

# A Pilot Study on the Diagnostic Performance of DMS-IV and DMS-V for Autism Spectrum Disorder

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## ABSTRACT

The new diagnostic criteria for autism spectrum disorders (ASD) are due to be released in May 2013. The impact of changes made in the new criteria is yet to be evaluated. Here we performed a retrospective study on a cohort of ASD patients diagnosed by DSM-IV criteria, aimed to compare the diagnostic performances between DSM-IV and DSM-V. We reviewed the medical records of 163 patients with possible clinical diagnosis of ASD. Ninety-three (57%) of them met the DSM-IV criteria for Autistic disorder, the rest 70 cases were either PDD-NOS (n=39) or Asperger's disorder (n=3) or without sufficient information in medical record to perform a clinical diagnosis (n=28). Upon re-evaluation using the new diagnostic criteria in DSM-V, only 60% of patients with previous diagnosis of autistic disorder met the new criteria. One individual who was previous diagnosed as PDD-NOS met the new diagnostic criteria for autistic disorder. The present study revealed a significant difference in diagnostic yield by new and old criteria. This pilot comparative study reveals that the ASD diagnostic criteria in DSM-V are stricter than that in DSM-IV and autism patients diagnosed using DMS-V criteria tend to be more severely affected. The new criteria will have immediate impact on the clinical diagnosis and management of individuals with neurodevelopmental disorders and it will affect the prevalence estimate of ASD in population as well.

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**KEY WORDS:** *autism spectrum disorder, autistic disorder, PDD-NOS, DSM-IV, DSM-V*

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## INTRODUCTION

Autism spectrum disorders are a group of clinically diagnosed neurodevelopmental disorders who share a set of complex behavioral phenotype involving difficulties in communication and reciprocal social interaction, as well as stereotypic repetitive behavior and unusually narrow interest.<sup>1</sup> The current clinical diagnostic criteria were delineated in the Diagnostic and Statistical Manual of Mental Disorders, 4th. Edition, better known as the DSM-IV, published by the American Psychiatric Association in 1994 (text revision in 2000).<sup>2</sup> Based on DSM-IV, autism spectrum disorder (ASD) refers to three categorical groups: autistic disorder, Asperger's disorder (AS) and pervasive developmental disorder - not otherwise specified (PDD-NOS). By DSM-IV criteria, patients with autistic disorder exhibits common characteristics such as severe difficulty in relations and communication associated with deficit in regulating sensory, attention, cognitive, motor and affective processes, with onset before age 3. The diagnostic criteria include qualitative impairment in social interaction, qualitative impairment in communication and restricted, repetitive and stereotyped patterns of behavior, interests and activities. AS patients are distinct from autistic disorder by their relatively normal language and cognitive development but involves all other diagnostic criteria in DSM-IV.<sup>2,3</sup> PDD-NOS are referred to as atypical PDD or atypical autism for children who do not meet full criteria in all three diagnostic domains. This group includes children with milder symptoms in all three diagnostic domains and those meeting full criteria for autism in two of the three domains. Sometimes PDD-NOS is used as an initial or tentative diagnosis for younger children or before diagnostic evaluations are completed.<sup>3</sup> The lack of clear definitions for this relatively heterogeneous group of children presents problems for research on this condition as well as for patient care. The DSM-IV had been criticized for relaxed criteria that may have lead to the widening of ASD diagnosis.

The draft DSM-V has been published recently.<sup>4</sup> In the proposed revision, there are only two domains as opposed to the three domains in DSM-IV (see the appendix 1 for details of the diagnostic criteria of both DSM-IV and DSM-V). The two domains are: 1) social/communication deficits, 2) fixated interests and repetitive behaviors. Unusual sensory behaviors are included within a subdomain of stereotyped motor and verbal behaviors with examples particularly relevant for younger children. In DSM-V, Asperger's disorder will be subsumed into an existing disorder: autism spectrum disorder.

