>
<td>i.e. memory (short term, long term, recall, explicit, working, etc.) or attention (orienting, selective, sustained, switch, etc)</td>
</tr>
</tbody>
</table>

LEVEL OF COMBINED FUNCTIONING

<table>
<thead>
<tr>
<th>QI</th>
<th>- associations and synergies of cognitive functions</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>- executive functions (higher level cognitive functions - ICF b164)</td>
</tr>
</tbody>
</table>

Is the age limit of 18 an useful criterion for ID?

WPA-SPID Position

Although the specific age limit of 18 is clearly arbitrary, WPA-SPID members expressed general agreement on the importance to keep a distinction between a persistent process that begins at birth (or early in life) and a change occurring after a normal development.

DSM-5 Position

While ID does not have a specific age requirement, an individual’s symptoms must begin during the developmental period and are diagnosed based on the severity of deficits in adaptive functioning.
There is an emphasis on the brain-based science of IDD, instead of a just a disability-based number.

This is particularly relevant in Forensics where the careful analysis of all the available information should lead to a CLINICAL (medical) diagnosis which should never be based on a single number (either IQ and/or adaptative behaviour)

This is the standard approach used in the diagnosis of other complex meta-syndromes such as dementia, and unfortunately, IDD/ID has been an exception in the medical diagnosis to this date

At issue (in a sense) is life or death because the IQ bright line number of 70 is used in several states in the US to determine eligibility for the death penalty. Thus disenfranchising the IQ total score as determinative (as you seem to do) of an ID diagnosis without giving any consideration to adaptive functioning is the consideration for the US Supreme Court case to be heard next Monday March 3.
PREVALENCE RATE (%) 
ASD AND ID

- PDD in ID = 30-40%
- ID in autism = 25-80%
- risk of underestimating ASD in ID in favour of schizophrenia

1. Kraijer 1997 (N=718); Morgan et al. 2002 (N=571); La Malfa et al., 2004 (N=1st adults); Cooper et al., 2007
2. Hoekstra et al., 2009 BJP; Centers for Disease Control and prevention USA, 2006; Edelson, 2006; Rescorla & Szatmari, 2009; Roid et al. 2006; Noterdaeme e Wriedt, 2010; Byun and Smith, 1998
3. Patucka et al., 2009; Savage et al., 2007

INCREASE OF AUTISM AND ASDs PREVALENCE RATE

More Autism ... or Different Labeling?

Soaring numbers of children reported as autistic by school districts are often cited as proof that the incidence of the condition is rising (left). But changes in diagnostic criteria could also account for that pattern. One study found that as autism seemingly increased, the prevalence of learning disabilities and mental retardation dropped—which suggests that in some cases, one diagnosis substituted for another (right).

---The Edition

Autism - Newschaffer et al., 2007
ASD - Lazoff et al., 2010; Baron-Cohen et al., 2009
PREVALENCE RATE (%)  
ASD AND SCHIZOPHRENIA

- around 50% of people with autism also meets criteria for schizophrenia disorganised-type
- at least 1.5% of psychiatric outpatients don’t receive the right diagnosis of ASD; 26% of these is diagnosed with schizophrenia

1. Konstantareas and Hewitt, 2001  
2. Nylander and Gilberg, 2001

SCHIZOPHRENIA AND ID/IDD

- The majority of people with SCZ have cognitive impairments
- Subtle ID already in the premorbid phase
  - in first-onset SCZ, IQ reduction is about more than 1 SD
  - in children who will develop SCZ in adulthood IQ reduction is 0.5 SD

Most common cognitive dysfunctions:
- attention deficit, especially in the reaction speed to sensory stimuli;
- working memory deficit;
- deficit of short-term and long-term verbal memory;
- deficit in categorization of information
- perseverative errors

SEQUENTIAL COMORBIDITY

- in ID around 50% of ASDs has been previously diagnosed with schizophrenia\(^1\)
- 21% of people with schizophrenia receive a lifetime diagnosis of PDD-NOS\(^2\)
- history of low intellectual functioning in many schizophrenics\(^3\)

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2. Sporn et al., 2004; Towbin et al., 2005
3. Hedman et al., 2013

