Relationship of Schizotypal Personality Disorder to Schizophrenia: Genetics

by Svenn Torgersen

Abstract

The adoptive, family, and twin studies show that schizotypal personality features are found among the relatives of schizophrenics. However, it has not been shown that there is a higher risk of schizophrenia among the relatives of schizotypals. An explanation may be that the current DSM-III criteria of schizotypal personality disorder do not adequately define schizotypals genetically related to schizophrenia. While some of the cases that meet DSM-III criteria are within the schizophrenia spectrum, others are unrelated to schizophrenia. There is reason to believe that schizotypals characterized by distant relationship to others, suspiciousness, eccentricity, peculiar communication, and dysfunctional school and work performance are within the schizophrenic sphere, while individuals with psychotic-like symptoms phenomenologically similar to schizophrenia and diagnosed as schizotypal personality disorders in DSM-III represent decompensation of other personality disorders.

Asking whether any genetic relationship exists between schizotypal personality disorder and schizophrenia might seem superfluous, considering the history of the concept. Earlier used terms—“ambulatory schizophrenia,” “latent schizophrenia,” “pseudo-neurotic schizophrenia,” and “borderline schizophrenia”—all imply that the disorder is a milder, covert, or masked variant of schizophrenia.

The term “schizotypal” indicates even more strongly the common genetic basis of schizotypal personality disorder and schizophrenia, as the term is an abbreviation of “schizophrenic phenotype.” Rado (1956), who first used the term, chose it to indicate that the disorder is a phenotypic representation of a genotype common to schizophrenia. Meehl (1962), who borrowed the term from Rado, did so because he believed that the “schizotypes” shared the “schizotaxic” endowment with schizophrenia.

There is reason to believe that the American Psychiatric Association’s Task Force on Nomenclature and Statistics held the same view when they decided to apply the term “schizotypal personality disorder” to the milder schizophrenic-like syndrome that has been designated by varying labels throughout the nosological history of psychiatry. They state:

There is some evidence that chronic schizophrenia is more common among family members of individuals with schizotypal personality disorder than among the general population. [American Psychiatric Association 1980, p. 312]

Furthermore, an empirically derived relationship to schizophrenia was expected since the criteria for the diagnosis of DSM-III schizotypal personality disorder are derived from the case records of the Danish extended family study (Kety et al. 1968; Spitzer, Endicott, and Gibbon 1979). The extended family study was intended to demonstrate the importance of genetic factors in the development of schizophrenia by studying the biological relatives of schizophrenic adoptees. Many of these relatives had a syndrome called “borderline schizophrenia,” and the criteria for schizotypal personality

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disorder were developed by studying the case records of these cases of borderline schizophrenia. (See Kendler’s review in this issue for an extensive discussion of the development of the diagnosis of schizotypal personality disorder.) Thus, it might seem self-evident that schizotypal personality disorder is genetically related to schizophrenia. However, a concept does not necessarily cover what it is intended to, and in this article the evidence of a genetic relationship between schizotypal personality disorder and schizophrenia is critically examined.

Adoptive Studies

As noted above, the DSM-III criteria for schizotypal personality disorder (SPD) are derived from the Danish adoptive studies of schizophrenia. Siever and Gunderson (1979) have thoroughly reviewed these studies and their implication for borderline disorders in general. Accordingly, I only briefly outline their implications for the topic under discussion.

The Danish adoptive studies are of two kinds—the extended family study (Kety et al. 1968) and the adopted-away study (Rosenthal et al. 1968). The extended family study’s point of departure was 34 schizophrenic adoptees and a control group of nonschizophrenic adoptees. The diagnosis of schizophrenia was divided into chronic schizophrenia, acute schizophrenia, and borderline schizophrenia. The results showed a higher prevalence of chronic and borderline schizophrenia among relatives of the index adoptees than among relatives of the control adoptees (Kety et al. 1976). Of interest here, however, is the relationship between borderline schizophrenia and other disorders within the schizophrenia spectrum. Siever and Gunderson (1979) reanalyzed the sample from this point of view. Since acute schizophrenia did not show any genetic relationship to the other schizophrenic disorders, they examined the relationship between chronic and borderline schizophrenia. Their analysis showed a statistically significantly higher prevalence of borderline schizophrenia among the relatives of chronic schizophrenic adoptees compared to relatives of controls. However, no chronic schizophrenia was observed among the relatives of borderline schizophrenic adoptees. Instead, they found a 13 percent prevalence of borderline schizophrenia, compared to 1 percent in the relatives of controls. Thus, biological relatives of chronic schizophrenic adoptees appear to have a higher prevalence of borderline schizophrenia, but biological relatives of borderline schizophrenic adoptees do not have a higher prevalence of chronic schizophrenia.

