SUMMARY

Schizophrenia is a multifactorial disease with high genetic heterogeneity and complex inheritance. In Boyacá, Colombia, we studied a group of 20 schizophrenic patients (16 men and 4 women) to establish their sociodemographic and clinical characteristics as well as their genetic and precipitating factors. The patients were analyzed using cytogenetic studies and a descriptive analysis of qualitative and quantitative variables. The disease frequently first manifested in young adults (average age of initiation: 22.5 years). The predominant subtype (8/20) was paranoid schizophrenia, and the onset was typically gradual (14/20). Precipitating factors were found in 15 patients: physical factors in nine patients, social factors in five patients and economic factor in one patient. All karyotypes were normal. Clinical features did not associate with either the sociodemographic characteristics or the genetic and predisposing factors, supporting the clinical heterogeneity of schizophrenia. Patients and their families received genetic counseling and explanations of the study’s results, the possibility of recurrences and the risk of suffering the disease given an affected relative. Further and larger studies are required to determine if the factors evaluated in this study influence the development of the disease.

KEY WORDS

Genetic Factors; Multifactorial; Precipitating Factors; Schizophrenia
RESUMEN

Estudio descriptivo de una muestra de pacientes con esquizofrenia residentes en el departamento de Boyacá, Colombia

La esquizofrenia, enfermedad multifactorial, tiene gran heterogeneidad genética y herencia compleja. En Boyacá, Colombia, se estudió un grupo de 20 pacientes esquizofrénicos (16 hombres y cuatro mujeres) y se establecieron las características sociodemográficas y clínicas y los factores genéticos y precipitantes. Se hicieron estudio citogenético y un análisis descriptivo de las variables cualitativas y cuantitativas. Hubo predominio del comienzo de la enfermedad en adultos jóvenes (promedio de edad en el momento de la aparición: 22,5 años). Predominaron la esquizofrenia paranoide (8/20) con modo de aparición progresivo (14/20). Se hallaron factores precipitantes en 15 pacientes: físicos en nueve, sociales en cinco y económicos en uno. Todos los cariotipos fueron normales. Los rasgos clínicos no se asociaron con las características sociodemográficas ni con los factores genéticos y precipitantes, lo que evidencia gran heterogeneidad en las formas de manifestación de la enfermedad. Se dio asesoría genética a los pacientes y sus familias y se les explicaron los resultados, el riesgo de recurrencias y el de padecer la enfermedad cuando se tiene un pariente afectado. Es necesario analizar una serie mayor de casos, para poder determinar si los factores evaluados influyen en el desarrollo de la enfermedad.

PALABRAS CLAVE
Esquizofrenia; Factores Genéticos; Factores Precipitantes; Multifactorial

INTRODUCTION

Schizophrenia is a multifactorial disease that belongs to the most genetically complex psychiatric disorders. Heredity of schizophrenia is variable, and the overall prevalence of the disease is approximately 1% (1). Gottesman and Bertelsen, in 1989, argued that the unaffected subject of a pair of identical twins has a 50% risk of developing the disease; relatives with the first degree of consanguinity have a risk of 5% to 16%, whereas second- and third-degree relatives exhibit a 2%-5% and 2% risk, respectively (2,3).

Descripción descriptiva de una muestra de pacientes con esquizofrenia residentes en el departamento de Boyacá, Colombia

La esquizofrenia, enfermedad multifactorial, tiene gran heterogeneidad genética y herencia compleja. En Boyacá, Colombia, se estudió un grupo de 20 pacientes esquizofrénicos (16 hombres y cuatro mujeres) y se establecieron las características sociodemográficas y clínicas y los factores genéticos y precipitantes. Se hicieron estudio citogenético y un análisis descriptivo de las variables cualitativas y cuantitativas. Hubo predominio del comienzo de la enfermedad en adultos jóvenes (promedio de edad en el momento de la aparición: 22,5 años). Predominaron la esquizofrenia paranoide (8/20) con modo de aparición progresivo (14/20). Se hallaron factores precipitantes en 15 pacientes: físicos en nueve, sociales en cinco y económicos en uno. Todos los cariotipos fueron normales. Los rasgos clínicos no se asociaron con las características sociodemográficas ni con los factores genéticos y precipitantes, lo que evidencia gran heterogeneidad en las formas de manifestación de la enfermedad. Se dio asesoría genética a los pacientes y sus familias y se les explicaron los resultados, el riesgo de recurrencias y el de padecer la enfermedad cuando se tiene un pariente afectado. Es necesario analizar una serie mayor de casos, para poder determinar si los factores evaluados influyen en el desarrollo de la enfermedad.

