Assessing Vulnerability to Schizophrenia or Manic-Depression in Borderline States*

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Abstract

In a discussion of the article on genetic determinants of borderline conditions by Siever and Gunderson, a phenotypic continuum between pure schizotypal and pure affective conditions is postulated. Many “borderline” cases are seen as attenuated forms of schizophrenia, schizoaffective psychosis, or manic-depression. A Venn diagram illustrates differences among syndromes described by Gunderson, Kernberg, Spitzer, and Klein (“hysteroid dysphoria”). Evidence is presented suggesting that Gunderson’s borderline syndrome contains more schizotypal individuals than Kernberg’s, whereas hysteroid dysphoria is nearer the affective pole of the continuum. A second diagram illustrates how the strength and nature of the genetic factors vary according to the syndrome.

Surrounding the universally accepted categories in our psychiatric nomenclature—schizophrenia and primary affective disorder—is a larger interstellar space of “borderline conditions.” Though many authors have offered descriptions of an imprecise nature, some have attempted to define these conditions in a rigorous fashion (Gunderson and Kolb 1978; Kernberg 1977; Klein 1977). The questions arise: (1) Do the conditions currently being labeled borderline represent fragments of psychopathology still affiliated, though at some distance, to the parent conditions—or are they truly “separate”? (2) Is there a subset of borderline conditions that are genetically related to the major psychoses—alongside another subset where no such connection is discernible? (3) If there is a subset related to clear-cut schizophrenia, schizoaffective psychosis, or to the affective psychoses—does this represent a majority of borderline cases, or only a small portion?

The painstaking and methodical review of Siever and Gunderson (1979) addresses itself to all these questions; their work represents an important step toward the demystification of this borderline region. As the authors make clear at the outset, there are two broad uses of the term “borderline”: one, genetic; the other, clinical. It is important to keep in mind, however, that this bipartite division does not in itself do justice to the large number of diagnostic labels, some embodying the word “borderline,” which currently are used to describe similar samples of patients. One needs to be familiar with all these labels, and with the typical patient samples they designate, lest the adherents of one school of thought misunderstand the clinical findings and hypotheses put forward by another. Perry and Klerman (1978), having recently examined in detail the classificatory systems of Grinker, Knight, Kernberg, and Gunderson (all of whom use the term “borderline”), incline toward the view that these various usages are not interchangeable. Instead, they overlap in certain particulars, but in many other respects remain distinct as subtypes within the general realm of borderline conditions. Elsewhere (Stone, in press) I have compared

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four contemporary systems in which an attempt at precise definition has been made: Gunderson’s borderline personality disorder, Kernberg’s borderline personality organization, Klein’s hysteroid dysphoria, and Spitzer’s borderline personality. Again, certain items (impulsivity, impaired object relations) are common to those four sets of descriptions; other items (histrionic, abuse of drugs, chronic emptiness) are emphasized by only one system.

As Siever and Gunderson mention, the Spitzer checklist of borderline personality items (Spitzer, Endicott, and Gibbon 1979) evolved out of an effort to refine the concept of borderline schizophrenia, as used by psychogeneticists to denote a penumbral region around a central “core schizophrenia”—where both borderline and core cases were expressions of the same genotype (cf. Kety et al. 1968). The Spitzer checklist comprises items that can be differentiated into two broad subtypes: the “schizotypal” and the “unstable.” The former answers to the borderline schizophrenia (B3 type) of Kety et al. The unstable items tend, on the other hand, to single out a patient sample with many abnormalities in the affective realm (e.g., mood lability, self-destructive acts, inappropriate anger). From a psychogenetic standpoint, many unstable borderlines appear to be borderline with respect to primary affective disorder (or, if you will, to manic-depressive illness in all its forms) in much the same way that the schizotypal group is related to core schizophrenia. There is probably less specificity with respect to the unstable group than in the realm of schizophrenia, because depression would appear to be, etiologically, more heterogeneous than mania and schizophrenia (for which hereditary factors may well be necessary—though-not-sufficient antecedents). Evidence for a genetic factor in at least some cases of depression is growing, not only from twin study results (Zerbin–Rüdin 1968), but also from a recent study of adoptees with depressed biologic mothers (Cadoret 1978).

