Published in final edited form as:

Autism Res. 2017 April ; 10(4): 653–662. doi:10.1002/aur.1706.

‘Frank’ presentations as a novel research construct and element of diagnostic decision-making in autism spectrum disorder

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Scientific Abstract

Many individuals with ASD have a distinctive behavioral presentation that is recognizable within moments, a phenomenon we call “frank” ASD. This phenomenon has been discussed informally for decades, perhaps as “classic” ASD; however, there is no unitary “classic” presentation, and classic autism does not seem to correspond to level of functioning. Thus, neither “frank” nor “classic” autism has been delineated or studied as a research construct. To initiate the empirical study of frank ASD, we surveyed 151 clinicians, from a range of disciplines that diagnose ASD, about this phenomenon. Respondents completed a 13-item questionnaire about frank ASD, which was analyzed using a mixed-methods approach. 97% of respondents were familiar with the phenomenon. Respondents estimated that 40% of the ASD population has a frank presentation. Respondents reported the most highly specific behaviors associated with frank presentations were a general sense of impaired reciprocity, quality of eye contact, atypical vocal prosody, presence of motor mannerisms, and atypical gait or posture. In general, respondents reported detecting frank features rapidly, with the majority forming their impressions within the first ten minutes of interaction or observation. Although unstudied empirically, “frank” presentations of ASD are familiar to diagnosing clinicians, and appear to be based on behaviors both central to ASD diagnostic criteria (e.g., impaired reciprocity), and absent from diagnostic criteria (e.g., atypical gait or posture). We discuss these findings within the context of diagnostic decision-making and behavioral phenotyping of ASD.

Lay Abstract

Many individuals with ASD have a distinctive behavioral presentation that is recognizable within moments. We call this phenomenon “frank” ASD, a term we adopted from the medical literature, where “frank” is used to mean “clinically evident and unmistakable.” This phenomenon has been discussed within clinical practice for decades, perhaps as “classic” autism, though there is not a unitary “classic” presentation and it does not correspond to level of functioning. Rather, there are likely several “frank” presentations. Neither “frank” nor “classic” autism has been specifically outlined by researchers, or studied scientifically. To initiate the scientific study of frank ASD, we

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surveyed 151 clinicians, from a range of disciplines that diagnose ASD, about this phenomenon. Respondents completed a 13-item questionnaire. 97% of respondents had experienced the phenomenon of frank autism. Clinicians estimated that 40% of the ASD population has a frank presentation. When asked an open-ended question about the single most specific behavior that contributes to a frank presentation, clinicians reported: an overall sense of impaired reciprocal interactions, the quality of eye contact, atypical vocal prosody, presence of motor mannerisms, and atypical walking or posture. In general, respondents reported detecting frank features rapidly, often within the first ten minutes of interaction or observation. Frank presentations of ASD seem to be familiar to diagnosing clinicians, and are based on behaviors both central to diagnostic criteria (e.g., impaired reciprocity), and not (e.g., atypical gait or posture). We discuss these findings within the context of diagnostic decision-making and understanding behavior in ASD.

Keywords

Autism spectrum disorder; diagnostic decision-making; behavioral phenotype

Clinicians with expertise diagnosing autism spectrum disorder (ASD) often report that some patients with ASD have such a clear behavioral presentation that the diagnosis is apparent within the first moments of interaction – a phenomenon we refer to as "frank" ASD[1] though others may have used the term "classic" ASD. Frank ASD, though discussed anecdotally, has not been delineated as a construct, though perhaps it should be. Frank features of ASD may have important roles in clinical care and research that may or may not be currently recognized.

In clinical care, if rapid impressions accurately predict diagnosis, the utility of resource-intensive diagnostic evaluations should be examined. More efficient tools and diagnostic processes could be introduced in both clinical and clinical research settings. If rapid impressions are inaccurate, however, then clinicians need a method for ensuring they do not fall victim to confirmation bias or diagnostic overshadowing.

In research, frank features may be the biobehavioral markers that are the most feasible and inexpensive to measure, and these features may provide insight into more specific biological mechanisms than broader characteristics. For example, the specific mechanisms behind atypical prosody may be more accessible than the multitude of mechanisms involved in a child's difficulty maintaining friendships. Finally, in our understanding of ASD and how it affects individuals, understanding the extent to which frank features do –or do not— correspond to outcome and level of functioning may be important. There are many highly accomplished individuals who also have frank features of ASD. Understanding when to provide intervention to improve functioning, and when to step back and appreciate human diversity is critical to cultivating a healthy society.

If frank ASD is a valuable construct, it will require consistent terminology. Here we aim to integrate multiple literatures relevant to the study of frank ASD, and to consolidate expert opinion on frank presentations within the context of ASD diagnosis.

[1]We have adopted this term from the medical literature, where "frank" is used to mean "clinically evident and unmistakable."