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INTRODUCTION

Schizophrenia is a multifactorial disease that belongs to the most genetically complex psychiatric disorders. Heredity of schizophrenia is variable, and the overall prevalence of the disease is approximately 1% (1). Gottesman and Bertelsen, in 1989, argued that the unaffected subject of a pair of identical twins has a 50% risk of developing the disease; relatives with the first degree of consanguinity have a risk of 5% to 16%, whereas second- and third-degree relatives exhibit a 2%-5% and 2% risk, respectively (2,3).

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autoimmune disease (17), family history of schizophrenia or mental illness, father’s age when the patient was conceived (18), parents’ consanguinity (19), and chromosome complement and karyotype (16,20-22); and precipitating factors or social and economic events (23), family relationships (24), physical (25) and psychological factors, and psychoactive substance use (26,27).

**Cytogenetic study by high-resolution GTG banding:** Chromosomes were obtained from heparinized peripheral blood samples with metaphases between 550 and 850 bands per genome according to the protocol of Ikeuchi (1984) (28). Thirty (30) metaphases were read, and the results were reported according to the ISCN 2009 (International System for Human Cytogenetic Nomenclature) (29).

**Statistical analysis:** A descriptive analysis of all qualitative and quantitative variables was performed using SPSS Statistics 17.0® Windows XP software. In addition, Fisher’s exact probability test was performed to determine the association or independence between the following variables: mode of onset of symptoms, type of symptoms, subtypes of schizophrenia, psychoactive substance use, number of medications and chromosome complement. An analysis was also performed to determine if there was (p < 0.05) or was not (p ≥ 0.05) a relationship between the father’s age when the patient was conceived and the age of onset of symptoms. The data were processed using Statgraphics Plus 5.0 software (30) (Table 1).

**Table 1. Association between variables assessed by the Fisher's exact test.**

None of the p-values supported an association between the variables

<table>
<thead>
<tr>
<th>Variable 1</th>
<th>Variable 2</th>
<th>p value</th>
</tr>
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<tbody>
<tr>
<td>Positive and negative symptoms</td>
<td>Subtype of schizophrenia (undifferentiated and paranoid)</td>
<td>0.36</td>
</tr>
<tr>
<td></td>
<td>Subtype of schizophrenia (paranoid and residual)</td>
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<tr>
<td></td>
<td>Subtype of schizophrenia (undifferentiated and residual)</td>
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<tr>
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<td>Mode of symptom onset (acute and gradual)</td>
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<td>Positive and disorganized symptoms</td>
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<tr>
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<td>Subtype of schizophrenia (undifferentiated and paranoid)</td>
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<td></td>
<td>Subtype of schizophrenia (paranoid and residual)</td>
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<tr>
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<td>Subtype of schizophrenia (undifferentiated and residual)</td>
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<td></td>
<td>Mode of symptom onset (acute and gradual)</td>
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<td>Negative and disorganized symptoms</td>
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<td></td>
<td>Mode of symptom onset (acute and gradual)</td>
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<tr>
<td>Chromosome complement</td>
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<tr>
<td></td>
<td>Disorganized and paranoid subtypes</td>
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</tr>
<tr>
<td></td>
<td>Disorganized and residual subtypes</td>
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</tr>
<tr>
<td></td>
<td>Undifferentiated and paranoid subtypes</td>
<td>0.73</td>
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</table>
Ethical considerations: This research was approved by the Ethics Committee of the School of Medicine, University College Our Lady of the Rosary, as required by ethical guidelines, in accordance with the Declaration of Helsinki (31) and resolution 8430 of 1993 of the Ministry of Health of Colombia (32). Patients, relatives, or guardians and external witnesses gave written informed consent to participate in this research, and the objectives, guidelines, methodology, scope and limitations were specified.

