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Status of Adults With X-Linked Agammaglobulinemia:

Impact of Disease on Daily Lives, Quality of Life, Educational and Socioeconomic Status, Knowledge of Inheritance, and Reproductive Attitudes

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Abstract

Since many children with X-linked agammaglobulinemia (XLA) can now be expected to reach adulthood, knowledge of the status of adults with XLA would be of importance to the patients, their families, and the physicians caring for these patients. We performed the current study in adults with XLA to examine the impact of XLA on their daily lives and quality of life, their educational and socioeconomic status, their knowledge of the inheritance of their disorder, and their reproductive attitudes. Physicians who had entered adult patients with XLA in a national registry were asked to pass on a survey instrument to their patients. The patients then filled out the survey instrument and returned it directly to the investigators. Adults with XLA were hospitalized more frequently and missed more work and/or school than did the general United States population. However, their quality of life was comparable to that of the general United States population. They achieved a higher level of education and had a higher income than did the general United States population. Their knowledge of the inheritance of their disease was excellent. Sixty percent of them would not exercise any reproductive planning options as a result of their disease. The results of the current study suggest that although the disease impacts the daily lives of adults with XLA, they still become productive members of society and excel in many areas.

INTRODUCTION

X-linked agammaglobulinemia (XLA) is a primary immunodeficiency disease characterized by defective development of B lymphocytes, profound hypogammaglobulinemia, and markedly deficient antibody formation and function [7,19,20]. It is caused by mutations in the gene for a B-lymphocyte tyrosine kinase, termed Btk [7,15,19,20,24,25]. Affected individuals have a marked susceptibility to infection with encapsulated bacteria, such as the pneumococcus and *Haemophilus influenzae*, type b, as well as *Giardia lamblia* and enteroviruses [5,7,9,13,19,20,21,26].

The first patient with XLA was identified by Colonel Ogden C. Bruton, MD, in 1952 [2]. Initially, intramuscular gammaglobulin was the only replacement therapy available. Unfortunately, since gammaglobulin could be given only by the intramuscular route, the dose was limited, and immunoglobulin replacement therapy in the first patients was inadequate. In a multi-institutional survey of United States residents with XLA performed in 1985, which

was largely reflective of the experience of patients on intramuscular gammaglobulin, 17% of the patients had died and over 50% had chronic pulmonary diseases [13]. In a survey of Italian patients with XLA, 80% of the patients who had been followed for 2 decades had chronic lung disease [21].

In the late 1970s and early 1980s, intravenous preparations of gammaglobulin were developed that allowed patients to receive larger doses of gammaglobulin and sustain higher levels of serum IgG [4]. In addition, in recent decades, patients with XLA have been diagnosed earlier in life [26], allowing for earlier replacement therapy and increased attention to the diagnosis and therapy of their infections. Because of these improvements in initial diagnosis, immunoglobulin replacement therapy, and treatment of infections, patients with XLA are avoiding long-term pulmonary insufficiency [22] and can be expected to survive into adult life. In fact, in a survey of 201 patients published in 2006, 43% of the patients were aged 18 years or older [26]. In the same study, during a prospective follow-up period of 4.5 years, only 3 patients died, and 2 of these 3 died from iatrogenic causes [26].

Since many children with XLA can now be expected to reach adult life, knowledge of the status of adults with XLA would be important for physicians caring for those patients, and certainly also for the patients themselves and their families. In fact, a 2006 study [10] that examined the health status and quality of life of 41 adult patients with XLA demonstrated that most adults with XLA are moderately healthy and have a reasonable quality of life. The current study was performed in adults with XLA to examine the impact of XLA on their daily lives and quality of life, their educational and socioeconomic status, their knowledge of the inheritance of their disorder, and their reproductive attitudes.

PATIENTS AND METHODS

Ascertainment of Patients

Patients were ascertained for this study using the National Registry of United States Residents with X-linked Agammaglobulinemia [26]. This registry is supported by a National Institutes of Health contract, and among its purposes is to provide a resource for individuals who wish to investigate different aspects of the disease. The registry is anonymous with respect to patients' names, addresses, or other specific identifiers. Patients are entered into the registry by their physicians, and the only patient identifiers are birth dates, initials, sex, and race.