Clinicians have been reliable in making the diagnosis of infantile autism since the introduction of the term in the DSM-III (Mattison, Cantwell, Russell, & Will, 1979), suggesting that certain features are highly specific to the disorder. As part of the DSM-IV Autism Field Trial, Klin and colleagues (2000) examined clinical reliability of PDD diagnoses in 131 cases. They compared reliability among "clinician-assigned" diagnoses (i.e., the clinician assigned a diagnosis based on clinical judgment only – without using the DSM) and "DSM-IV assigned" diagnoses (i.e., the clinician determined which symptoms the patient had, and diagnosis was determined via DSM-IV algorithm). Among pairs of experienced clinicians, reliability for clinician-assigned diagnosis was extremely high (kappa = .94), supporting earlier findings of high clinician agreement for ASD diagnosis. Kappa for DSM-IV assigned diagnoses, while still in the excellent range (kappa = .84), was *lower* than for clinician-assigned diagnoses, suggesting that experienced clinicians consider additional diagnostic information that is not fully captured by DSM symptoms. We propose that frank features may be part of this missing information.

Informally, clinicians describe detecting frank ASD rapidly, on the basis of a patient's overall behavioral presentation. A recent study (Gabrielsen et al., 2015) supports these anecdotal reports, showing that diagnosing providers are able to identify ASD in a portion of cases based on brief behavioral observations. Licensed psychologists watched 10-minute segments of the Autism Diagnostic Observation Schedule (ADOS; C. Lord, Rutter, DiLavore, & Risi, 2002) in toddlers who had failed an ASD screener. One-third of the sample was ultimately diagnosed with ASD, one-third with language delay (no ASD), and one-third were deemed typically developing (false positives on screeners). Following each 10-minute observation, clinicians indicated whether they would refer the child for an ASD-specific assessment. 61% of toddlers diagnosed with ASD were referred, compared to 25% of the language-delayed group, and 11% of the typical group. Overall sensitivity and specificity of the referral accuracy were 0.61 and 0.82, respectively. These findings suggest that trained clinicians are able to rapidly discern behavioral features indicative of ASD by observation alone. They also suggest that rapid appraisal of ASD may only be possible in a subset of children with ASD – even with the conservative judgment used in this study (i.e., a decision to refer, not a diagnosis) only 61% of toddlers were correctly identified. Toddlers who are rapidly identified may present with specific "frank" behavioral characteristics that differentiate them from toddlers who are not rapidly identifiable but also meet strict ASD diagnostic criteria.

Experimental studies focused on the behavioral presentation of ASD also suggest that even non-experts can rapidly detect differences in behavior between people with and without ASD, at least at the group level. For example, after watching short video clips (some as short as one second) of cases and controls, untrained undergraduates successfully discriminate groups, with large effect sizes (de Marchena & Eigsti, 2010; Fusaro et al., 2014; Grossman, 2014) (Fusaro et al., 2014), suggesting that there is something about the behavioral presentation of ASD that is readily apparent to the untrained eye. Two of these studies included only participants with intact cognitive and language skills, suggesting that these qualitative judgments go beyond an individual's level of function. Unlike the Gabrielsen (2015) study that looked at expert decision-making in a heterogeneous, real-world clinical sample, these studies are limited in that they compare cases with ASD to control participants

with no clinical symptoms of any kind, which may be too straightforward a comparison to have clinical utility.

In summary, while frank ASD has not been a specific focus of empirical research, there is evidence from the literature on diagnostic decision making and referral making suggesting both that there are features of behavior in ASD that can be detected rapidly, even by novices, and that there are features of behavior relevant to ASD diagnosis that are not fully captured by existing diagnostic criteria. We believe that frank presentations encapsulate many of these features, along with well-known features central to ASD diagnostic criteria. Here we consolidate expert opinion on frank ASD with the goal of formally defining the construct so that the field can begin to measure frank ASD more systematically for research and clinical purposes.

Method

Questionnaire

We designed a brief questionnaire (Appendix A) to address several central questions related to frank ASD, including 1) were clinicians familiar with the phenomenon? 2) how did clinicians think they formed these impressions of frank ASD? We also invited clinicians to comment freely, and collected basic information about respondents themselves.

Sources of Recruitment

Target participants were clinicians with professional qualifications to make a medical diagnosis of ASD (i.e., psychologists and physicians), who diagnose ASD as part of their clinical practice. With the goal of recruiting a broad sample of clinicians, participants were invited in several ways. 1) We used the *Autism Speaks Resource Guide* (https://www.autismspeaks.org/family-services/resource-guide) to find individual practitioners and facilities that self-identify as providing ASD diagnoses. Every provider in the “Where to get an autism diagnosis” section with a valid email address was contacted and invited to complete the questionnaire, for a total of 823 emails sent. 2) We sent a mass email to providers on the listserv maintained by the Center for Autism Research at the Children’s Hospital of Philadelphia, resulting in 501 additional emails sent. 3) We sent the questionnaire to current and former colleagues with experience diagnosing ASD, for a total of 35 emails sent. This protocol was deemed exempt by the Institutional Review Board at the Children’s Hospital of Philadelphia.