The adopted-away study used the opposite strategy compared to the extended family study. Here the starting point was schizophrenic (and manic-depressive) parents whose children had been adopted away (Rosenthal et al. 1968). Normal parents with adopted-away offspring made up the controls. When the offspring were interviewed, a higher prevalence of schizophrenia was found in the adopted-away offspring of index parents (Rosenthal et al. 1971). Siever and Gunderson (1979) again reanalyzed the observations to look at the relationship between chronic and borderline schizophrenia. They observed two borderline schizophrenics and one schizophreniform borderline among the 30 adopted-away offspring of chronic schizophrenic parents, but again no chronic schizophrenia among the four offspring of borderline schizophrenic parents.

Thus, the adopted-away study led to the same conclusion as the extended family study: If the index cases are chronic schizophrenic patients, one finds borderline schizophrenia in the relatives, but if one starts with borderline schizophrenic patients, one finds no chronic schizophrenia in the relatives. We have now examined studies of borderline schizophrenia. However, although the criteria for DSM-III SPD are derived from studies of borderline schizophrenia, SPD and borderline schizophrenia appear not to be the same.

Kendler, Gruenberg, and Strauss (1981) applied the DSM-III criteria for SPD to the interview records of the relatives in the Danish extended family study. The criteria were not applied to the adoptees. This study showed a moderate correspondence between the diagnosis of borderline schizophrenia and DSM-III SPD. While the specificity of SPD in relation to borderline (including uncertain borderline) schizophrenia was 98 percent, the sensitivity was only 31 percent (calculated by this author). The specificity decreased to 91 percent, and the sensitivity increased to 40 percent when uncertain borderline schizophrenia was excluded from the calculation. Both borderline and uncertain borderline schizophrenia thus seem to be more inclusive diagnoses than DSM-III SPD.

Kendler, Gruenberg, and Strauss (1981) found that 11 percent of the biological relatives of the schizophrenic adoptees had SPD, compared to none among the adoptive relatives. However, almost all the SPD cases were found among the biological relatives of the chronic schizophrenic adoptees. Only one (5 percent) was found among the
relatives of acute schizophrenic adoptees, and none among the relatives of borderline and uncertain schizophrenics.

The authors conclude: “Although based on a small number of relatives and hence best regarded as tentative, these results support a genetic link between SPD (schizotypal personality disorder) and chronic schizophrenia but not acute or borderline schizophrenia” (p. 984). They note that the criteria for SPD may select persons who are in a prodromal phase of schizophrenia, although this would be rare considering that the mean age of the relatives was 39 years old.

Another more important objection, which Kendler, Gruenberg, and Strauss (1981) also mention, is the circularity of their research design. The very criteria for DSM-III SPD were derived from the interviews in the Danish extended family study. Given this, it was noteworthy that only 9 of the 29 cases with borderline and uncertain borderline schizophrenia fulfilled the DSM-III SPD criteria. One reason may be that Kendler, Gruenberg, and Strauss (1981) were very restrictive in using the diagnosis of SPD. In a later article (Kendler and Gruenberg 1984), they state that when they were presented with a case with a mixture of psychotic and personality disorder features, their tendency was to apply a psychotic diagnosis rather than SPD. Their study is also limited by the fact that the adoptees were not evaluated by DSM-III SPD criteria. This was done, however, in the aforementioned recent study (Kendler and Gruenberg 1984) (reviewed below).