The changes made in DSM-V are quite substantial. It is not known if this revision will have significant impact in the clinical diagnosis of patients with suspected ASD and how the new criteria will affect the ASD prevalence in the population level. Here we performed a retrospective pilot study to compare the diagnostic performance of DSM-V with that of DSM-IV.

## PATIENTS AND METHODS

### Subject and Data Collection

We reviewed the medical records of 163 patients with suspected ASD diagnosis at the Department of Developmental Medicine in Children's Hospital Boston. A standard coding system including information of initial and follow-up evaluation was used to score each individual using both DSM-IV and DSM-V diagnostic criteria (see appendix 2). The clinical characteristics of the cohort are as following: 80% of the cases were boys and 20% were girls (M:F=4:1); the average age was 2 years and 10 months at their initial diagnosis of ASD (range from 18 months to 56 months, SD=10.4 months). All patients underwent ASD clinical (by DSM-IV) and neurological evaluation, as well as play

observation. Two thirds of patients also underwent psychological, MRI and EEG evaluations. About half of the patients had metabolic evaluation (thyroid function (43%), plasma amino acids/plasma and urine organic acids (53%)). Regarding genetic evaluation, the majority of patients had chromosomal microarray analysis (94%), 44% had G-banding karyotyping, 38% had Fragile X testing, and 20% had MECP2 testing.

### Statistical Analysis

Subject characteristics were described according to study group (cases met the criteria or not by DSM-IV or DSM-V) and compared by using  $\chi^2$  tests. Association of items in DSM-IV with the positive diagnosis of autistic disorder by DSM-V was estimated using unconditional logistic regression. All p-values are two-sided, and all analyses were carried out using SPSS software packages (version 11, SPSS, USA).

The study is approved by the internal review board of Children's Hospital Boston.

**Table 1.** Results of physical examination and clinical evaluation by DSM-IV and DSM-V.

Variables	DSM-IV Cases (%)		DSM-V Cases (%)	
	93 cases met the diagnosis of autistic disorder by DSM-IV	70 cases did not meet the diagnosis of autistic disorder by DSM-IV	57 cases met the diagnosis of autistic disorder by DSM-V	106 cases did not meet the diagnosis of autistic disorder by DSM-V
ID/MR	45(48.39)	27(38.57)	28(49.12)	44(41.51)
Seizures	37(39.78)	23(32.86)	22(38.59)	38(35.84)
Positive family history	51(54.84)	26(37.14)	33(57.89)	44(41.51)
ASD	20(21.51)	8(11.43)	12(21.05)	16(15.09)
ID/MR, learning disability, language delay	20(21.51)	14(20.00)	15(26.32)	19(17.92)
Mood problem	11(11.83)	4(5.71)	7(12.28)	8(7.55)
Hypotonia	29(31.18)	18(25.71)	19(33.33)	28(26.42)
Hypertonia	10(10.75)	3(4.29)	5(8.77)	8(7.55)
Macrocephaly	15(16.13)	11(15.71)	9(15.79)	17(16.04)
Microcephaly	9(9.68)	7(10.00)	8(14.04)	8(7.55)

## RESULTS

### Characteristics of participants by DSM-IV and DSM-V

**Table 1** listed all patients in the cohort, their physical examination and clinical evaluation by both DSM-IV and DSM-V. As a result, 93 out of 163 cases met the DSM-IV criteria for autistic disorder. 48% of patients had intellectual disability/mental retardation (ID/MR) and 40% had seizure; 55% of patients had positive family history of ASD (22%), ID/MR, learning disability, language delay (22%) or mood problem (12%). Of the 163 cases, 70 patients did not meet the DSM-IV criteria for autistic disorder. Among them, 39% had ID/MR and 33% had seizure; 37% of patients had

positive family history of ASD (11%), ID/MR, learning disability, language delay (20%) or mood problem (6%). Also detailed results of physical examination and clinical evaluation by DSM-V are shown in **Table 1**. There is no statistically significant difference in physical examination and clinical evaluation between two groups classified by DSM-IV or DSM-V.