Respondents

195 providers completed the survey (14% response rate). Forty-four respondents had expertise in ASD but were not from disciplines that make ASD diagnoses (e.g., speech pathology, social work). As the emphasis of the current study was on the diagnostic process, these respondents were excluded from the current study, resulting in a total of 151 eligible participants.

Clinicians from across the United States (34 states) responded to the survey. Providers were primarily clinical psychologists (n=121), but several medical disciplines were also

represented, including developmental-behavioral pediatrics (n=23), psychiatry (n=3), neurology (n=2), and general pediatrics (n=2). Providers varied considerably in their actual experience diagnosing ASD, with a range of 1–40 years of experience (median 13 years) and 1–5,000 ASD diagnoses given (median 250 diagnoses). Collectively, by their own estimates, respondents had provided over 62,000 ASD diagnoses in the course of their careers. 38% of respondents identified as ASD researchers as well as clinicians.

Analytic Approach

Given the quantitative and qualitative aspects of the information gathered by the survey, we took a mixed methods approach to analyzing and reporting our data, following the recommendations of the NIH working group on Best Practices for Mixed Methods in the Health Sciences (Creswell, Klassen, Plano Clark, & Smith, 2011).

Quantitative analyses were performed using SPSS; whenever inferential statistics were computed, variables were tested for normality and transformed as appropriate prior to statistical testing.

Qualitative data are reported following the Consolidated Criteria for Reporting Qualitative Research (Tong, Sainsbury, & Craig, 2007). Two items from the survey were considered qualitative and subjected to data coding: Question 6 (behaviors associated with frank features), and free-form comments about frank ASD.

Results

Clinician Familiarity with Frank ASD

97% of eligible providers responded “yes” to the question, “have you ever experienced the ‘you know it when you see it’ [frank ASD] phenomenon?” demonstrating that a very high proportion of diagnosing clinicians have encountered patients with a frank presentation. Providers who responded “no” to this question were not prompted to answer other questions specific to frank ASD.

To estimate whether frank presentations are evident across the autism spectrum, we asked clinicians to indicate whether they had seen frank cases in four subgroups that varied by age and verbal ability. Clinicians endorsed seeing frank cases across all four subgroups, see Table 1.

When asked to estimate what percentage of individuals with ASD present with frank features, clinicians provided a wide range of responses – from 2% to 95% of cases. On average, clinicians reported that 40% of patients diagnosed with ASD have a frank presentation. We note that the questionnaire was specifically worded, “of children you have *diagnosed*” to ensure that clinicians were reporting on diagnosed patients, not the broader population of ASD referrals. For mean, median, and mode of percent with frank presentations, see Table 2.

These findings suggest 1) that the overwhelming majority of ASD clinicians endorse the phenomenon of frank ASD, 2) that frank cases are seen across the ASD spectrum, and 3)

that frank features are evident in a subset of ASD cases (i.e., not everyone with the diagnosis).

Speed of Judgments of Frank ASD

To test the hypothesis that clinicians often detect frank features rapidly, we asked providers to report how quickly they thought they formed impressions of frank ASD. We left this question open-ended to reduce the chance of biasing respondents toward any particular time frame. 133 clinicians provided valid responses, which were categorized based on commonly provided values. Twelve responses that were judged to be irrelevant to the question, or not specific enough to categorize (e.g., “very quickly”) were considered invalid and excluded. A wide range of values was provided, from seconds to multiple assessment sessions. The most commonly provided value was five minutes or “a few minutes” (collapsed into the same category; 24% of respondents). 52% of participants described themselves as detecting frank features within 10 minutes, supporting the hypothesis that these impressions are often made rapidly. Table 3 describes these responses in more detail.

Clinician Factors Influencing Judgments of Frank ASD

We collected demographic data from participants to determine if clinical impressions of frank ASD varied in any way as a function of discipline or experience. There were no differences between physicians and psychologists in terms of either the proportion of patients with ASD they considered frank $t(125) = 1.01$, $p = .32$, Cohen’s $d = 0.22$, or the speed at which they formed impressions of frank ASD, $t(131) = 0.94$, $p = .35$, Cohen’s $d = 0.21$. To test the effect of experience, we divided the sample in two ways: 1) we conducted a median split on number of years of experience (median = 13 years), and 2) we conducted a median split on number of cases diagnosed (median = 250 diagnosed). Years of experience did not significantly predict either the speed at which clinicians formed impressions of frank ASD, $t(129) = 0.23$, $p = .82$, Cohen’s $d = 0.05$, or the proportion of cases considered frank, $t(123) = 0.61$, $p = .55$, Cohen’s $d = 0.11$. In contrast, when the group was split by number of cases assessed, we found that more experienced clinicians did form impressions of frank ASD differently. Specifically, they were marginally faster in forming their impressions, $t(108) = 1.98$, $p = .05$, Cohen’s $d = 0.37$, and considered a greater proportion of cases to be frank, $t(102) = 2.01$, $p = .047$, Cohen’s $d = 0.39$. These findings suggest that experience with ASD itself (and not just general clinical practice) may change how clinicians detect frank features.