Genetic counseling: Cytogenetic results were reported by interconsultation with medical genetics; advice was also provided to the patients and their families concerning the pathogenesis of the disease, heredity, the impact of precipitating factors, the risk of recurrence and of developing the disease, and making informed decisions. This information complemented the psychiatric screening conducted in these patients.

RESULTS

Sociodemographic characteristics: The sample consisted of 16 men and 4 women, age 18 to 64 years (mean 35.15 ± SD 12.6). Men’s ages ranged from 18 to 55 years (mean 33 ± SD 11.6), and women were between 24 and 64 years old (mean 29.5 ± SD 18.4).

Fifteen patients (75%) lived in the same places of birth, and the remaining five patients lived in other locations of the same department. Seventeen patients (85%) had lived most of their lives in urban areas, and the remaining three patients in rural areas. Thirteen (65%) had low socioeconomic status, and seven (35%) were middle status. Regarding the level of education, 10 patients (50%) had completed some degree of high school, 6 (30%) had completed only primary school, and four (20%) had attended college. All patients were Caucasian.

Clinical features: Eight patients (40%) suffered from paranoid schizophrenia (F20.0x), five (25%) from undifferentiated schizophrenia (F20.3x), four (20%) from disorganized schizophrenia (F20.1x), and three (15%) from residual schizophrenia (F20.5x). The age of onset of symptoms was between 14 and 44 years (mean 22.70 ± SD 7.2). By Fisher’s statistical test, there was no relationship between this variable and the subtype of schizophrenia (p = 0.9668).

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<table>
<thead>
<tr>
<th>Table 1 (continuation)</th>
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<tr>
<td><strong>Chromosome complement</strong> (continuation)</td>
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</table>

**Father’s age when the patient was conceived.** **Age of symptom onset** 0.90
The mode of onset was gradual in 14 patients (70%) and acute in six (30%). Using Fisher’s exact test, the presence or absence of symptoms was not associated with the mode of onset of the disease (table 1).

Eleven patients (55%) were currently taking only one drug, eight (40%) were medicated with two or more drugs, and only one (5%) did not use any medication. Clozapine (Clozaril®) was the most commonly prescribed antipsychotic drug among the alternatives offered by the health system, (12 patients, 60%), followed by haloperidol (Haldol®) (six patients, 30%), and pipotiazine palmitate (Piportil L4®). Among the drugs not covered by the health system, risperidone (Risperdal®) (10%) and sulpiride (Dogmatil®) (5%) were prescribed.

Genetic factors: Genetic factors were identified in 12 patients (60%), consanguinity among relatives in three, family history of mental illness in six, and relatives with schizophrenia in three. The age of the parents at the time of gestation was between 20 and 60 years (mean 36.00 ± SD 13.03). There were no phenotypic traits associated with the 22qDS syndrome identified in any of the patients. In addition, all patients had normal karyotypes (46, XX in 4 patients and 46, XY in 16 patients [figure 1]). It should be noted that in two 24- and 28-year-old women and in a 31-year-old man, tetraploid cells were observed at a low frequency, ranging from 2% to 4% of clonality (92,XXXX[2]/46,XX[98], 92,XXXX[4]/46,XX[96], and 92,XXYY[4]/46,XY[96], respectively), but based on the type of abnormality, they were not considered part of the constitutional karyotype of the patients.

Figure 1. Normal karyotype observed with 100X magnification in a woman with schizophrenia
Precipitating factors: In nine patients (45%), there were physical factors associated with working in the military and police, mental stress by study, and work situations. In five patients (25%), social factors associated with religion, family and relationships with the environment were identified. In one patient (5%), the precipitating factor was economic, and in the remaining five (25%) a precipitating factor was not determined. Furthermore, 13 (65%) patients did not consume any psychoactive substance that could have affected their disease. The variables evaluated in the study with Fisher’s exact test are presented in Table 1. No p-value validated an association between these variables.

DISCUSSION

Sociodemographic characteristics: Most patients were men (80%), a trend also observed in previous studies (16,21,22,33-36), which suggests that men have a greater risk of developing the disease. This trend is explained by the interaction between sex hormones, differences in neural development and psychosocial differences.