If one thinks of all patients who exhibit enough positive items on the Spitzer checklist to qualify as borderline in this schema, it should be clear that the domain embraces the Kety B3 borderline schizophrenia (i.e., the “spectrum” cases), many spectrum cases of the affective disorders, some mixed types, and, in addition, a certain number of patients whose conditions may not be etiologically related to liability to either classical psychosis to any appreciable extent. This represents, to my way of thinking, the broadest usage of the term “borderline” where the criteria are still quantifiable.

The domain of Kernberg’s (1977) borderline personality organization is smaller than, and included within, the Spitzer domain. Phenotypically, it occupies a region that is tilted toward the affective pole and away from the schizophrenic pole (Stone 1977). In the psychostructural language of Kernberg’s model, the borderline patient shows the capacity to test reality (cf. Frosch 1964), even though he may start out with a faulty relationship to reality. If a patient, free of gross delusory ideation, nonetheless holds rigid, contradictory, and highly unrealistic views about himself or about the important people in his intimate world, and if, furthermore, these ideas do not yield on confrontation by the interviewer, then the capacity to test reality is seriously impaired, and the patient is said to exhibit psychotic (rather than borderline) structure. It so happens that many borderline schizophrenics (à la Kety et al.), while free (by definition) of productive signs (i.e., delusions and hallucinations) do hold tightly to certain unrealistic impressions within the sphere of object relations; one sees this characteristic more often among borderline schizophrenics than among spectrum cases of manic-depressive illness. Hence, in the Kernberg domain, borderline psychic structure (or borderline personality organization) overlaps with borderline schizophrenia only in a few instances. These tend to involve highly intelligent schizotypal persons whose distortions in the interpersonal realm can be corrected without inordinate difficulty on the part of the examiner. Hypomanic patients are seldom chosen for analytically oriented psychotherapy because they do not usually respond well. This leaves, almost by exclusion, a large number of individuals with significant depressive pathology—among those to whom the Kernberg label would apply. Though the percentage will vary from sample to sample, a significant proportion of the structurally borderline patients have full-blown or spectrum affective disorders. Some Kernberg borderline patients, when first seen, do not have enough affective symptoms (especially the more “vegetative” ones) to permit confident placement within the manic-depressive core or spectrum. A number of these patients, if followed long enough, have, in my experience, gone on to develop unmistakable signs of such a disorder. The presence of unipolar or bipolar relatives...
(first or second degree) may heighten the suspicion one is dealing with a spectrum affective disorder, but is not in itself a diagnostic sign. It should be underlined that borderline patients (of whatever diagnostic schema) are often young; many are adolescents. As such, they have scarcely entered—let alone passed through—the age of risk for either of the classical psychoses. It is therefore hard to distinguish, when they first come to our attention, which cases owe their borderline pathology (in large part) to genetic liability and which develop their disorder in response to non-genetic factors. In this connection it is of interest to note that of 24 "undiagnosed" adolescents followed up by Fard, Hudgens, and Welner (1978) 7 years after initial evaluation, six received a psychiatric diagnosis: one schizophrenic, two schizoaffective, and three manic-depressive disorders.

The Gunderson (Gunderson and Kolb 1978) schema overlaps in many features with that of Kernberg. Both emphasize impulsivity and disordered object relations; both allow for the presence of brief psychotic episodes. The Gunderson criterion set does not insist, however, upon the capacity to test reality in the interpersonal realm in quite the same way emphasized by Kernberg. Therefore, certain Kety B3 borderline schizophrenics, who would be excluded by Kernberg, might be included in Gunderson’s system. Contrariwise, certain alcoholics, sociopaths, etc., who exhibit borderline “structure,” fail to satisfy some of the Gunderson items (e.g., good socialization). The net result of these differences is that the Gunderson domain encompasses a smaller set of patients than that of Kernberg, although the former has a thin “tail” into the region of borderline schizophrenia. This latter point may make more comprehensible the effort of Siever and Gunderson to review in such depth the results of the twin, adoption, and other genetic transmission studies of schizophrenia. There are enough Gunderson borderline patients who are reminiscent of the Kety borderline schizophrenic group to have justified this focus,1 whereas to adherents of the Kernberg system such an effort would appear somewhat irrelevant.