Clinician Terms for Frank ASD

Of the 146 providers who endorsed the phenomenon of frank ASD, 81 (55%) answered ‘yes’ to the question, “Do you have a term for this phenomenon (even if used only informally),” suggesting that the phenomenon is common and/or salient enough to be labeled. Some providers used generic expressions alluding to a clear ASD presentation (e.g., “clearly ASD,” “obvious autism”). Many clinicians endorsed terms associated with a prototypical presentation; specifically, 13 providers spontaneous used the term “classic.” Finally, consistent with the hypothesis that detection of frank features happens rapidly, 28 clinicians (one-third of those who provided a term) endorsed using a phrase consistent with rapid impression formation from the first few moments of meeting the patient, reflecting the

fact that a brief interaction, such as an initial handshake or greeting, or even a glimpse of the patient interacting in the waiting room, was sometimes sufficient to form an impression of frank ASD.

Behavior Associated with Frank Presentations

A major goal of this study was to describe how clinicians form their impressions of frank ASD, including identifying what specific behaviors, and/or features of behaviors might give rise to an impression of frank ASD.

Coding frank behaviors—We took a data-driven approach to analyzing the spontaneously reported behaviors associated with frank presentations, to identify specific behavior categories that clinicians believe are responsible for their impressions of frank ASD. The first author reviewed all responses and created categories of commonly provided example behaviors. Specific behaviors spontaneously endorsed in at least 1% of responses were included. 21 categories were identified. Both authors then coded 200 of the reported behaviors (32%) to establish reliability. Kappa was .83, demonstrating excellent reliability.

Five thematic categories emerged: “social reciprocity,” “nonverbal communication,” “repetitive behaviors and interests,” “atypical motor behavior (non-repetitive),” and “other.” The first three were divided into subcategories. The fourth reflected atypicalities in general motor behavior that were frequently endorsed, and did not clearly fit into any of the above categories. Finally, “other” was included to capture both low-frequency behaviors that could not easily be categorized into another group (these were separated into ASD-specific behaviors, and behaviors more broadly associated with psychopathology and developmental disabilities) and responses that were too vague to categorize. Data are presented in Table 4, ordered from most frequently endorsed to least frequently endorsed.

Clinicians’ Open-ended Commentary on Frank ASD

Coding—At the end of the questionnaire, clinicians were invited to comment on frank ASD in an open-ended manner. All comments were reviewed by the first author and commonly-endorsed themes were extracted. After the list of themes was developed and reviewed by the second author, each individual comment was revisited and individually coded; comments could be coded as reflecting multiple themes.

53 clinicians (36% of respondents) provided free-form comments. Responses ranged in length from 7 to 587 words. Clinician comments primarily centered around two themes: 1) What is frank ASD, and 2) How should the existence of frank presentations affect the diagnostic process? Table 5 summarizes these themes, along with several subthemes, along with illustrative quotations from survey respondents.

What is frank ASD?—27 respondents (51% of those who provided free responses) spontaneously discussed the nature of frank ASD in their comments. Ten respondents discussed the relationship between frank ASD and *severity*. Interestingly, clinicians did not agree on the direction of this relationship, with some clinicians reporting that they observe frank presentations more often among more impaired individuals, and others reporting frank

presentations to be more common in non-intellectually disabled cases. Furthermore, eight respondents spontaneously reported a relationship between clinical *experience* and recognition of frank ASD, with more experienced clinicians being more able / more likely to identify frank cases. Finally, four respondents suggested that frank features are often tied to either the *singularity* of the ASD in the particular individual, or the *specificity* of the ASD diagnosis. Clinicians endorsing this theme commented that frank features are most apparent in the least complex cases, for example, those lacking significant comorbidities or complicated differential diagnosis. Several mentioned that frank features are associated with a behavioral presentation that is *only* seen in ASD.

Frank ASD and the diagnostic process—29 respondents (55% of those who provided free responses) discussed how judgments of frank ASD might affect clinical practice, for better or worse. The majority of clinicians discussing process (69%) described how they proceed with their standard evaluation regardless of initial impression. Clinicians varied in the degree to which they attend to frank presentations, with a small number of clinicians reporting that they actively suppress or attempt to ignore these initial impressions. Several clinicians reported that when evaluating individuals with frank presentations, testing results are often more important for clinical needs such as assessing strengths and weaknesses, and communicating the diagnosis to families, than for establishing the ASD diagnosis per se.

Discussion

ASD is widely regarded as a heterogeneous condition with a wide variety of presentations. And yet, data from this study as well as our clinical experience suggest there is some aspect of the behavioral presentation that can be detected rapidly, and that it is highly specific to ASD. We sought to determine whether the idea of frank features would be endorsed by practicing diagnosticians, what they would include, and whether these features would correspond with current diagnostic criteria or level of functioning. We found that clinicians did endorse the idea of frank ASD, and notice it in about 40% of diagnosed patients. Our data also suggests that frank features do not correspond to level of functioning, and that some frank features are outside the current diagnostic criteria (e.g., posture and gait). We propose that a better understanding of frank ASD would inform both clinical practice and research, since frank features do not constitute the whole of diagnostic criteria, and since some possible frank features are not even included in diagnostic criteria.