Physicians who had patients aged 18 years or older were sent a letter explaining the study and requesting that they pass the study instrument on to their adult patients. Letters were sent to 43 physicians who had, or had had in the past, 83 potential patients. A total of 25 patients returned the survey instrument. Of these, none had been included in a previous study of adults with XLA [10].

Diagnosis Criteria

Patients with XLA entered in the registry must fulfill 1 or more of the following diagnostic criteria [6]: 1) have a mutation in the Btk gene and/or defective expression of the Btk protein, 2) have a positive family history of a maternally related lateral male relative with XLA (for example, either a mutation in the Btk gene, defective expression of BTK protein, or markedly reduced numbers of B lymphocytes in their blood and hypogammaglobulinemia) and markedly reduced numbers of B lymphocytes in their blood and hypogammaglobulinemia, or 3) have markedly reduced numbers of B lymphocytes in their blood and hypogammaglobulinemia.

Survey Instrument

The survey instrument was a 15-page pencil and paper questionnaire. Most items on the questionnaire were in the form of multiple-choice questions. A few asked open-ended questions (for example, "What is/was your primary occupation?"). The 12-item short form health survey, version 2, (SF-12v2) quality of life instrument was included within the larger questionnaire and was scored using software from Quality Metrics, Inc (Lincoln, RI). The group means of the patients' responses to the SF-12v2 questionnaire were compared to the fixed scores of adult males in the United States by use of a Student t-test.

Institutional Review Board

The study was approved by Western Institutional Review Board (#20031427).

RESULTS

Impact of Medical Care on Daily Lives

Medical Visits—Twenty-one patients had been hospitalized at some time. Most patients (13/25; 52%) had been hospitalized within the last 5 years, but only 24% (6/25) had been hospitalized in the preceding 12 months (Table 1). The reasons for the most recent hospitalizations varied. Infection-related causes accounted for 15/21 (71%) of the most recent hospitalizations (pneumonia, 7 patients; sepsis/bacteremia, 3 patients; otitis media, 2 patients; upper respiratory infection, 1 patient; infected ankle/foot, 1 patient; osteomyelitis, 1 patient). Surgery accounted for 4/21 (19%) of the most recent hospitalizations (appendicitis, 2 patients; back surgery, 1 patient; and cholecystectomy, 1 patient), and infusion of intravenous immunoglobulin (IVIG) accounted for 2/21 (10%) of the most recent hospitalizations. The number of days of hospitalization during the last 12 months varied from 1 to 25. Twenty-three patients had 1 or more outpatient/office/clinic visits in the preceding 12 months (see Table 1).

Days Missed From Work or School—Twenty-three of the 25 patients reported that they either worked or attended school; 1 patient was unable to work and 1 patient was retired. Thirty-five percent of the patients who worked and/or attended school (8/23) reported that they did not miss any days of work or school in the preceding 12 months. Most patients, 65% (15/23), did miss at least 2 days of work or school in the preceding 12 months; 10 individuals (43%) missed fewer than 10 days, while 5 (22%) missed 10 or more days.

Medications—Twenty-three patients were receiving IVIG replacement therapy at the time of the survey; 1 was receiving intramuscular gammaglobulin and 1 was receiving subcutaneous immunoglobulin. Most patients who received IVIG therapy received it at 3–4 week intervals, in their homes and from a nurse (Table 2). Some patients received IVIG as frequently as every 2 weeks (n = 5); some received IVIG in their physicians' offices (n = 4), outpatient department (n = 2), or inpatient unit (n = 1); and some were infused by a family member (n = 5) or self-infused (n = 1).

Seventeen patients were on chronic prescription medications; 10 were on bronchodilators and/or inhaled steroids, 8 were on chronic antibiotics, and 2 were on statins.

Health Insurance—Health insurance concerns and problems varied. Ten patients reported that they had not had any significant problems with health insurance. The remaining 15 patients reported a variety of problems; most patients experienced more than 1 kind of problem. Ten patients reported that they had insurance denied at some point, 5 had their condition excluded from coverage, 5 reported that they had their immunoglobulin replacement denied or delayed, and 6 had their insurance canceled at some point in time.