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![CNV (COPY NUMBER VARIATION)](image)
GENETICS OF NEURODEVELOPMENTAL DISORDERS

<table>
<thead>
<tr>
<th>Chromosome</th>
<th>Gene(s)</th>
<th>Function(s)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1q21.1</td>
<td>CHD1, PBKAB2</td>
<td>chromatin and AMP kinase regulation</td>
</tr>
<tr>
<td>16p11.2</td>
<td>SEZ6L2</td>
<td>expressive language and socialisation</td>
</tr>
<tr>
<td>16p13.1</td>
<td>NDE1, NTAN1</td>
<td>synaptic plasticity, memory</td>
</tr>
<tr>
<td>GRIN2</td>
<td>NMDA rec</td>
<td>working memory and perceptual binding</td>
</tr>
<tr>
<td>TCF4</td>
<td>TCF4</td>
<td>memory and attention (N150)</td>
</tr>
<tr>
<td>NRXN1</td>
<td>neurexine</td>
<td>synaptic functions</td>
</tr>
<tr>
<td>CNTNAP2</td>
<td>contactin-L2</td>
<td>cell adhesion and receptors</td>
</tr>
<tr>
<td>SHANK 3</td>
<td>ProSAP2</td>
<td>synapse and dendritic spine formation</td>
</tr>
<tr>
<td>UPF3B</td>
<td>UPF3B</td>
<td>neurons growth and differentiation</td>
</tr>
</tbody>
</table>

WPA-SPID proposed to include in a cluster of disorders sharing salient cognitive symptoms and similarities on risk factors, clinical factors, genetic phenotype, early onset, course, and co-morbidity.
06 Mental and behavioural disorders

- **Neurodevelopmental disorders**
  - Disorders of intellectual Development
    - 6A00 DID, mild
    - 6A01 DID, moderate
    - 6A02 DID, severe
    - 6A03 DID, profound
    - 6A04 DID, provisional
    - 6A0Y Other specified DID
    - 6A0Z DID, unspecified
  - KA00 Conditions with DID as a relevant clinical feature

NDD: DSM-5 Position

- NDD include conditions with onset in the developmental age, typically early, often occurring before school age and characterised by developmental deficits which determine impairment of the personal, social, school and work functioning

- deficits range from very specific difficulties in learning and executive functions to a wide impairment of social skills and intelligence

- NDD often co-occur, i.e people with ASD often have also ID/IDD and many children with ADHD often have also a specific learning disorder
DSM-5 METASTRUCTURE

NEURODEVELOPMENTAL DISORDERS

F70. Intellectual Disability (Intellectual Developmental Disorder)
F80. Communication Disorders
  F80.0 Language Disorder
  F80.0 Speech Sound Disorder
  F80.81 Childhood-Onset Fluency Disorder (Shuttering)
  F80.89 Social (Pragmatic) Communication Disorder
  F80.9 Unspecified Communication Disorder
F84. Autism Spectrum Disorder
F90. Attention-Deficit/Hyperactivity Disorder
  F90.2 Combined
  F90.0 Predominantly Inattentive
  F90.1 Predominantly Hyperactive/Impulsive
  F90.8 Other Specified Attention-Deficit/Hyperactivity Disorder
  F90.9 Unspecified Attention-Deficit/Hyperactivity Disorder
F81. Specific Learning Disorder
  F81.0 with impairment in Reading
  F81.81 with impairment in Written Expression
  F81.2 with impairment in Mathematics
F82. F96. F95 Motor Disorders
F88. F89 Other Neurodevelopmental Disorders

ICD-11 AND DIAGNOSIS OF MENTAL DISORDERS IN PEOPLE WITH DISORDERS OF INTELLECTUAL DEVELOPMENT (DID)

SUBMISSION TO THE WHO
ICD-11/DID WORKING GROUP
ON BEHALF OF THE FACULTY OF INTELLECTUAL DISABILITY
ROYAL COLLEGE OF PSYCHIATRISTS-UK

IN COLLABORATION WITH NATIONAL AND INTERNATIONAL EXPERTS AND STAKEHOLDERS
1. GENERAL COMMENTS ON THE ICD-11 WITHIN THE CONTEXT OF DID

Insertion of the following paragraph to the introductory chapter of the ICD-11:

“With regard to the application of ICD-11 criteria for mental disorders to individuals with limited cognitive functions and ability to communicate, efforts must be made to seek the presence of signs and symptoms using developmentally appropriate methods of investigation since chronological age and developmental level (mental age) are by definition discrepant in this group. Such methods should include the ascertainment of signs of disorder that are observable, or that are reported by others familiar with the person.