We define a frank feature of ASD as a behavioral phenotype that gives rise to a rapid impression of ASD. Some frank features may be present continuously and thus would be immediately apparent upon meeting an individual (e.g., unusual prosody, which has been proposed as the most rapidly identifiable feature of ASD, Mesibov, 1992). Other behaviors may not be present continuously but once exhibited can lead to a rapid impression of ASD (e.g., repetitive motor behavior or speech). There may be a range of frank presentations based on different specific behaviors or clusters of behaviors.

In our data, the behaviors that stood out to the largest number of clinicians included: body mannerisms, repetitive language, repetitive behaviors generally, minimal social overtures, atypical social engagement, poor eye contact, and odd prosody. These are elements that have

long been reported in ASD (Kanner, 1943), but as broad categories none of them are specific to ASD alone. There are other disorders with social communication deficits, or with unusual behavior (e.g., internalizing disorders, externalizing disorders, psychosis). Even in combination with each other, it is not difficult to imagine patient scenarios where several of these behaviors are evident but the *quality* is not resonant with ASD. Thus, there is something particular about the quality of the poor eye contact, or the quality of the repetitive behavior, that distinguishes ASD.

Just as not all frank features of ASD are immediately apparent, not all features of ASD are likely frank. For example, struggling to adjust behavior to fit the social context is common to our conceptualization of ASD, but may not be apparent in a one-on-one clinical interaction with a kind and friendly evaluator. Likewise, subtle problems with nonverbal communication, emotional reciprocity, peer relationships, or behaviors and interests may only be appreciated over an extended period of time and across multiple contexts.

In our data, frank features did not correspond to a person's level of functioning—both frank and subtle features may be present at any level of either end of the cognitive continuum. We also hypothesize that frank features exist at both ends of the social continuum, or adaptive continuum. Individuals with frank features such as odd prosody and repetitive behaviors can be socially motivated, affectionate, caring, and independent. Some frank features (being highly aloof, or a particularly stiff or awkward social interaction style) may predict social impairments, but other frank features (odd prosody, unusual interests) may function as differences rather than deficits, depending on the individual and the social context. Some frank features may never change with intervention (e.g., prosody), while some frank features may or may not change with intervention (e.g., very limited social responding). This may suggest that some frank features exist independently from skill development, and others are tied closely to it.

We suspect that other psychiatric disorders, particularly those associated with distinctive social-communicative styles, may also have specific frank presentations, consisting of rapidly observable quantitative and qualitative features of behavior. For example, some patients with depression or schizophrenia might present with an interactive style that is readily apparent to experts, and which contributes to the diagnostic process. Describing behavioral qualities, rather than simple presence/absence, is consistent with the field of behavioral phenotyping, and has been used to try and elucidate the relationship between biology and behavior in other disorders. For example, a characteristic pattern of eye gaze avoidance has been described in Fragile X syndrome (Wolff, Gardner, Paccia, & Lappen, 1989), linking behavior to genetics, and distinctive aprosodias have been described in right hemisphere stroke (Ross & Monnot, 2008), linking behavior to anatomy. We propose that the behavioral features that give rise to impressions of frankness are highly specific to the ASD phenotype, and would be easily distinguishable from frank features of other disorders. More research is needed to test this prediction, and to test whether frank features of ASD change as function of other clinical features, such as age, level of functioning, and co-occurring diagnoses.

Many clinicians reported that they notice frank features quickly, often in ten minutes or less. Further, of those who reported using a term for the experience of noticing frank features in their clinical practice, over one third provided a term suggestive of rapid impression formation, such as within the time frame of the initial greeting or interaction in the waiting room. Several clinicians pointed out the risks of false first impressions, including halo effects (Nisbett & Wilson, 1977) wherein initial impressions of frank features might lead a clinician to see every behavioral atypicality through the lens of ASD, and confirmation bias (Nickerson, 1998), where one goes looking for evidence to support an established theory (in this case, that a patient has ASD), while ignoring incompatible or missing information that might contradict that theory. However, none of the open-ended responses suggested that clinicians are actually experiencing a significant number of "false positive" experiences, wherein frank features are apparent in the first few minutes, but then clearly explained by other factors that are uncovered over the course of the evaluation. Future work can investigate this more specifically by studying the relationship between behavior in the first few moments of interaction and the overall diagnostic conclusion.