Gunderson, Siever, and Spaulding (1983) undertook another study of the interviews from the Danish extended family study. Two of the researchers blindly and independently reviewed 27 interview records of borderline schizophrenic cases and 27 records from the same study of other cases to identify schizotypals and borderlines. The main aim of their study was to investigate whether the borderline schizophrenic biological relatives of schizophrenic adoptees were different from index borderline schizophrenics or control borderline schizophrenics and how the SPD criteria were related to borderline schizophrenia. Schizotypal personality disorder was established by means of two sets of criteria, DSM-III and their own criteria. The main difference between the two definitions was that the research group focused more on affective impoverishment, asociality, and shallow relationships. By using their own criteria, they avoided some of the circularity that was found in the study by Kendler, Gruenberg, and Strauss (1981) even if they, of course, were influenced by the original Danish adoptive studies in developing the criteria.

When the diagnosis of schizotypal was compared with borderline schizophrenia, Gunderson, Siever, and Spaulding (1983) obtained a sensitivity of 59 percent and a specificity of 81 percent applying the DSM-III criteria, and a sensitivity of 52 percent and a specificity of 89 percent by use of their own criteria. Thus, as was also shown by the study of Kendler, Gruenberg, and Strauss (1981), the overlap between schizotypals and borderline schizophrenics is not that impressive, even if the criteria for SPD are derived from a study of borderline schizophrenia in the Danish adoptive studies.

Contrary to what would be expected, they found that fewer of the borderline schizophrenic biological relatives of chronic schizophrenic adoptees fulfilled the criteria for SPD than did the other borderline schizophrenic relatives, index cases, and controls.

Symptoms such as self-destructiveness, psychotic-like experiences, demandingness, and past psychiatric contacts were less common in borderline schizophrenic relatives of chronic schizophrenics than in borderline schizophrenic relatives of other index cases, index cases, and controls. The relatives of chronic schizophrenics who received a diagnosis of borderline schizophrenia in the original Danish study thus seem to have a less dramatic and severe symptomatology than other borderline schizophrenics in the same study.

Furthermore, they found that the borderline schizophrenic relatives had more dysfunctional social adaptation by being more often unemployed and eccentric, were more often interpersonally and affectively detached, more depressed and less unstable in affects and relationships, less impulsive, and had more somatic problems compared to individuals with another type of personality disorder, DSM-III borderline personality disorder (BPD).

These observations led Gunderson, Siever, and Spaulding (1983) to propose the following criteria for a schizotypal personality disorder possibly related to chronic schizophrenia: (1) social isolation and anxiety; (2) suspicious, superficial, and distant interpersonal relationships; (3) odd, eccentric, “off-putting” appearance and behavior; (4) frequent somatic problems; (5) detached, constricted, and flattened affect; and (6) serious social dysfunction at school and work.

The results of this study by Gunderson, Siever, and Spaulding (1983) are intriguing for advocates of a genetic link between SPD, as defined today, and schizophrenia, for many features of DSM-III SPD are
found more seldom among borderline schizophrenic relatives of chronic schizophrenics than among other borderline schizophrenic cases.

Khouri et al. (1980) used the other Danish adopted-away study of Rosenthal et al. (1968), as a starting point for identification of schizotypals genetically linked to schizophrenia. On the basis of the description by Kety et al. (1968) of borderline schizophrenia, they developed the Symptom Schedule for the Diagnosis of Borderline Schizophrenia. The schedule focused on perceptual and behavioral changes, with items such as "soft" hallucinations, altered perception, feelings of unreality, muddled thinking, ideas of reference and persecution, perverse sexuality, and violence. From their own clinical experience, they added self-inflicted injuries in the absence of suicidal depression. The schedule was applied to the interview records of 14 adoptees who had received the diagnosis of borderline schizophrenia in the original Danish adopted-away study (the index group) and 17 others with mixed nonpsychotic diagnoses, including 3 normals (the control group). Two researchers rated the interview records blindly and independently, obtaining an interrater agreement correlation of .83. Their scores were averaged, and with an appropriate cutting point, they claimed to have obtained a sensitivity of 79 percent and a specificity of 100 percent in differentiating the indexes from the controls.