DM-ID (Diagnostic Manual – Intellectual Disability)

Table 5. Changes in Diagnosis from DSM-IV-TR to the Diagnostic Manual-Intellectual Disability (DM-ID)\textsuperscript{a,b,c,d}

<table>
<thead>
<tr>
<th>Description of Change</th>
<th>Anxiety Disorder</th>
<th>Mood Disorder</th>
<th>Psychotic Disorder</th>
<th>FDD</th>
</tr>
</thead>
<tbody>
<tr>
<td>Change from NOS to specific diagnosis in same broad category</td>
<td>15</td>
<td>42</td>
<td>9</td>
<td>8</td>
</tr>
<tr>
<td>Change from NOS to specific diagnosis in a different broad category</td>
<td>2</td>
<td>1</td>
<td>15</td>
<td>2</td>
</tr>
<tr>
<td>Change from one specific diagnosis to another in the same broad category</td>
<td>2</td>
<td>11</td>
<td>5</td>
<td>3</td>
</tr>
<tr>
<td>Change to a specific diagnosis in a different broad category</td>
<td>3</td>
<td>1</td>
<td>6</td>
<td>1</td>
</tr>
<tr>
<td>No. of new diagnoses by DM-ID</td>
<td>10</td>
<td>4</td>
<td>2</td>
<td>7</td>
</tr>
</tbody>
</table>

\textsuperscript{a}Records were included only if the clinician responded yes to the question.
\textsuperscript{b}Around 100 diagnoses per group.
**PSYCHIATRIC DISORDERS IN ID WITH AND WITHOUT AUTISM**

<table>
<thead>
<tr>
<th>Prevalence</th>
<th>Tool</th>
<th>with A</th>
<th>without A</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bradley &amp; Bolton, 2006</td>
<td>SAPPA</td>
<td>50</td>
<td>16.7</td>
</tr>
<tr>
<td>Bradley et al., 2004</td>
<td>DASH</td>
<td>&gt;50</td>
<td>25</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Disorder</th>
<th>Prevalence</th>
<th>with A</th>
<th>without A</th>
</tr>
</thead>
<tbody>
<tr>
<td>Depression</td>
<td>50</td>
<td></td>
<td>8</td>
</tr>
<tr>
<td>Mania</td>
<td>67</td>
<td></td>
<td>8</td>
</tr>
<tr>
<td>Eating Disorders</td>
<td>58</td>
<td></td>
<td>25</td>
</tr>
<tr>
<td>Schizophrenia</td>
<td>8</td>
<td></td>
<td>8</td>
</tr>
</tbody>
</table>


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**PROBLEM BEHAVIOURS**

PROPOSAL OF LINEAR STRUCTURE FOR ICD-11

F: MENTAL AND BEHAVIOURAL DISORDERS (meta-structure)

F1: NEURO-DEVELOPMENTAL DISORDERS (meta-category)

F1.Y PROBLEM BEHAVIOURS/BEHAVIOUR DISORDER (category)

F1.Y.1 Mild and infrequent
F1.Y.2 Mild and frequent
F1.Y.3 Severe and infrequent
F1.Y.4 Severe and frequent
F1.Y.5 External boundary prevents expression of behaviour
F1.Y.8 Unspecified

F1.Y.1-8.1 Physical aggression to others
F1.Y.1-8.2 Verbal aggression (e.g. screaming)
F1.Y.1-8.3 Destructive to property (e.g. throwing/pulling objects)
F1.Y.1-8.4 Self-injury
F1.Y.1-8.5 Oppositional
F1.Y.1-8.6 Overly-demanding
F1.Y.1-8.7 Sexually inappropriate (e.g. repeated stripping)
F1.Y.1-8.8 Other

Can multiple sub-sub categories be specified or not? – alternative descriptors if not