One theme that arose from clinicians' freeform comments was that experience matters when it comes to detecting frank presentations. Consistent with this theme, reliability of clinically-assigned diagnoses of autism are poor for pairs of inexperienced clinicians (kappa = .34) despite being excellent for pairs of experienced clinicians (kappa = .94; Klin, Lang, Cicchetti, & Volkmar, 2000). In Klin et al. (2000), clinically-assigned diagnoses were made by clinical impression, and *not* by completing a DSM checklist. Interestingly, reliability among inexperienced clinicians improves with the use of DSM-IV symptom checklists, while reliability among experienced clinicians gets worse. This suggests that attending to individual symptoms is especially helpful for less experienced clinicians who are learning what to look for. Eventually, clinicians may internalize those symptoms, as well as other factors relevant to the construct of ASD (e.g., frankness, information relevant to differential diagnosis) and make diagnostic conclusions that go beyond what is explicitly described in DSM. We speculate that clinicians form prototypes of ASD that they refer to when evaluating a patient. With experience, these prototypes become reified, and clinicians become more confident in them. This phenomenon could help explain recent findings that clinicians *within* sites are more likely to agree on an ASD diagnosis than clinicians *across* sites (Lord et al., 2012): prototypes are formed and strengthened based on the types of referrals seen at each clinic, and consultations between close colleagues. This could also reflect a form of observer drift (Kazdin, 1977).

Implications for Clinical Work and Research

Our results suggest that diagnosticians identify frank features in about 40% of the individuals they evaluate. Thus, there is something specific, at least in a significant segment of individuals with ASD, that leads to a rapid impression of autism. However, current diagnostic and research practices generally combine all individuals with ASD into a single group. So, what exactly are we studying when we combine heterogeneous individuals into one "ASD" group, and what might we be *failing* to study by not going deeper into the most frank cases?

A better understanding of frank features of autism may help both clinical endeavors and research. In the clinic, if frank features are indeed pathognomonic, we could create more efficient diagnostic tools and processes that give greater weight to frank features. We could also modify our clinical training programs. It may be more useful to help junior clinicians distinguish frank features that can be identified rapidly and are specific to ASD, from non-frank features which are specific to ASD but can only be appreciated when understood across time and multiple contexts, and from behaviors that could contribute to an ASD presentation but may not be specific to ASD.

At the level of clinical screening, efforts at rapid screening for ASD by parent-report questionnaires tend to have higher sensitivity than specificity (Charman et al., 2007; Warren et al., 2012); however, when clinicians are asked to make rapid ASD referral decisions for preschool age children (i.e., based on 10-minute observations), specificity for ultimate ASD diagnosis is higher than sensitivity (i.e., 0.82 vs. 0.61; Gabrielsen et al., 2015). Perhaps if frank features can be formalized and quantified, they can be better incorporated into screening measures that yield stronger overall diagnostic classification power.

In research, frank features may be biobehavioral markers worthy of study either as independent mechanisms or as predictors of course, etiology, or response to treatment. For example, a narrow frank feature (e.g., active avoidance of eye contact, or unusual prosody) may be more proximal to a biological mechanism than a broad ASD feature (e.g., difficulty reading social cues). Perhaps it would be more effective to follow the trail of a particular frank feature rather than to attempt to map the full landscape of ASD. This field of study will depend on highly precise quantification of behavior to determine how people with ASD may cluster together behaviorally; this has proven difficult thus far (e.g., in the study of prosody, Fusaroli, Lambrechts, Bang, Bowler, & Gaigg, 2016), and will remain a challenge for the field.

Limitations and Future Directions

This study was an initial attempt to gather a cross section of impressions from practicing diagnosticians in order to develop testable hypotheses for future studies. Thus, it is limited by the fact we do not have extensive information about the specific training, setting, or types of referrals (e.g., age, IQ, co-occurring conditions) seen by the respondents. We also did not gather data on the actual outcomes of the diagnostic process. Future work can refine the conceptual basis and measurement of frank ASD, and elucidate the accuracy of frank features vis-a-vis ASD diagnosis. It can also work to understand if and how frank features of ASD are conceptualized by non-diagnosticians, such as treatment providers and educators, and whether the presence of frank features impacts the behavior of a social or communicative partner. Impressions of frank ASD could affect treatment and educational decisions made by professionals, for better or worse. Future research can address how perceptions of frank ASD affect practices such as teaching style and classroom placement (e.g., inclusion vs. ASD-specific), and how this ultimately affects outcomes for children with frank presentations.

Future work can also shed light on which frank features of ASD are differences to be appreciated as part of human diversity, and which are functional impairments we can address to help individuals reach their potential.

Acknowledgments

We thank the many ASD clinicians who generously gave their time to complete the frankness survey. Thanks to the many wonderful people at the Center for Autism Research at the Children's Hospital of Philadelphia, in particular the clinicians who provided feedback on the survey, and Armen Bagdasarov, for assistance with data collection and coding. We also thank Drs. Sally Ozonoff and Benjamin Yerys who provided helpful feedback on the manuscript, as well as three anonymous reviewers and an associate editor at this journal. This project was (in part) supported by Award number T32NS007413 from the National Institute of Neurological Disorders and Stroke (NINDS). The content is the sole responsibility of the authors and does not necessarily represent the official views of the NINDS of the National Institutes of Health.