The study by Khouri et al. (1980) does not prove anything about the genetic relationship between chronic and borderline schizophrenia, or between schizophrenia and SPD, because of the circularity of the design. However, contrary to the results of the study of Gunderson et al. (1983), the high sensitivity and specificity of their interview schedule might seem to demonstrate the preponderance of psychotic-like symptoms in the borderline schizophrenic adopted-away offspring of schizophrenics. However, the low intrarater agreement on individual symptoms weakens their results, as Gunderson, Siever, and Spaulding (1984) have stated.

Lowing, Mirsky, and Pereira (1983) have also applied DSM-III criteria to the adopted-away offspring in the study of Rosenthal et al. (1968). Their first step was to examine the interview protocols of the parents. Only parents who fulfilled DSM-III criteria for chronic schizophrenia, acute schizophrenia, borderline schizophrenia, and mixed schizophrenic symptoms were included. Thus, all parents with manic-depressive symptoms, other affective symptoms, or unclear psychotic symptom patterns were excluded. The same was true if there were doubts as to whether the schizophrenic "father" was the real biological father. In this way, the number of parents was reduced from 156 to 78 and the index adopted-away offspring to 39. Application of DSM-III criteria to the index offspring and their controls showed that six (15 percent) index compared to three (8 percent) control offspring had an SPD. Even if there is a difference between the prevalence of SPD in index and control offspring, the difference is not convincing. The authors mention that the results are similar if the analysis is restricted to the 29 offspring of chronic schizophrenics. However, they do not report results for this more restricted index group.

Recently, Kendler and Gruenberg (1984) sought to improve their earlier (1981) study by also applying DSM-III criteria to the index adoptees in the Danish extended family study. They found that 6 percent of the relatives of the schizophrenic adoptees had schizophrenia and 14 percent, SPD. Grouping schizophrenia, schizophrenic schizoaffective disorder, and SPD in a group of schizophrenia spectrum adoptees resulted in almost the same percentages—4 percent schizophrenia and 13 percent SPD in the relatives. However, none of the six relatives of SPD adoptees had schizophrenia, and two had borderline personality disorder.

It would appear that the adoptive studies have demonstrated that the biological relatives of schizophrenics have a higher risk of the rather vaguely defined syndrome called borderline schizophrenia, which only partially overlaps with DSM-III SPD. When borderline schizophrenics or subjects with SPD are index cases, however, chronic schizophrenia has not yet been observed among the biological relatives, though the number of such index cases remains small.

Another major problem with the studies is the circularity of the designs. The criteria for the diagnosis of SPD are derived from the same study in which its genetic relationship to schizophrenia is tested.

**Family Studies**

Adoptive studies are undoubtedly the best way to demonstrate the genetic link between two disorders. However, studies of relatives of probands reared in their biological families may also give some hints about the importance of genetic factors. Generally, such family studies are better able to disconfirm than confirm a genetic relationship. If the relatives of a proband with one disorder have a higher risk of another disorder, the observation
may be interpreted as evidence of an environmental as well as a genetic link between the two disorders. If, however, no higher risk of the other disorder is observed among the relatives, neither a genetic nor an environmental link may exist between the disorders. A few family studies aiming to disclose the relationship between schizotypals and schizophrenics have been performed.

Stone (1979) studied consecutive inpatient admissions to a New York psychiatric hospital. A psychostructural diagnosis was given according to Kernberg's (1977) criteria. The relatives of probands with a psychotic, a borderline, and a normal personality structure were compared. Stone mentions that eight of the probands with a borderline structure fulfilled criteria for SPD. These probands had no schizophrenic relatives, but there were two relatives with SPD. No SPD was found among the first-degree relatives of 13 DSM-III borderline personality disorder probands. Stone's study does not support a relationship between SPD and schizophrenia. However, the study indicates that SPD runs in families.

Soloff and Millward (1983) studied admissions to inpatient services in Pittsburgh during a 4-year period. The borderline sample was defined by Spitzer, Endicott, and Gibbon's (1979) criteria encompassing both DSM-III SPD and BPD. Patients suffering from unipolar, nondelusional depression and schizophrenia constituted two control groups. Family histories of psychiatric disorders were obtained through interviews with the patient and family informants, and through chart review. They found the prevalence of schizophrenia to be the same in the relatives of SPD, BPD, and depressive patients, and about half that in the relatives of schizophrenics. On the other hand, eccentric/peculiar behavior was far more common among the relatives of SPD and BPD probands than in the relatives of depressives and schizophrenics.