In this study, we found that a high percentage (85%) of patients resided in urban areas. Sundquist et al. and Mortensen et al. (37,38) concluded that a high level of urbanization is associated with a high risk of developing psychosis and depression. One possible explanation for this relationship is the frequent exposure to infections during pregnancy and childhood due to the difficult living conditions in urban areas (39). Other studies have found no difference between urban and rural environments on mental health (40-41).

Thirteen patients (65%) were of low socioeconomic status, suggesting that this may be a risk factor that can lead to delays in seeking treatment in the initial phase of the disease, thereby contributing to the chronicity of the clinical picture or severity of symptoms (42). However, this disadvantage is not sufficient or necessary to increase the likelihood of developing the disease because this can manifest in families of any social level (42,43).

Due to economic constraints and factors related to living conditions and family environment, 80% of our patients had no college education. The education level influenced the incidence of the disease, which could be direct (patients who did not continue their education because of the stress of study), indirect (patients who did not continue because of an accident), or unrelated (patients who did not continue to study because of cultural beliefs), suggesting that there was another type of triggering event in these cases.

Clinical features: Luengo (44) and Contreras et al. (45) reported that schizophrenia manifests between 20 and 39 years of age. According to the Colombian Association for Mental Health (ACSAM, for its initials in Spanish), the age of onset recorded for men is between 15 and 25 years old and 25 to 30 years old for women, which corresponds with the range in which the first psychotic episode took place in the study population (average of 22.65 years). Similarly, ACSAM reports that mental disorders begin in adolescence and early adulthood, interfering with the achievement of important social, educational, and work-related goals, and can cause lifelong disability.

In our patients, the prevalence of the subtypes of schizophrenia, from highest to lowest, was as follows: paranoid (40%), undifferentiated (25%), disorganized (20%), and residual (15%). This result agrees with that of Contreras et al. (45), who concluded that one of the most common subtypes is paranoid schizophrenia. Further, Luengo (44) and Chinchilla (46) reported that paranoid schizophrenia produces less functional impairment, as it is the most productive from the cognitive standpoint, and affects volitional capacity (capacity for initiative) the least. Because of this, paranoid schizophrenia is regarded as the subtype with the best prognosis and the least likelihood to become chronic. Another study that supports this trend (47) highlights the high prevalence of paranoid schizophrenia (12%) in a sample of 82 patients. Espina et al. (48) studied 50 patients and found the following distribution by subtypes: paranoid (54%), undifferentiated (29%), residual (12%), disorganized (10%), and simple (9%). Although the paranoid subtype is the most common presentation of schizophrenia (45), a patient’s disease frequently makes the transition to another subtype (disorganized, negative, undifferentiated), so that the course of the disease is very different than expected (31,32,44,45). Contreras et al. (45) suggest that many patients have episodes with symptoms that do not correspond to a unique type of schizophrenia, and thus, a higher percentage of undifferentiated schizophrenia is being diagnosed. In their study of 297 patients with schizophrenia, subtypes were distributed as follows: undifferentiated (45%), paranoid (29.1%), disorganized (15.9%), and others (10.1%).
In our sample, eight patients (40%) had positive symptoms (paranoid schizophrenia), five (25%) had concurrent symptoms (undifferentiated schizophrenia), four (20%) had disorganized symptoms (hebephrenic or disorganized schizophrenia), and three (15%) had negative symptoms (residual schizophrenia). However, Rosenthal et al. (49), in their study of 29 patients, found concurrent symptoms in 58.6%, negative symptoms in 24.1%, and positive symptoms in 17.3%. Espina et al. (48) also found a higher incidence of concurrent symptoms (58%), followed by negative symptoms (50%) and positive symptoms (12%). These differences in the frequency of symptoms compared to our study can be explained because the progress of schizophrenic patients is highly variable; it is quite common that symptoms change from positive to negative, disorganized or simultaneous throughout the patient’s life (44).

In our patients, disease onset was gradual in 14 patients (70%) and acute in six (30%). Ey et al. have determined that the onset of schizophrenia is slow in more than half of the cases and acute in 30% to 40% (50).

During the course of our study, the most frequently used antipsychotics in Colombia were clozapine, haloperidol and pipotiazine palmitate because they are included in the Mandatory Health Plan; it is possible, however, to recognize the effectiveness of the new antipsychotics yet to be included in the plan, each of which has special indications. The additional cost of using such medications is a limitation for those cases in which their use would otherwise be justified to achieve a better response.