Quality of Life

Quality of life was assessed using the SF-12v2 survey instrument. Although the patients scored lower than the United States adult male population in each of the 8 individual scales, as well as on the 2 composite scales reflecting physical and mental health (Table 3), their quality of life was comparable to that of other adult males in the United States general population since the differences were not statistically significant.

In addition to the formal quality of life assessment presented above, the patients were also asked about the kinds of physical activity in which they participated. The majority of patients participated in some regular physical activity. For example, 14 of the 25 walked 30 minutes or more at least weekly, and 5 jogged at least a few times each week.

Socioeconomic Status

Thirteen of the patients were married at the time of the survey, 2 were currently members of an unmarried couple, and 1 was currently divorced; 9 had never been married or a member of an unmarried couple. Fifteen of the patients lived with either a spouse or other life partner, 4 lived with their parents, 3 lived alone, and 1 lived with a roommate; 2 listed “other” for living arrangements.

All but 1 patient had completed high school (Table 4). Twenty patients (20/25; 80%) had attended college. Of these, 7 (28%) had graduated from either a 2-year or 4-year college, and an additional 6 (24%) received either a graduate or a professional degree.

Fifteen of the patients were employed for wages at the time of the survey, 5 were self-employed, 3 listed themselves as students, and 2 were unable to work or were retired (Table 5). The kinds of jobs or professions that the patients listed as their primary occupation, either currently or when they last worked, are listed in Table 5. Patients’ personal and household incomes are displayed in Table 6.

Genetic Knowledge and Reproductive Attitudes

Twelve of the 25 patients had children. The patients’ knowledge of the genetics of their disease and the risk that they calculated of specific outcomes of pregnancies are listed in Table 7. Only 1 of the patients failed to know that his risk for having an affected son was 0%, and the same patient was the only one who failed to know his risk for having an unaffected son was 100%. Another patient was the only one who failed to know that his risk for having an affected daughter was 0%. Although 7 of the patients failed to understand that their risk of having a carrier daughter was 100%, those 7 did understand that there was a risk of 50% or greater, and none thought there was no risk at all.

The patients’ reproductive attitudes and choices are displayed in Table 8. The majority of patients (60%) had chosen, or would choose in the future, not to exercise any reproductive planning options. Only 2 of the 25 patients had chosen, or would choose in the future, not to have children. None of the patients had tried not to have sons or would try in the future not to have sons; 3 of the patients had tried not to have daughters or would try in the future not to have daughters. However, 8 patients stated that they had in the past, or would in the future, have prenatal diagnosis.

DISCUSSION

The primary immunodeficiency diseases are a family of more than 125 disorders in which the defect is intrinsic to the immune system rather than caused by an extrinsic agent such as chemotherapy, steroids, or a viral infection. The most common clinical manifestations are an

increased susceptibility to infection and/or autoimmune diseases. The overwhelming majority of the disorders are genetically determined. When they were initially described over 50 years ago, children with the genetically determined primary immunodeficiency diseases generally did not survive until adult life. However, with improvements in early diagnosis [17,26] and remarkable advances in therapy [3,4], effective treatments exist for most of the diseases, and many, if not most, of the children born with these diseases now survive until adulthood [23]. This is especially true for XLA. The average age of diagnosis of XLA has fallen significantly over the last few decades allowing earlier initiation of immunoglobulin replacement therapy [26]. In addition, the development of immunoglobulin preparations suitable for intravenous or subcutaneous infusion has allowed patients to receive doses that keep their serum immunoglobulin levels at, or near, that of normal individuals [4,22].

Unfortunately, relatively few studies have examined the status of adults with genetically determined primary immunodeficiency diseases. A previous study [10] of adults with XLA showed that they were moderately healthy and led productive lives. The current study was performed in adults with XLA to determine the impact of the disease on their daily lives and quality of life, their educational and socioeconomic status, their knowledge of the inheritance of their disease, and their reproductive attitudes and practices. Patients were not ascertained or contacted directly but rather were sent the questionnaire by their physicians, who had previously entered them anonymously in a national registry of XLA patients [26].