### Items Comparison between Two Groups By DSM-V

We found a significant ASD diagnostic difference between DSM-IV and DSM-V criteria. Among the 93 patients who met the diagnostic criteria by DSM-IV, there are only 56

individuals that also met the diagnostic criteria by DSM-V. The concordant rate between the two criteria is about 60%. In order to identify the cause of such significant difference, we examined the presence or absence of each diagnostic item in DSM-V for every patient. The results are showing in **Table 2**. By definition, all autistic disorder patients have to meet all three items in domain A, thus their positive rate are 100%; for domain B items, B1 has the highest positive rate (98%) and B2 has the lowest positive rate (32%) in patients with autistic disorder. The items that contributed significantly to the difference of diagnostic yield are A2, A3, B2, B3 and B4 ( $P \leq 0.001$ ).

We further performed logistic regression analysis for the 93 cases with diagnostic items in the three domains of DSM-IV.

In **Table 3**, the item C1 (stereotypic and restricted interest) of DSM-IV showed statistically significant association (OR=12.97, 95%CI: 2.76-60.89) with the positive diagnosis of autistic disorder by the new criteria. The item C 2 (inflexible to routine) appears to have the least predictive power in DSM-IV for positive diagnosis by DSM-V.

In addition, there is one PDD-NOS patient by DSM-IV met the diagnosis of autistic disorder by DSM-V (patient # 67 in **Appendix 2**).

**Table 2.** Comparison of positive rates of items between two groups classified by DSM-V.

Items DSM-V	56 cases met the diagnosis of autistic disorder by DSM-V		37 cases did not meet the diagnosis of autistic disorder by DSM-V		70 cases did not meet the diagnosis of autistic disorder by DSM-IV		$\chi^2$ *	P-value
	YES	NO	YES	NO	YES	NO		
A1	56	0	35	2	25	45	1.06	0.3037
A2	56	0	27	10	19	51	14.26	0.0004
A3	56	0	29	8	24	46	10.64	0.0011
B1	55	1	37	0	30	40	0.04	0.8338
B2	18	38	0	37	2	68	12.76	0.0004
B3	32	24	4	33	4	66	18.25	0.0001
B4	42	14	4	33	15	55	34.2	0.0001

\* Chi square tests were between the group of 56 cases who met the diagnosis of autistic disorder by DSM-V and the group of 37 cases who did not meet the diagnosis of autistic disorder by DSM-V

**Table 3.** Logistical regression coefficients and their standard errors (S.E) for items in DSM-IV and autistic disorders by DSM-V.

Items in DSM-IV	Correlation coefficient	S.E.	OR(95% CI)	P-value
A1	0.18	0.32	1.19(0.64-2.22)	0.580
A2	0.32	0.43	1.37(0.59-3.20)	0.465
A3	0.49	0.88	1.64(0.29-9.20)	0.575
A4	0.51	0.37	0.60(0.29-1.25)	0.175
B1	0.63	0.42	1.87(0.82-4.25)	0.135
B2	0.12	0.59	0.88(0.28-2.83)	0.835
B3	0.25	0.58	0.78(0.25-2.45)	0.672
B4	0.13	0.41	1.14(0.51-2.56)	0.757
C1	2.56	0.79	12.97(2.76-60.89)	0.001
C2	21.56	8.14E+03	2.30E+09(0.00-.)	0.998
C3	0.43	0.43	1.54(0.67-3.58)	0.310
C4	1.60	1.15	4.94(0.52-47.29)	0.166
Constant	1.73	1.14	0.18	0.129

## DISCUSSION

It is known to the autism research and clinical community that DSM-IV has limitations when it applies to children with ASD. There has been a lot of criticisms regarding the ASD

diagnostic criteria in DSM-IV.<sup>5</sup> In particular, the PDD-NOS subgroup is not well defined in terms of what should be excluded from their definition, and the distinction between Childhood Autism and PDD-NOS in terms of functioning is

unclear. Sometimes PDD-NOS is used as a tentative diagnosis for young children, this practice may have led to the widening of ASD diagnosis. Newly published DSM-V reduced three domains to two and three subgroups of PDD in DSM-IV were combined to one: no more PDD-NOS and Asperger diagnosis by DSM-V. While the new criteria is aimed to remedy the deficit existed in the old criteria, and their performance in real clinical practice are yet to be systematically evaluated. This pilot study used a cohort of patients with prior ASD diagnosis by DSM-IV. The preliminary results showed that there are significant differences in diagnostic yield using the old and new criteria.