Soloff and Millward (1983) also presented the results for the relatives of the pure SPD probands, the SPD/BPD probands (who met the criteria for both SPD and BPD), and the pure BPD probands. It appears that almost all schizophrenic relatives of SPD and BPD probands were relatives of the mixed SPD/BPD cases. None of the relatives of the pure SPD probands and only one relative of the pure BPD cases were schizophrenic. Eccentric/peculiar behavior was also somewhat more common among the relatives of the mixed SPD/BPD probands. Unfortunately, the presentation of the data does not permit one to compare the SPD and SPD/BPD group with the control group.

In discussing their results, the authors hypothesize that the mixed SPD/BPD group may be more closely related to schizophrenia than pure SPD or BPD. However, the low number of probands in each of the three subgroups makes such an interpretation questionable.

The fact that eccentric/peculiar behavior was clearly more common in the families of SPD and BPD patients as compared to the families of schizophrenics and depressives indicates a familial transmission of borderline states (including SPD). As noted above, however, family studies are better able to disconfirm than confirm a genetic basis for a disorder. From this perspective, Soloff and Millward's study may be considered as a disconfirmation of any relationship between SPD and schizophrenia. However, the study leaves open the possibility that SPD is genetically transmitted.

Baron et al. (1983) conducted a methodologically well-done study of consecutively admitted patients to New York hospitals. The probands were 74 chronic schizophrenics. Their parents and siblings were interviewed with the Schedule for Affective Disorders and Schizophrenia (SADS) (Endicott and Spitzer 1978) and the Schedule for Interviewing Borderlines (SIB) (Baron and Gruen 1980). The Research Diagnostic Criteria (RDC) (Spitzer, Endicott, and Robins 1978) and DSM-III criteria were used for diagnostic assessment. Schizophrenia was defined according to RDC. The relatives were interviewed and rated blindly in three-quarters of the cases. All the probands and 85 percent of the relatives were personally interviewed. In the data analysis, the investigators used an ingenious procedure to elucidate the relationship between schizophrenia and schizotypal personality disorder. The families were divided into three groups according to the manifestation of SPD in the parents: the SPD × SPD group, when both parents had definite or probable SPD; the SPD × N group, when one parent had SPD and the other did not; and finally the rest of the probands who had no parents with SPD constituted the N × N group. The age-corrected morbidity risk for schizophrenia and SPD in the probands' sibs was calculated in each group. The results showed that the prevalence of SPD was highest among the sibs in the SPD × SPD group, somewhat lower among the sibs in the SPD × N group, and lowest in the N × N group. The same was true for the prevalence of schizophrenia among the sibs in the three groups.

The investigators interpret these results as confirmation of two hypotheses: SPD is familially transmitted and SPD is genetically related...
to schizophrenia. The first interpretation is obvious since the morbidity risk of SPD was highest if both parents had the same disorder, lower when only one parent had the disorder, and lowest if none of the parents had SPD. The second interpretation is more doubtful. What the study demonstrated was that parents who had a schizophrenic child had a higher risk of more such offspring if they were both schizotypals, and a lower risk if only one of them was schizotypal. If none of them were schizotypals, the probability of having more than one schizophrenic child was lowest.

Several interpretations of the results are possible. One interpretation is that schizophrenia and SPD have an analogous genotype. This genotype will then be more strongly aggregated in the SPD × SPD group and make schizophrenia more likely to appear among the sibs. Another interpretation, based on research with genetic markers (Siever, personal communication), is that specific genes that predispose to schizotypal characteristics may, in conjunction with other genes, which may not necessarily be specific to schizophrenia but rather reflect a genetic vulnerability to decompensation, result in a schizophrenic phenotype. A third possibility is that an adverse family atmosphere created by schizotypal parents lowers the threshold for schizophrenic phenotypic representation. In view of the results of the Danish adoptive studies, perhaps a fourth hypothesis is most likely: Among the schizotypals there exists a smaller subgroup that is genetically related to schizophrenia. Baron et al. (1983) seem to favor this view. Fully aware of the sampling problems related to the fact that the families were ascertained through schizophrenic probands, they state:

The schizotypal sample identified in this study differs from a random population sample of schizotypal persons. Hence, it would be incorrect to suggest that the presence of schizotypal features greatly increases the risk of having a schizophrenic or schizotypal child, whereas this may not be the case unless a schizophrenic individual is already present in the pedigree. Put differently, it would be misleading to consider SPD in the population a regular expression of the genetic disposition to schizophrenia. [p. 11]

Even so, they state: “This study suggests, however, that schizotypal features constitute an important ingredient of schizophrenia-related disorders” (p. 12).