Patients were given the opportunity to remain anonymous when they returned the survey instrument, so that in many cases the results of the survey could not be linked to the medical data already in the registry. As a result, we could not correlate any of the data obtained in the current survey (for example, income level, reproductive attitudes or practices, marital status) with any of their medical data (for example, age of diagnosis, type of facility where they receive care, presence or absence of positive family history). Another potential limitation of the current study is that the patients may represent a skewed sample of XLA patients, since more than half of the physicians who entered patients in the registry had academic affiliations [26], and the patients in the current study may have received different care than patients with XLA who were served exclusively by physicians in private practice. In addition, the patients in the current study may represent a skewed sample because they were willing to take the time and effort to fill out and return the form. Finally, the results of the current study apply only to those patients who are well enough to have reached adult life, and may not reflect outcomes or expectations of all patients with XLA at birth.

Their disease did have an impact on the daily lives of these patients. Although 24% of the patients with XLA had been hospitalized during the last year, only 7% of the general population of adults in the United States have been hospitalized in the previous year [18]. Similarly, 65% of the patients missed work or school due to illness during the past year, compared to 46% of the general population of adults [18]. Finally, 22% of patients reported missing 10 or more work days in the past year, compared to only 8% of the general adult population of the United States. [18]. These data on hospitalizations and missed days from work or school are comparable to a previous study [10] of adults with XLA in which 32% had been hospitalized in the last year and 14% missed 10 or more days of work or school in the last year. Although the majority of patients in the current study received IVIG at home (70%), a substantial minority (30%) received it in medical facilities, thereby contributing to days missed from work and/or school.

The adult patients with XLA in the current study had a quality of life that was comparable to that of the general United States adult male population. Although they scored lower in each of the individual scales of a standardized measure of quality of life, the SF-12v2, these differences were not significant. It is possible that study of a larger number of patients would have revealed

significant differences. It is noteworthy that the composite scores for mental health were virtually identical to those of other adult males, suggesting that the patients' psychological reaction to their illness was excellent. A previous study [10] of adults with XLA measured quality of life using the same instrument, and found that with the exception of the patients' perception of their general health, adults with XLA also had a comparable quality of life to that of the general population. In a previous study [1] of men with human immunodeficiency virus (HIV) infection, quality of life was related to the severity of disease, as measured by CD4 positive cells and symptoms, suggesting that the normal quality of life in adults with XLA reflects relatively mild clinical involvement from the patients' perspective.

Patients' educational and socioeconomic status was also comparable to, or better than, the general adult United States population. For example, they appeared to be better educated than the general population. Only 20% of the patients had no college experience compared to 44% of the general population [8]. More remarkably, nearly half (48%) of the patients had 4-year college degrees or higher, while only 27% of the general population have 4-year college degrees [8]. Of those patients who had attained a 4-year college degree, one-half went on to obtain a graduate or professional degree. Although their employment status (80%) was comparable to that of adult males in the general United States population (74%) [8], their income appeared to be better. In terms of household income, only 12% of adults with XLA reported annual incomes less than \$30,000 compared to 27% of the adult general population [8]. By contrast, 44% of the adults with XLA reported household annual incomes of \$75,000 or more, compared to 32% of the general adult population [8]. Finally, patients' marital status (52%) was comparable to that of the general adult population (56%) [8].

Patients had an excellent understanding of their genetic risks, substantially better than typically found among adults with other X-linked conditions [11,14]. Only a single individual believed he had a substantial chance of having an affected child. Although one-third of participants did not understand that a daughter would definitely be an XLA carrier, all knew there was at least a 50% chance that she would be. Perhaps reflecting their understanding that their children had virtually no chance of inheriting XLA (although their grandchildren were at risk), most participants indicated that the disease had not, or would not, in any way influence their reproductive practices. One-third said that they would use, or had used, prenatal diagnosis, and 2 indicated they would choose to adopt due to XLA. While previous studies have investigated the affect of various X-linked conditions on reproductive planning and use of prenatal diagnosis among female carriers [12], relatively little attention has been paid to how X-linked disease influences reproductive decisions among affected men. One study [16] of men with classic X-linked hemophilia reported results similar to the current results in that few men altered reproductive planning as a result of the disease. Thus, XLA does not cause a significant reproductive burden for these men, most likely because of their accurate understanding of their negligible personal reproductive risk of having an affected child, and possibly because of their own successful adaptation to the disease.