Generally, ASD patients diagnosed by DSM-V have more symptoms on all three domains compared to those by DSM-IV (**Table 2**). DSM-V criteria tend to be stricter since only 60% of children with autistic disorder diagnosed by DSM-IV met the criteria of DSM-V. Almost all patients, except one, with PDD-NOS no longer meet the diagnostic criteria of DSM-V. Overall, the adjustment made in the new diagnostic criteria may be responsible for such difference. In DSM-V, 12 items in all of three domains were reduced to 7. Language related items were reduced to a part of item 1 in B domain. Six positive items required by DSM-IV were reduced to five in DSM-V. In the social/communication domain, all three items must be met in DSM-V while in DSM-IV only 2 of 4 items need to be met. In the domain of fixated interests and repetitive behaviors, 2 of 4 items in DSM-V and 1 of 4 items in DSM-IV are needed. It seems that DSM-V has less selection and thus, is stricter than DSM-IV. It also suggests that the new criteria place more focus on the social/communication, fixated interests and repetitive behaviors aspects of the clinical manifestation. The language function on the other hand is less emphasized. Emphasizing language deficit by DSM-IV may account for higher prevalence of ASD diagnosis when using DSM-IV. Based on the positive rate comparison between the two groups of patients in **Table 2** (ASD patients still met the diagnostic criteria by DSM-V and those did not meet the new diagnostic criteria), it showed that the majority of patients in both groups have very high positive rate for the item 1 in A domain (social-emotional reciprocity) and the item 1 in B domain (stereotypic repetitive behavior) of DSM-V. These are essential features of patients with ASD but insufficient for the new diagnostic criteria. The item 2, 3 in A domain and the item 2, 3, 4 in B domain of DSM-V are the ones that differentiate patients in this cohort whether or not meet the new diagnostic criteria. Consistent with the previous study,<sup>6</sup> these items that showed significant differences ( $P < 0.001$ ) between the two groups indicate the severity of ASD and may distinguish young children with autistic disorder from those with PDD-NOS. Our data is in line with researchers who suggest that PDD-NOS is a less severe variant of autism.<sup>7</sup> Thus, according to our results, the current draft guidelines for DSM-V may exclude such patients who may have autistic spectrum. Interestingly, we conducted a logistic regression, the item C1 in DSM-IV has the best predictive power for positive diagnosis of autism by DSM-V

(OR=12.97, 95%CI: 2.76-60.89). Thus “restricted interest and preoccupied focus” may be an important key feature to determine whether or not patients will meet DSM-V criteria. This finding is consistent with both the empathizing-systemizing theory and the central coherence theory of autism. Both theories predict excellent attention to detail in autism patients.<sup>8,9</sup>

In summary, the DSM-V criteria tend to be stricter and may target ASD cases with high severity, and most of patients with PDD-NOS diagnosed by DSM-IV will not meet the new diagnostic criteria. This could help to correct the trend of widening the diagnostic criteria for ASD in routine clinical practice as well as in population based epidemiological study. Clinically, diagnostic evaluation will place emphasis on the social/communication, fixated interests and repetitive behaviors aspect of clinical features when using DSM-V. Patients with certain autistic features, especially in the language domain, but lacking solid evidence for their deficit in other domains will not be diagnosed with ASD. Our study is limited by retrospective design which is based on the review of medical records, not on clinical diagnosis in person. With this limitation, the conclusion should be considered as preliminary but with instructive utility.

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## Appendix 1. Diagnostic criteria of DSM-IV for autistic disorder and draft DSM-V for ASD.