References

Charman T, Baird G, Simonoff E, Loucas T, Chandler S, Meldrum D, Pickles A. Efficacy of three screening instruments in the identification of autistic-spectrum disorders. The British Journal of Psychiatry. 2007; 191(6):554–559. [PubMed: 18055961]

Creswell JW, Klassen AC, Plano Clark VL, Smith KC. Best practices for mixed methods research in the health sciences. Bethesda (Maryland): National Institutes of Health. 2011:2094–2103.

de Marchena A, Eigsti IM. Conversational gestures in autism spectrum disorders: Asynchrony but not decreased frequency. Autism Research. 2010; 3(6):311–322. [PubMed: 21182208]

Fusaro VA, Daniels J, Duda M, DeLuca TF, D'Angelo O, Tamburello J, Wall DP. The Potential of Accelerating Early Detection of Autism through Content Analysis of YouTube Videos. PloS One. 2014; 9(4):e93533. [PubMed: 24740236]

Fusaroli R, Lambrechts A, Bang D, Bowler DM, Gaigg SB. Is voice a marker for Autism spectrum disorder? A systematic review and meta-analysis. Autism Research. 2016 n/a-n/a. http://doi.org/10.1002/aur.1678.

Gabrielsen TP, Farley M, Speer L, Villalobos M, Baker CN, Miller J. Identifying Autism in a Brief Observation. Pediatrics. 2015; 135(2):e330–e338. [PubMed: 25583913]

Grossman RB. Judgments of social awkwardness from brief exposure to children with and without high-functioning autism. Autism. 2014; 19(5):580–587. [PubMed: 24923894]

Kazdin AE. Artifact, bias, and complexity of assessment: The ABCs of reliability. Journal of Applied Behavior Analysis. 1977; 10(1):141–150. [PubMed: 16795543]

Klin A, Lang J, Cicchetti DV, Volkmar FR. Brief report: Interrater reliability of clinical diagnosis and DSM-IV criteria for autistic disorder: Results of the DSM-IV autism field trial. Journal of Autism and Developmental Disorders. 2000; 30(2):163–167. [PubMed: 10832781]

Lord C, Petkova E, Hus V, Gan W, Lu F, Martin DM, et al. A multisite study of the clinical diagnosis of different autism spectrum disorders. Archives of General Psychiatry. 2012; 69(3):306–313. [PubMed: 22065253]

Lord, C., Rutter, M., DiLavore, PC., Risi, S. Autism Diagnostic Observation Schedule (ADOS). Los Angeles: Western Psychological Services; 2002.

Mattison R, Cantwell DP, Russell AT, Will L. A comparison of DSM-II and DSM-III in the diagnosis of childhood psychiatric disorders: II. Interrater agreement. Archives of General Psychiatry. 1979; 36(11):1217–1222. [PubMed: 485779]

Nickerson RS. Confirmation bias: A ubiquitous phenomenon in many guises. Review of General Psychology. 1998; 2(2):175–220. http://doi.org/10.1037/1089-2680.2.2.175.

Nisbett RE, Wilson TD. The halo effect: Evidence for unconscious alteration of judgments. Journal of Personality and Social Psychology. 1977; 35(4):250–256.

Ross ED, Monnot M. Neurology of affective prosody and its functional–anatomic organization in right hemisphere. Brain and Language. 2008; 104(1):51–74. [PubMed: 17537499]

Tong A, Sainsbury P, Craig J. Consolidated criteria for reporting qualitative research (COREQ): a 32-item checklist for interviews and focus groups. International Journal for Quality in Health Care. 2007; 19(6):349–357. http://doi.org/10.1093/intqhc/mzm042. [PubMed: 17872937]

Warren Z, Vehorn A, Dohrmann E, Nicholson A, Sutcliffe JS, Veenstra-VanderWeele J. Accuracy of phenotyping children with autism based on parent report: what specifically do we gain phenotyping "rapidly"? Autism Research. 2012; 5(1):31–38. http://doi.org/10.1002/aur.230. [PubMed: 21972233]

Wolff PH, Gardner J, Paccia J, Lappen J. The greeting behavior of fragile X males. American Journal on Mental Retardation. 1989; 93(4):406–411. [PubMed: 2522786]

Appendix A: Frankness Questionnaire

A. Introduction

Some clinicians with expertise diagnosing autism/ASD report that "you know it when you see it," a phenomenon we refer to as *frankness* (i.e., frankly ASD). We are interested in understanding this phenomenon from the clinician perspective.

Thank you for taking the time to improve our understanding of frankness. Please answer the following questions to the best of your ability.