Recently, Kendler et al. (1984) conducted a blind family history study of the first-degree relatives of consecutive admissions to a VA Medical Center. Criteria for a schizoid-schizotypal personality disorder were defined by social isolation, eccentric behavior, and deviant communication. Their results showed a morbidity risk of 4 percent for schizoid-schizotypal personality disorder among the first-degree relatives of schizophrenics, compared to zero among the relatives of controls. The study is of limited importance for the questions dealt with in this review because the control group was made up of normals and not a contrast group of other psychiatric patients. There is reason to believe that a morbidity risk as low as 4 percent for schizoid-schizotypal personality disorder also may be found among the relatives of other psychiatric patients. The study has thus not demonstrated a specific familial link between SPD and schizophrenia.

Baron et al. (1984) recently conducted a family history study comparing relatives of SPD and BPD patients with normal controls. They found no schizophrenia among the relatives. However, they observed an increased risk of SPD among the relatives of the pure SPD compared to mixed SPD/BPD, pure BPD, and normals.

The family studies reviewed above seem to lead to the same conclusion as the adoptive studies: SPD is related to schizophrenia, but schizophrenia is not related to SPD. Put less paradoxically: A hypothesis about the relationship between SPD and schizophrenia is strengthened when the probands are schizophrenics, but disconfirmed when the families are ascertained through schizotypal probands.

Twin Studies

Twin studies represent a unique opportunity to study the spectrum of variation in an etiologically homogeneous diagnostic grouping. As twin partners experience a similar family environment, nosological variants with similar familial transmission are expected to appear in the same twin pair. Furthermore, as monozygotic (MZ) twins have an identical genetic endowment and dizygotic (DZ) twin partners are no more similar genetically than sibs in general, a genetic link between two different diagnostic variants means that both diagnoses are more often represented in the same MZ pair than in the same DZ pair.

Siever and Gunderson (1979), in an extensive review of the relevant twin studies, made the following main observations: From the variably detailed descriptions of the cotwins in the schizophrenic twin studies of Inouye (1961), Kringlen (1967), Tienari (1968), Essen-Møller (1970), Fischer (1972), and Gottesman and Shields (1972), Siever
and Gunderson (1979) estimated that 7-25 percent of the MZ twins could be broadly conceived as borderline cases, compared to 6-10 percent among the DZ cotwins. In addition to such disorders, many cotwins of MZ schizophrenics showed nonpsychiatric peculiarities:

When specified, these characteristics seemed to resemble the cognitive distortions and atypical mentalizations found in the adoptive studies rather than any uniform personality type or style of social adaptation. Since these characteristics are mild and do not impair the individual's personal functioning, it would seem unwise to assign a psychiatric label to such individuals. [p. 78]

It is also important to note that no schizophrenia spectrum twin study reported a borderline schizophrenic index twin with a chronic schizophrenic cotwin. Siever and Gunderson (1979) conclude:

In summary, the twin studies do not greatly clarify the relationships of the borderlines to chronic schizophrenics. This is in large part due to lack of explicit or homogenous diagnostic criteria, as well as appropriate controls. If a relationship between borderline and chronic schizophrenia exists, it is not a simple one. The presence of mild psychotic-like symptoms correlates better with genetic relationship to chronic schizophrenia than any identifiable personality type such as borderline or schizoid. Nevertheless, there is some evidence that a borderline adaptation is seen more frequently in the co-twins of chronic schizophrenics than any other identifiable personality disorder. [p. 78]