In summary, the results of the current study suggest that although the disease impacts the daily lives of adults with XLA (for example, days missed from work and hospitalizations) these patients still become productive members of society and excel in many areas (for example, income and education).

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Abbreviations

IVIG	intravenous immunoglobulin
XLA	X-linked agammaglobulinemia

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TABLE 1

Hospitalizations and Outpatient Visits

No. of Events	0	1	2	3	4	5	6	8	10	12	20	25
No. of hospitalizations												
In last 5 yr	12*	6	2	2	0	1	1	1				
In last 12 mo	19	4	1	1								
No. of days in hospital												
In last 12 mo	19	1	0	0	0	1	0	1	1	1	0	1
No. of outpatient physician visits												
In last 12 mo	2	5	2	3	2	1	2	3	1	1	2	1

* No. of patients.

TABLE 2**IVIG Treatment**

	No. of Patients
Frequency of infusion	
Q2 wk or less	5
Q3 wk	8
Q4 wk	9
Q5 wk	1
Location of infusion	
Home	16
Physician's office	4
Hospital inpatient	2
Hospital outpatient	1
Individual administering infusion	
Nurse	16
Family member	5
Physician	1
Patient	1

TABLE 3

Quality of Life Scores

SF-12v2 Scale	XLA Patients *	US Adult Males
Individual scale		
Physical strength	47.56 [†]	51.68
Physical activity	46.94	51.54
Bodily pain	49.52	51.33
Perception of general health	46.82	50.87
Vitality	48.87	51.44
Social functioning	47.59	51.17
Role of emotions	50.69	51.49
Level of depression	49.87	51.26
Summary scale		
Physical	46.82	51.05
Mental	50.36	50.70

* Mean score.

[†] Norm-based scoring where 50 is the mean score for the general US population.

TABLE 4

Educational Status

Highest School Grade Completed	No. of Patients
Some high school	1
12th grade/GED	4
Some college	7
2-year college graduate	1
4-year college graduate	6
Graduate/professional degree	6

Abbreviations: GED = General Educational Development high school equivalency diploma.

TABLE 5

Employment Status

Status	No. of Patients
Employed for wages	15
Self-employed	5
Student	3
Unable to work/retired	2
Type of Employment	
Accountant	1
Arts administrator	1
Carpenter	1
Computer support	1
Construction estimator	1
Express mail courier	1
Firefighter/EMS	1
Fundraiser	1
Owner food processing business	1
PhD research biologist	1
PhD psychologist	2
Physician	2
Podiatrist	1
Project manager	1
Retail sales	2
Stock trader	1
Inventory control supervisor	1
Systems analyst	1
Teacher's aide	1
Truck driver	1
Waiter	1

Abbreviations: EMS = emergency medical services.

TABLE 6

Income of Patients With XLA

Annual Income (US\$)	Personal Income		Household Income	
	Patients No. (%)	General US Population * (%)	Patients No. (%)	General US Population * (%)
<5000	3 (12)	(14)	0 (0)	(2)
5000–14,999	2 (8)	(13)	1 (4)	(12)
15,000–29,999	4 (16)	(22)	2 (8)	(13)
30,000–49,999	7 (28)	(21)	7 (28)	(19)
50,000–74,999	3 (12)	(12)	4 (16)	(20)
75,000–99,999	3 (12)	(5)	6 (24)	(13)
>100,000	3 (12)	(12)	5 (20)	(19)

* Data from 2005 United States Census.

TABLE 7

XLA Patients' Knowledge of Genetic Risk

Chance of:	0%	25%	50%	75%	100%
Son to have XLA	24 *	0	1	0	0
Son to be carrier	24	0	1	0	0
Daughter to have XLA	24	0	1	0	0
Daughter to be carrier	0	0	6	1	18

* No. of patients responding; correct answer is in **bold**.

Table 8

XLA Patients' Reproductive Attitudes and Practices

Did you or would you:	No. of Patients[*]
Choose not to have children	2/25
Delay having children	4/25
Choose to adopt	2/25
Try not to have boys	0/25
Try not to have girls	3/25
Use prenatal diagnosis	8/25
None of the above	15/25

* Some patients selected more than 1 option.