### DSM-IV diagnostic criteria for autistic disorder:

- I. A total of six (or more) items from (A), (B), and (C), with at least two from (A), and one each from (B) and (C)
- (A) Qualitative impairment in social interaction, as manifested by at least two of the following:
1. marked impairments in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body posture, and gestures to regulate social interaction;
  2. failure to develop peer relationships appropriate to developmental level;
  3. a lack of spontaneous seeking to share enjoyment, interests, or achievements with other people, (e.g., by a lack of showing, bringing, or pointing out objects of interest to other people);
  4. lack of social or emotional reciprocity ( note: in the description, it gives the following as examples: not actively participating in simple social play or games, preferring solitary activities, or involving others in activities only as tools or "mechanical" aids ).
- (B) Qualitative impairments in communication as manifested by at least one of the following:
1. delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime);
  2. in individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation; with others
  3. stereotyped and repetitive use of language or idiosyncratic language;
  4. lack of varied, spontaneous make-believe play or social imitative play appropriate to developmental level.
- (C) Restricted repetitive and stereotyped patterns of behavior, interests and activities, as manifested by at least one of the following:
1. encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal either in intensity or focus;
  2. apparently inflexible adherence to specific, nonfunctional routines or rituals;
  3. stereotyped and repetitive motor mannerisms (e.g hand or finger flapping or twisting, or complex whole-body; movements)
  4. persistent preoccupation with parts of objects.
- II. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (A) social interaction, (B) language as used in social communication, (C) symbolic or imaginative play

### Draft DSM-V diagnostic criteria for ASD:

Must meet criteria A, B, C, and D:

- A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays, and manifest by all 3 of the following:
1. Deficits in social-emotional reciprocity; ranging from abnormal social approach and failure of normal back and forth conversation through reduced sharing of interests, emotions, and affect and response to total lack of initiation of social interaction,
  2. Deficits in nonverbal communicative behaviors used for social interaction; ranging from poorly integrated- verbal and nonverbal communication, through abnormalities in eye contact and body-language, or deficits in understanding and use of nonverbal communication, to total lack of facial expression or gestures.
  3. Deficits in developing and maintaining relationships, appropriate to developmental level (beyond those with caregivers); ranging from difficulties adjusting behavior to suit different social contexts through difficulties in sharing imaginative play and in making friends to an apparent absence of interest in people
- B. Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least two of the following:
1. Stereotyped or repetitive speech, motor movements, or use of objects; (such as simple motor stereotypies, echolalia, repetitive use of objects, or idiosyncratic phrases).
  2. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change; (such as motoric rituals, insistence on same route or food, repetitive questioning or extreme distress at small changes).
  3. Highly restricted, fixated interests that are abnormal in intensity or focus; (such as strong attachment to or preoccupation with unusual objects, excessively circumscribed or perseverative interests).
  4. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment; (such as apparent indifference to pain/heat/cold, adverse response to specific sounds or textures, excessive smelling or touching of objects, fascination with lights or spinning objects).
- C. Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities)
- D. Symptoms together limit and impair everyday functioning.