One more recent twin study has ascertained twin pairs through schizotypal probands (Torgersen 1984). In a study of all same-sexed twins admitted to inpatient and outpatient facilities in Norway, 59 index twins were diagnosed as DSM-III SPD. Fifteen of them also met the criteria for DSM-III BPD, while 10 index twins received only a BPD diagnosis. The diagnoses were based on personal interviews, including the structured Present State Examination (PSE) (Wing, Cooper, and Sartorius 1974), an anamnestic interview, and a personality questionnaire. A sample of nonpsychotic (neurotic) twins who did not meet the criteria for SPD or BPD and who were matched for age, sex, and zygosity constituted a comparison group. The twins had earlier been diagnosed independently and blindly by the author and two psychiatrists according to Norwegian use of the term "borderline psychosis" based on the description of Hoch and Polatin (1949), Knight (1953), and Kernberg (1977). The author did not know the borderline psychosis diagnosis when the DSM-III diagnoses were given. If the criteria for borderline psychosis were that at least two of the three judges diagnosed the twin as borderline psychotic, then the sensitivity of BPD and SPD in selecting the borderline psychotics was 97 percent and the specificity, 67 percent. The concept of borderline psychosis thus seems to be more narrow than the BPD and SPD concept, since almost all borderline psychotics were designated BPD and/or SPD, but some others also received these diagnoses. When only SPD diagnoses were considered, the sensitivity was 89 percent and the specificity, 74 percent. (The sensitivity and specificity are not given in the article, but can be calculated from the tables.)

Twenty-eight probands (21 SPD, 4 SPD/BPD, and 3 BPD) were MZ twins, and 41 probands (23 SPD, 11 SPD/BPD, and 7 BPD) were DZ twins. None of the 69 cotwins proved to have a DSM-III diagnosis of schizophrenia. Furthermore, none of the cotwins had an ICD-9 (World Health Organization 1978) diagnosis of schizophrenia, according to independent and blind ratings by judges or the PSE computer classification of schizophrenia. Thus, the prevalence of schizophrenia in cotwins in the sample was less than 1/69 (1.5 percent), or more specifically, the prevalence in cotwins was less than 1.7 percent when only the 59 SPD and SPD/BPD were considered. Among the MZ cotwins, the prevalence was less than 4.0 percent in the total proband group and less than 4.1 percent in the group of SPD and SPD/BPD twin pairs.

Although some of the cotwins may develop schizophrenia in the future, such cases will be rare since the mean age of SPD and SPD/BPD probands was 40 years old. Therefore, the study does not confirm the existence of any genetic link between DSM-III SPD and schizophrenia.

However, the study showed that 7/21 (33 percent) of the cotwins of MZ schizotypals were also SPD, against 1/23 (4 percent) of the DZ cotwins. None of the cotwins were schizotypals in the SPD/BPD or the BPD group. When the SPD and SPD/BPD groups were combined, the MZ concordance was 7/25 (28 percent) and the DZ concordance, 1/34 (3 percent). These results strongly support the hypothesis that SPD is genetically determined. The lack of SPD among the cotwins of the mixed SPD/BPD probands is parallel to the results of Baron et al. (1984), who only found an increased risk of SPD among the relatives of the pure SPD, not among the relatives of the mixed BPD/SPD group.

Furthermore, a comparison of the cotwins of SPD probands with the
cotwins of the matched non-SPD control group showed that the only differences in symptom frequency were ideas of reference, social anxiety, self-depreciation, lack of self-confidence, and depressed mood. Psychotic-like symptoms such as soft perceptual distortions and delusions were no more prevalent among the cotwins of the probands than among the cotwins of controls, even if such symptoms were common among the schizotypal index twins. In addition, the cotwins of schizotypals more often felt inadequate at work, and more isolated and distressed by company. The common features among the schizotypal twin and his cotwin seem to be a schizoid, low self-esteem, paranoid-like personality structure—not a psychotic-like, near-to-schizophrenia symptom formation.

The twin study results parallel the results of the adoptive and family studies: If schizophrenics are probands, schizotypals are found among the cotwins. If schizotypals are index cases, no schizophrenia is observed in the cotwins. Furthermore, the schizotypal personality disorder as such seems to have a strong genetic basis.