Mata et al. (51) suggest that atypical neuroleptics are being introduced in patients according to the clinical features, clinical progress and type of response of the disease. In addition, the use of several types of atypical neuroleptics remains essential in treatment because ideally, the goal is to achieve the maximum benefit in terms of disease improvement.

**Genetic factors:** The parents of 13 patients (65%) were over 30 years of age at the moment of conception. Paternal age can affect the risk of developing schizophrenia, as de novo mutations can appear in the paternal germ line (52-55).

With regard to the family history of schizophrenia for 12 of our patients (60%), Robert et al. (56) demonstrated the existence of a threshold for the addition of factors, below which the trait is not expressed. Some individuals exceed this threshold, which is supported by two segregation models: 1) a recessive gene of major effect in addition to the participation of two or three genes of smaller effect and interactions with the environment; and 2) many genes of small effect in addition to the interaction with the environment. Consanguinity between cousins, present in three of our patients, is a risk for familial schizophrenia (19).

Table 2 presents the cytogenetic findings of the patients, all normal, and compares the results with those reported in other populations of patients with schizophrenia. Among them, DeLisi et al. (53) studied 46 patients and reported that all of them had normal karyotypes. Other studies report normal karyotypes in 68% to 97.4% of cases.

The conventional technique used by the seven investigations comprised the standard protocol for the culture of peripheral blood lymphocytes. GTG banding was performed for the extended chromosome, and two high resolution studies are prominent (16,21). For chromosome reading, 20 to 30 metaphases were analyzed, and in mosaicisms, 100 metaphases were counted, a similar approach to that used in this study (table 2).

With regard to non-constitutional additional findings, corresponding to low-proportion mosaics with tetraploid cell lines, it should be noted that this type of anomaly has also been reported in other studies, such as Demirhan and Tastemir (22), who found mosaics with aneuploidies, including the following: 47, XY, + mar[1]/hyperploidy[60]/46, XY[48], 47, XX, +21[4]/hyperploidy[5]/46, XX[93], hyperploidy 54[1]/triradial figure[1]/46, XY[68], hyperploidy 55[1]/46, XY, del (22) (q11) [1]/46, XY [18]. Furthermore, Iourov et al. (57) claim that schizophrenia is most likely associated with an increase in aneuploidy and polyploidy. In their study, they demonstrated a high incidence of mosaic aneuploidies in individuals with psychiatric disorders and argued that such anomalies are present in samples from different tissues, mainly in brain. Due to the type of abnormality and their low incidence, it is advisable to develop further studies on skin or gonadal tissue biopsies to establish a cytogenetic and diagnostic interpretation of the findings, offering a better-informed genetic counseling to patients and their families (58). Finally, chromosomal heteromorphisms 9qh+ and 13ps+ were similar to those reported by Demirhan and Tastemir (22).
Table 2. Comparison of conventional cytogenetic findings from global studies in patients with schizophrenia reported between 1988 and 2003 (4).  

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<tbody>
<tr>
<td><strong>Total population</strong></td>
<td>20</td>
<td>134</td>
<td>161</td>
<td>250</td>
<td>47</td>
<td>38</td>
<td>120</td>
<td>46</td>
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<td><strong>Women</strong></td>
<td>4/20 (20%)</td>
<td>33/134 (24.6%)</td>
<td>62/161 (38.5%)</td>
<td>128/250 (51.2%)</td>
<td>19/47 (40.4%)</td>
<td>17/38 (44.7%)</td>
<td>61/120 (50.8%)</td>
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<td><strong>Men</strong></td>
<td>16/20 (80%)</td>
<td>101/134 (75.4%)</td>
<td>99/161 (61.5%)</td>
<td>122/250 (48.8%)</td>
<td>28/47 (59.6%)</td>
<td>21/38 (55.3%)</td>
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<td>17-61</td>
<td>17-86</td>
<td>41.7 (average)</td>
<td>14.3 (average)</td>
<td>9-18</td>
<td>18-60</td>
<td>19-63</td>
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<td><strong>Normal karyotype</strong></td>
<td>20/20 (100%)</td>
<td>91/134 (68%)</td>
<td>140/161 (87%)</td>
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<td><strong>Universal numerical abnormality</strong></td>
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<td>2/15 (13.3%)</td>
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<td>10/15 (66.7%)</td>
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<td>0/1 (0%)</td>
<td>4/7 (57.2%)</td>
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<td><strong>Mosaic</strong></td>
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<td>15/21 (71.4%)</td>
<td>3/15 (20%)</td>
<td>0/5 (0%)</td>
<td>1/1 (100%)</td>
<td>3/7 (42.8%)</td>
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<td>B-GTG in SP</td>
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<td>B-GTG, FISH Test in SP. AR</td>
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<td>20 metaphases</td>
<td>20 metaphases</td>
<td>20 metaphases</td>
<td>20 metaphases and 100 for Fragile X</td>
<td></td>
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</tbody>
</table>