Psychiatric diagnosis or symptoms and Level of evidence

<table>
<thead>
<tr>
<th>Psychiatric diagnosis or symptoms</th>
<th>Level of evidence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Total sample: 648.</td>
<td></td>
</tr>
<tr>
<td>Sample with diagnosis: n 286</td>
<td></td>
</tr>
<tr>
<td>Depressive illness</td>
<td>Level II:</td>
</tr>
<tr>
<td>with psychotic symptom: n 93</td>
<td>at least one well-designed controlled study without randomisation</td>
</tr>
<tr>
<td>Obsessive-Compulsive behaviour: n 74 (25%)</td>
<td>Level III:</td>
</tr>
<tr>
<td>Psychotic illness (cycloid psychosis): n 70 (24.5%)</td>
<td>at least one well designed quasi-experimental study, such as a cohort study</td>
</tr>
<tr>
<td>Bipolar disorder: n 19 (6.6%)</td>
<td></td>
</tr>
<tr>
<td>PDD: n 15 (5.2%)</td>
<td></td>
</tr>
<tr>
<td>ADHD: n 14 (4.9%)</td>
<td></td>
</tr>
<tr>
<td>Anxiety: n 3 (1%)</td>
<td></td>
</tr>
<tr>
<td>Cognition</td>
<td></td>
</tr>
<tr>
<td>• Level II: 2 studies</td>
<td></td>
</tr>
<tr>
<td>• Level III: 9 studies</td>
<td></td>
</tr>
<tr>
<td>• expressive language</td>
<td></td>
</tr>
<tr>
<td>• visual-spatial processing</td>
<td></td>
</tr>
<tr>
<td>• Cognitive Rigidity</td>
<td></td>
</tr>
<tr>
<td>• Maths Skills</td>
<td></td>
</tr>
<tr>
<td>• attention switching</td>
<td></td>
</tr>
<tr>
<td>• sequential processing</td>
<td></td>
</tr>
<tr>
<td>• visual processing</td>
<td></td>
</tr>
<tr>
<td>• working memory</td>
<td></td>
</tr>
<tr>
<td>• short term memory</td>
<td></td>
</tr>
<tr>
<td>• visual-perceptual organization</td>
<td></td>
</tr>
</tbody>
</table>

Bertelli et al. (2012). PSICOGEN (PSIchiatric disorders and Cognition in GENetic syndromes)

A NEW CULTURAL MODEL FOR NEURODEVELOPMENTAL DISORDERS / CONDITIONS

PSYCHO-CHARACTERISATION

• SPECIF COGNITIVE FUNCTIONS
• INDIVIDUAL SKILLS
• INDIVIDUAL ATTRIBUTION OF IMPORTANCE
• PSYCHOPATHOLOGICAL VULNERABILITY
• SYMPTOMS PRESENTATION
• PROBLEM BEHAVIOURS

OFFER OF A WIDE RANGE OF OPPORTUNITIES AND SUPPORTS

IMPROVEMENT OF THE INDIVIDUAL IMPORTANCE/SATISFACTION
INDIVIDUAL QoL IMPROVEMENT

Bertelli et al. Advances in Mental Health and Intellectual Disabilities, in press
We are delighted to invite you to the 10th European Congress of Mental Health in Intellectual Disability to be held in Florence, Italy from the 9th – 11th September 2015.

In collaboration with and hosted by our Italian partners CDA and Misericordia, we look forward to meeting you in Florence, Italy.

Together we will create a place for reflection to exchange research, experiences and good practice. We will also bring new perspectives and support networking in the field of Mental Health and Intellectual Disability.

For all plenary sessions and many workshops and symposia we will provide simultaneous translation from English into French and Italian.

Add these dates to your agenda and be our guest in the beautiful surroundings of Florence. I look forward seeing you all in Italy!

Dr. Marco Botelli
President of the European Association for Mental Health in Intellectual Disability 2013-2015 – Psychiatrist, CREA (Research and Clinical Centre) della Fondazione San Sebastiano, Florence, Italy

Stefano Lassi
Vice-President of the European Association for Mental Health in Intellectual Disability 2013-2015 – Psychiatrist, Fondazione CDA Firenze Ostus of Florence, Italy
MARCO O. BERTELLI

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