**Conclusion**

Although the different studies apply various methods in elucidating the genetic transmission of SPD, the results are rather uniform: When the families are ascertained through schizophrenic probands, schizotypals are found in the biological family. However, when schizotypals are index cases, schizotypals but not schizophrenics are found among the relatives. The adoptive and twin studies indicate that the transmission of SPD has a genetic basis. Concerning the relationship between SPD and schizophrenia, the interpretation of the studies is difficult. Application of the polygenic multiple-threshold model (Reich, James, and Morris 1984), along with some diagnostic considerations, may explain the results.

As schizophrenia is a relatively rare disorder and SPD more common, a polygenic multiple-threshold model for the schizophrenia-schizotypal spectrum might predict a relatively higher prevalence of schizotypals among the relatives of schizophrenics and a relatively lower prevalence of schizophrenia among the relatives of schizotypals. Such a model can partly explain the results reviewed in this article. It can explain the prevalence of schizotypals in relatives of schizophrenics. However, it cannot explain why it is so difficult to demonstrate a higher prevalence of schizophrenia in relatives of SPD probands than in relatives of controls. It may be stated from the polygenic multiple-threshold model that the samples are too small to demonstrate the relatively rare cases of schizophrenia in the relatives. Although this may be the case, it is surprising that so few schizophrenic individuals are found among the relatives of schizotypals in the combined studies today.

It may be necessary, in addition to the multiple-threshold model, to make some assumptions about the Danish criteria for borderline schizophrenia and the DSM-III criteria for SPD. As mentioned earlier in this article and as Kendler and Siever discuss more extensively elsewhere in this issue, borderline schizophrenia and SPD only partly overlap. Furthermore, neither the criteria for borderline schizophrenia from the Danish adoptive studies nor the DSM-III criteria for SPD seem to encompass important aspects of the symptomatology of schizotypal spectrum relatives of schizophrenics. A hypothesis may be that the DSM-III criteria for SPD are able to pick out only some “true” schizotypals. At the same time, however, the criteria also cover many individuals with personality disorders unrelated to schizophrenia. If the “true” schizotypals are relatively rare, and the “false” schizotypals more common, a study of SPD probands will seldom reveal schizophrenic cases among the relatives. A study of schizophrenic probands, however, will give a number of “true” schizotypals diagnosed as DSM-III SPD among the relatives.

An important objective of future research will then be to find criteria that differentiate between “true” and “false” schizotypals. Gunderson, Siever, and Spaulding (1983) and Siever and Kendler (this issue) have given some directions. Gunderson, Siever, and Spaulding (1983) maintain that schizotypal relatives of schizophrenics are characterized by social isolation; distant, suspicious interpersonal relationships; eccentric behavior; frequent somatic problems; detached affects; and social dysfunction. Siever (this issue) states that marked social withdrawal, poor rapport, and decreased pleasure in social interaction are associated with biological markers of schizophrenia. Kendler (this issue) maintains that the description in the genetic tradition of borderline schizophrenia better defines the schizotypals genetically related to schizophrenia. These criteria are much the same as those proposed by Gunderson, Siever, and Spaulding (1983) and Siever (this issue). All three articles maintain that the psychotic-like symptoms that are phenomenologically similar to schizophrenia and that have been popular in the clinical tradition only poorly describe “true” schizotypals. This viewpoint concurs with a recent twin
study (Torgersen 1984), which showed that social dysfunction, and schizoid and milder paranoid features were observed in the cotwins of schizotypal probands, and not the more dramatic psychotic-like cognitive and perceptual distortions that were common in the probands. An explanation may be that the former symptoms constitute the genetic “core” of SPD, while the latter represent a decomposition of either schizotypal or other personality disorders.

On the basis of the findings of genetic research to date, it appears that while SPD seems to be genetically transmitted, a genetic link between the broad category of DSM-III SPD and schizophrenia has not been established.

References


Lowing, P.A.; Mirsky, A.F.; and Pereira, R. The inheritance of schizophrenia spectrum disorders: A


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**Announcement**

The annual meeting of the American Psychopathological Association will be held March 6-March 8, 1986. The theme of this year's meeting is "Alcoholism: A Mental Disorder." Papers from invited guests will be presented in plenary session. Continuing Medical Education credit is offered.

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