Hysteroid dysphoria, described by Klein (1977), defines a patient sample situated within the larger borderline domains of Spitzer and Kernberg. As the phrase suggests, the syndrome is characterized, within a three-dimensional framework, by depression (constitutional), borderline organization (functional), and histrionic personality traits (characterological). Restrictively defined in this way, hysteroid dysphorias are clustered on a phenotypic continuum between schizophrenia and manic-depression; they are seldom schizotypal. In a consanguinity study of 10 patients who satisfied Klein’s criteria (Stone, unpublished data), I noted the following: nine were female; the age range was 21 to 45; five had been hospitalized (once each); there were 39 first degree relatives (including three children older than 20) of whom 17 had serious psychiatric disturbances. These included borderline (B3) schizophrenia (2), predominantly affective schizoaffective disorders (3), bipolar affective illness (1), recurrent depression (7), and alcoholism or other addiction (4). In two of the families there were no first degree relatives with a disorder related to the classic psychoses. This is a small series, in which only 11 of the relatives were interviewed personally; nevertheless, the preponderance of affectively ill relatives suggests that hysteroid dysphoria is a variety of borderline condition closely allied in many instances to the primary affective disorders. These patients also satisfied the Gunderson and Kernberg criteria.

In trying to grasp the relationships among the currently popular borderline systems, it is useful to visualize a phenotypic continuum, with “pure” schizophrenia at one end, “pure” manic-depression at the other. Along this continuum, there are not only the psychotic cases but also forms frustes or borderline cases at each point along the way. If one focuses on the borderline realm of such a clinical universe, the loci of the range of patients within each system may then be mapped within this realm. Figure 1 depicts such a mapping. Not indicated in this diagram is an area assigned to patients without any discernible genetic connection to the classical psychoses. A certain proportion of borderline cases (the percentages are unknown and might differ for each system), as Siever and Gunderson suggest, would occupy this area. Siever and Gunderson imply that this proportion could be quite large, whereas I feel it is likely to be small.

If the psychiatrically ill first and second degree relatives of each borderline type were then analyzed

1 See Siever and Gunderson’s statement on p. 82, “... the psychotic-like characteristics Spitzer, Endicott, and Gibbon define as schizotypal are unlikely to demarcate a new diagnostic entity discriminable from borderline personality disorder.”
Figure 1. Location of various borderline domains

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Functional level: P = psychotic; B = borderline (the domain of all patients with six or more items on the Splitter Borderline Checklist); N = neurotic.

Regions: I = pure schizophrenic and schizotypal disorders; II = predominantly schizophrenic schizoaffective; III = schizoaffective; IV = predominantly affective schizoaffective; V = primary affective disorders ("pure" type).

Borderline domains: 1 = borderline schizophrenia; 2 = borderline personality disorder (Gunderson); 3 = borderline personality organization (Kernberg); 4 = hysteroid dysphoria (Klein) (shaded area).

with respect to the continuum diagram of figure 1, the frequency distributions within each cell would probably differ. Siever and Gunderson have indicated how the ill relatives of schizotypal borderlines are spread out beyond just the region of core and borderline schizophrenics, yet still slant toward the schizophrenic pole. If a larger and more methodical study of the ill relatives of hysteroid dysphorics were to corroborate my initial impressions, these relatives would also be spread out over a certain range within the continuum, but would be slanted toward the affective pole.