**Appendix 2.** Patients with ASD classified by DSM-IV and draft DSM-V respectively.

Cases	DSM-IV														DSM-V								
	12 items in three domains												Diagnosis (case number)		7 items in two domains							Diagnosis (case number)	
	A 1	A 2	A 3	A 4	B 1	B 2	B 3	B 4	C 1	C 2	C 3	C 4	AD (93)	AS (3)	PDD-NOS (39)	A 1	A 2	A 3	B 1	B 2	B 3	B 4	AD (57)
1	1	0	0	1	1	1	1	0	0	0	1	0	1	0	0	1	1	0	1	0	0	0	0
2	0	0	0	1	0	1	0	0	0	0	0	0	0	0	0	1	0	1	0	0	0	0	0
3	0	0	0	1	1	1	1	0	0	0	1	0	1	0	0	1	0	1	1	0	0	0	0
4	1	1	1	1	1	1	1	1	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
5	1	0	0	1	0	1	1	0	0	0	1	0	1	0	0	1	1	0	1	0	0	0	0
6	1	1	1	1	1	1	1	1	0	0	1	0	1	0	0	1	1	1	1	0	0	1	1
7	1	1	1	1	1	1	1	1	1	0	1	0	1	0	0	1	1	1	1	0	1	0	1
8	1	0	0	1	0	1	0	1	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
9	1	1	0	1	1	1	1	1	1	0	1	1	1	0	0	1	1	1	1	0	1	0	1
10	1	1	0	1	1	0	1	1	1	0	1	0	1	0	0	1	1	1	1	0	1	1	1
11	1	0	0	0	0	1	1	0	0	0	1	0	0	0	1	0	1	0	1	0	0	0	0
12	1	1	1	1	1	1	1	1	1	1	1	0	1	0	0	1	1	1	1	1	1	0	1
13	0	1	0	1	1	1	0	1	0	0	1	0	1	0	0	1	0	1	1	0	0	0	0
14	0	0	0	1	0	0	0	1	0	0	1	0	0	0	1	0	1	1	0	0	1	0	0
15	0	0	0	0	0	1	1	0	0	0	1	0	0	0	1	0	0	1	0	0	0	0	0
16	1	0	0	1	1	1	1	0	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
17	1	1	0	1	1	1	1	1	1	1	1	1	1	0	0	1	1	1	1	1	1	0	1
18	1	1	0	1	1	1	1	1	1	0	1	1	1	0	0	1	1	1	1	1	1	0	1
19	1	1	0	1	1	1	0	1	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
20	1	1	1	1	1	1	1	0	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
21	0	1	0	0	1	1	1	0	1	0	1	0	0	0	1	0	0	1	0	1	0	0	0
22	1	1	0	1	1	1	1	1	1	0	1	0	1	0	0	1	1	1	1	0	1	0	1
23	1	1	0	1	1	1	1	1	0	0	1	0	1	0	0	1	1	1	1	0	0	1	1
24	1	1	0	1	0	1	1	0	1	0	1	0	1	0	0	1	1	1	1	0	1	1	1
25	1	0	0	1	0	0	0	0	0	0	0	0	0	0	0	1	1	1	0	0	0	0	0
26	1	0	0	0	1	0	0	0	0	0	1	0	0	0	1	0	1	0	1	0	0	0	0
27	1	1	0	1	1	1	1	1	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
28	0	0	0	1	1	1	0	0	0	0	0	0	0	0	0	1	0	1	0	0	0	0	0
29	1	1	0	1	1	1	1	1	1	0	1	0	1	0	0	1	1	1	1	0	1	0	1
30	1	1	0	1	1	1	1	0	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
31	1	1	0	1	0	1	0	0	0	0	0	0	0	0	1	1	1	0	0	0	0	0	0
32	0	1	0	1	0	1	0	0	0	0	0	0	0	1	0	1	0	1	0	0	0	1	0
33	1	1	0	1	1	1	1	1	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
34	1	1	0	1	1	1	0	1	0	0	1	0	1	0	0	1	1	1	1	0	0	1	1
35	1	1	0	1	1	1	1	0	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
36	0	0	0	1	0	1	1	0	0	0	1	0	0	0	1	0	0	1	0	0	0	0	0
37	1	1	0	1	0	1	1	0	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
38	1	1	0	0	1	1	0	0	0	0	1	0	0	0	1	0	1	1	1	0	0	1	0
39	1	1	0	0	1	1	1	1	0	0	1	0	1	0	0	0	1	1	1	0	0	0	0
40	1	1	0	1	1	1	1	0	0	0	1	0	1	0	0	1	1	1	1	0	0	0	0
41	1	1	0	1	1	1	1	1	1	0	1	0	1	0	0	1	1	1	1	0	1	0	1
42	0	1	1	1	0	0	1	1	0	0	1	1	1	0	0	1	0	1	1	0	1	0	0
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