1 Have you ever experienced the "you know it when you see it" frankness phenomenon?

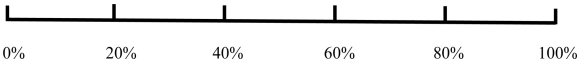
- Yes
- No* *(If respondents answered "No", they were taken to section C: "Comments")

B. Frankness Questions

2 Select the groups from which you have seen frank presentations of ASD (check all that apply):

- Younger, less verbal children
- Younger, more verbal children
- Older, less verbal children or adults
- Older, more verbal children or adults

3 Of the children and adults you have diagnosed with ASD, approximately what percent do you consider frank? (Please mark the line)



4 When a child is frankly ASD, how quickly do you form that impression?

5 Do you have a term for this phenomenon (even if used only informally)? If so, what?

6 What patient behaviors are most likely to contribute to you impression of frankness?

C. Comments

(Please provide any comments about frankness):

D. Information on Respondent

7 Discipline:

- Clinical Psychology
- Developmental-Behavioral Pediatrics
- Psychiatry
- Neurology
- Other:

8 In what state (or country, if not US) do you practice?

9 Years of experience diagnosing ASD (including training):

10 Approximate number of ASD diagnoses you have given:

11 Do you consider yourself an ASD researcher in addition to a clinician?

a. Yes

b. No

12 If you are willing to be contacted about future research on this topic, please provide contact information (OPTIONAL):

Table 1

Clinicians endorsed whether they had seen frank cases in the following four subgroups: Younger, less verbal children; Younger, more verbal children; Older, less verbal, children and adults; and Older, more verbal, children and adults. Proportion of total clinicians who endorsed seeing a frank case from each subpopulation is reported here.

	Less verbal	**More verbal**
Younger children	82%	62%
Older children and adults	78%	67%

Table 2

Clinician estimates of the percentage of people with ASD who have a frank presentation.

	Mean	Median	Mode
Entire sample	40%	31%	19%
More experienced clinicians	42%	37%	30%
Less experienced clinicians	38%	29%	19%

Mean, median, and mode are all presented (Note: sample skewness = 0.46) for the entire sample, as well as the sample split by experience. “Less experienced clinicians” self-reported having given 250 ASD diagnoses or less, while “more experienced clinicians” self-reported having given at least 300 ASD diagnoses.

Table 3

Spontaneously provided responses on how rapidly frankness impressions are made.

Speed of Impression	Percent of respondents (cumulative)
Within seconds	3%
Within 1 minute	8%
Within 5 minutes	37%
Within 10 minutes	52%
Within 15 minutes	64%
Within 20 minutes	74%
Within 30 minutes	87%
Within 60 minutes	94%

Note that categories are cumulative, e.g., “Within 1 minute” includes all participants who responded that they make frankness judgments within 1 minute, as well as all respondents who form impressions within seconds.

Table 4

Spontaneously reported behaviors associated with frankness.

Category	Subcategory	Sample response	% of responses
Repetitive Behaviors/Interests			**35.2**
	Motor mannerisms	"Stereotypies"	9.0%
	Repetitive use of language	"Echolalia/scripting"	7.6%
	Stimming/Sensory seeking	"Self-stimulatory behaviors"	4.4%
	Restricted interests	"Narrow interests"	4.2%
	Repetitive object use	"Lining up toys/items"	1.1%
	RBI – other/general	"Repetitive and ritualized behavior"	8.9%
Social Reciprocity			**26.7%**
	Amount of interaction	"No social overture"	10.8%
	Quality of interaction	"Atypical social engagement"	9.0%
	Conversation skills	"Really extensive monologues"	3.1%
	Response to name	"Little response to name"	2.3%
	Reciprocity – other/general	"Social skills"	1.5%
Nonverbal Communication			**22.3%**
	Eye Contact	"Poor eye contact"	10.0%
	Prosody	"Voice quality"	7.8%
	Facial expressions	"Flat affect"	1.8%
	Joint attention	"Response to joint attention"	1.5%
	Nonverbal – other/general	"Gestures (lack of)"	1.3%
General Motor Behavior		**"Awkward movements"**	**2.9%**
Other			**12.9%**
	ASD-related	"Rigid and concrete thinking"	3.7%
	General psychiatric / DD	"Inconsistent hygiene"	5.2%
	Unrelated/vague	"Language"	4.0%

Table 5

Themes extracted from free-form comments.

Theme	Subtheme	Illustrative quotation
What is frankness?	Frankness and severity *and* Frankness and specificity	“It's really not the severity, it's those telltale signs that are really only seen in ASD that trigger 'frankness' for me.”
	Detection of frankness improves with experience	“As I get more experienced in diagnosis, this occurs more and more frequently.”
How should frankness affect diagnostic process?	Recognize frankness but do not change diagnostic process	“Although I have seen many cases of "ASD frankness" I still do a complete evaluation before making a diagnosis.”
	Frankness as a hypothesis to be tested	“It is still important to rule out less common explanations for the same behavioral presentation. As scientists, the "frankness" of the presentation lends support to the hypothesis that the individual has ASD, but it is up to the clinician to look for evidence to refute that hypothesis.”
	Clinicians should be careful not to jump to conclusions	“I think as a clinician it is important to recognize your gut impression when you first meet a child/family, but not to let that be your final impression.”