Figure 2 shows how the curves for the affected relatives of different groups of borderline index cases might be drawn. A curve for chronic schizophrenic probands is included for purposes of comparison. Though figure 2 has been constructed only from preliminary data and impressions, it can serve as a hypothesis to be tested in future consanguinity studies. If a similar set of curves emerges, this would tend to confirm the notion that the current borderline systems represent patient samples which, while partially overlapping, are also partially distinct and, in some instances (Kety et al. vs. Klein), almost noncontiguous.

Siever and Gunderson make clear that the "borderline" region around core schizophrenia is better delineated than is the corresponding region surrounding primary affective illness because the family, twin, and adoptive studies are in a higher state of development within the schizophrenic realm. It is precisely because the adoptive studies are so impressive that one can no longer deny the importance of the hereditary factor in schizophrenia. This, in turn, has emboldened others of us to work backwards from borderline index cases toward the core(s) to which they may (or may not) be related. Before the adoption studies, there was no convincing way of (1) challenging the strict environmentalist's claim that variations in nurture accounted for all variation in clinical state, or (2) suggesting the psychiatric morbidity in the offspring of mentally ill parents might be a reflection, at least in part, of genetic factors as well as of rearing. After these studies it became possible to argue, for example, that given schizophrenia in a parent, borderline schizophrenia in any of the offspring might stem as much, or more, from genetic transmission than from the pattern of rearing, even where the affected child was reared by the affected parent.

Siever and Gunderson's careful analysis of the schizophrenia studies led them to conclude that borderline (including schizotypal) cases probably have less genetic loading for schizophrenia than core cases. Because the borderline probands
Figure 2. Probable loci of affected relatives of various borderline index cases

Index Cases: 1. Borderline schizophrenia (Kety et al., B3 type); 2. Hysteroid dysphoria (Klein); 3. Borderline personality disorder (Gunderson); 4. Borderline personality organization (Kernberg); 5. Borderline personality: schizotypal and unstable (Spitzer).

Comparison cases: X. Chronic schizophrenia.

had some borderline—but very few core schizophrenic—relatives, they even appeared to "breed true" rather than to be linked primarily to schizophrenia. The authors mention, however, that nonspecific factors may be present that modify the clinical manifestations in the

It is possible that the apparent tendency to breed true relates to the existence of pedigrees in which both genetic loading for a psychosis and favorable modifiers are passed down from one generation to the other, such that all the affected members appear borderline.

At present we cannot determine with precision, in many borderline cases, whether a genetic factor is applicable and, if so, what is its relative strength in the causal hierarchy. Siever and Gunderson, appraising this factor according to the best available hard data, represent a conservative viewpoint. They could be underestimating its significance. My tendency would be to err on the high side. I can imagine, for example, that there are borderline cases with only modest genetic loading, but also a few with fairly high loading—where the favorable modifying factors happen to be unusually strong. One could envision a gray area where modest loading, in the presence of favorable factors, might fail to push an individual beyond the threshold for a borderline condition. Subjects of this sort would not show up in our hard data; their recognition is, admittedly, largely an act of faith, stemming from very subtle clinical cues. It also seems possible for some borderline cases to have arisen out of liability for both schizophrenia and primary affective disorder. A continuum of genotypes may exist alongside the continuum of schizophrenic and affective mixtures that we see clinically (cf. Stabenau 1977). I have worked with a number of borderline patients in whom core or borderline schizophrenia was present in one parental line; primary affective disorder, in the other. The clinical state of the patients represented something intermediate between the two classical psychoses as well as something in between neurosis and psychosis. It is difficult to know what inferences can be drawn from such examples because, as Siever and Gunderson point out, there is a large problem in ascertainment: It is easy to select cases in accordance with whatever theory one favors. The time seems ripe now for carefully controlled consanguinity studies on large samples of rigorously defined borderline patients. It is to be hoped that such studies would be carried out with the elegance of the adoption and twin studies reviewed by Siever and Gunderson, and then evaluated with the same meticulousness demonstrated by the authors in their review.
References


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