B (bands), B-GTG, BC (Giemsa-Trypsin jolting and C), FISH (fluorescence in situ hybridization), SP (peripheral blood), AR (high resolution)

* The incidence of mosaic chromosomal abnormalities in patients with schizophrenia is 31.5% according to the seven values recorded.

† This classification includes mosaicsisms with one or more numerical or structural abnormalities present in only one cell 17/36 (47.2%), mosaics with at least one of the abnormalities in more than two cells 7/36 (19.4%), and mosaics with two or more cells in each of the complements found 12/36 (33.3%)
Precipitating factors: In one patient, the triggering event was the influence of the family structure, a feature within the range of environmental events that trigger the disease. Touriño et al. (59) support the influence of family factors with different theories: 1) the schizophrenogenic mother, 2) the double bind, 3) schism and marital skew, 4) pseudomutuality, and 5) speech and communication alterations. These theories are based on population studies.

The economic factor was crucial for two patients. Financial constraints and feelings of failure because of being unable to continue their higher education created a precipitating stress situation. Wiscarz et al. (23) explain that economic factors are one of the stress situations that have to be faced throughout life; the impact of low income is significant in the groups at risk of developing a psychiatric disorder; and the commonly held view is that the impact of all other risk factors is multiplied by poverty. Hoffman et al. also documented the relationship of poverty and severe financial stress with poor health (27).

Stress caused by the social environment and work-related problems were observed in three of our patients, and one developed the disease due to factors of an academic nature. Ballon et al. (60) reported that the disease is accelerated by a malfunctioning of personal, social, emotional, and academic aspects including the following: work, social independence, and social relations such as dating, suggesting that these are important precipitating factors that may predict mental health in an adult.

The situations involving military personnel were the most common factor in the male population of our study; these occurred in four of the 16 men, with an onset of symptoms between 18 and 25 years of age. Some patients reported having had disturbances during their military service, and others exhibited antipathy towards the military. The onset of schizophrenia in the military has also been studied in Perú; the Ombudsman’s Report No 42 of 2002 “The right to life and personal integrity in the context of military service in Perú” shows that recruits are subjected to conditions of severe rigor, which help trigger symptoms of mental health disorders. Other military stressful experiences have been documented (44,61).

Drug abuse is not a causal factor for schizophrenia but can trigger a faster development of the disease or cause further deterioration over the course of the disease (62). In our study, seven patients (35%) had consumed psychoactive substances, and this environmental factor may have contributed to the development of the disease. Neurobiological development and substance abuse explain how the loss of neurons with dopaminergic activity leads to neocortical hypofrontality and thus anhedonia and dysphoria states, which are important risk factors for chronic disease; loss of such neurons can also alter the remaining functionality of patients with positive symptoms (63).

This study has been a first approach to the clinical description of the schizophrenic population in Boyacá. A predominance of men and a disease onset in young adulthood with an average onset at 22.5 years were observed. Paranoid schizophrenia predominated (40%) with gradual onset (70%). Statistically, clinical features exhibited no association or particular trend, which demonstrates heterogeneity, and it follows that every patient has peculiarities in the forms of disease manifestation. However, it is necessary to continue with these types of studies to analyze a larger series of cases to identify whether the evaluated factors may influence the development of the disease and thus propose preventive measures to reduce its impact in Colombia.

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