Reproductive health in individuals with homozygous β-thalassemia: knowledge, attitudes, and behavior

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Objective: To review the reproductive health knowledge, attitudes, and behaviors of persons with homozygous β-thalassemia.

Design: Case–control study.

Setting: Patients treated at a tertiary hospital and community-based healthy controls.

Participant(s): One hundred and thirty persons, 16 years of age or older, who had homozygous β-thalassemia and were attending hospital for regular blood transfusion, and 99 demographically matched persons without thalassemia.

Assessment tool: Reproductive health questionnaire.

Result(s): 104 (80%) persons with homozygous β-thalassemia completed the questionnaire and were compared with 99 controls. Persons with homozygous β-thalassemia were as likely as healthy peers to be in a relationship, employed full-time, sexually active, and using contraception and to have had children. Hypogonadotrophic hypogonadism was present in 55 (52.8%) patients, 46 (83.6%) of whom were compliant with hormone replacement therapy. Understanding of genetics and reproductive potential was suboptimal among persons with homozygous β-thalassemia, and this group had a higher rate of unplanned pregnancy.

Conclusion(s): This study suggests that with optimal therapy, most young adults with homozygous β-thalassemia can achieve reproductive, sexual, and social experiences similar to those of their healthy peers. (Fertil Steril® 2002;77:119–27. ©2002 by American Society for Reproductive Medicine.)

Key Words: Homozygous β-thalassemia, reproductive health questionnaire, hormone replacement therapy, libido, assisted fertility, hypogonadotrophic hypogonadism

The reproductive and sexual health issues concerning persons who are homozygous for β-thalassemia (previously called “thalassemia major”) are complex. The chronic nature of the condition, the complexity of its treatment, and the well-documented complication of hypogonadotrophic hypogonadism with subsequent growth failure, delayed or absent sexual development, infertility, and osteoporosis (1–3), put persons with this disorder at particular risk for reproductive disorders and possible associated psychosocial problems.

In the past, children with homozygous β-thalassemia rarely survived beyond adolescence (4, 5); reproductive and sexual health were thus considered irrelevant in this patient group. Improved medical treatment for these patients introduced in the late 1970s, involving regular optimum red blood cell transfusion and almost daily subcutaneous iron chelation therapy, has significantly decreased patient morbidity and increased survival beyond adolescence into middle age (6–10). As a result, patient care has now expanded to include encouraging these patients to aspire to the same vocational, social, sexual, and reproductive goals as their peers without β-thalassemia (11).

An autosomal recessive disorder, homozygous β-thalassemia is characterized by reduced or absent production of the β-globin chains of adult hemoglobin. The genes for β-thalassemia are prevalent in several ethnic groups distributed in a broad geographic belt from the Mediterranean basin through the Middle East and the Indian subcontinent and into southeast Asia (5, 10). The incidence of β-thalassemia outside of this belt has increased because of large-scale immigration. In Australia in 1998, approxi-
mately 390 persons with a hemoglobinopathy received regular red blood cell transfusions.

Although thousands of people worldwide have β-thalassemia, have increased risk for reproductive health problems, and would benefit from information on good reproductive and sexual health for their overall wellbeing, the literature on these issues is scarce. Most published studies have investigated the hypothalamic–pituitary–gonadal axis of these patients from an endocrine viewpoint alone (3, 12–16).

We examined the reproductive complications that persons with homozygous β-thalassemia experience, including how they view reproductive health in the context of their illness and whether having β-thalassemia influences their reproductive behavior. To ensure that the information gathered from the patients with β-thalassemia was interpreted within the context of the broader community, we interviewed a control group without thalassemia that was matched for age, sex, and ethnicity.

**MATERIALS AND METHODS**

**Setting**

The study was conducted at the Medical Therapy Unit, Monash Medical Centre, Clayton, Victoria, Australia, where most of the known patients with transfusion-dependent β-thalassemia in the state of Victoria are managed. Monash Medical Centre is a 400-bed teaching hospital in metropolitan Melbourne. The study was approved by the Monash Medical Centre Institutional Research and Ethics Committee on May 14, 1998.

**Participants**

Sixty-three men and 67 women, 16 to 46 years of age, who had transfusion-dependent β-thalassemia were eligible and were invited to participate while they attended for regular review and blood transfusion from June through August 1998. To be included, patients had to speak English, have transfusion-dependent β-thalassemia, be 16 years of age or older, be in stable physical and mental health, provide informed consent, and present for treatment during the recruitment period. Patients 16 or 17 years of age required parental consent to participate. Potential participants completed a self-administered questionnaire in privacy at the clinic. One hundred and four (80%) of the 130 eligible patients with β-thalassemia completed the questionnaire.

We randomly recruited 50 women and 49 men without thalassemia who were demographically matched to the case-patients to act as a control group. Controls were recruited from primary care clinics and community groups, such as relevant ethnic, social, and sports clubs.

**Measurements**

Four reproductive health questionnaires were developed to obtain the information required to fulfill the objectives of the study (male- and female-specific patient questionnaires and male- and female-specific control questionnaires). Survey questions were developed by reviewing the published literature; incorporating questions from previously administered questionnaires (17); and consulting experts in thalassemia, ethics, and questionnaire development. After pilot testing, the questionnaire was modified to incorporate the suggestions of a sample of six young adults without thalassemia.

The questionnaires, which included closed and open-ended questions, took most participants 20 minutes to complete. The questionnaires included questions on sociodemographic data, specific sexual health issues, sexual activity, and fertility experience.

The data obtained from the questionnaires were supplemented with information on testosterone, estradiol, follicle-stimulating hormone, luteinizing hormone, and serum ferritin levels obtained from each patient’s medical record. A history of hormone replacement therapy (HRT) or use of the oral contraceptive pill was obtained. Competitive double-antibody radioimmunoassay kits were used to measure serum estradiol and testosterone concentrations (DPC, Los Angeles, CA, and Bayer, Tarrytown, NY, respectively). Women with estradiol levels <20 pmol/L [co-efficient of variation (CV) 14.9%] and men with testosterone levels <10 nmol/L (CV 9.9%) were considered hypogonadal. No cross-reactivity with exogenous estrogen preparations, such as in the oral contraceptive pill, was observed.

Mean serum ferritin levels were calculated and recorded for each patient from August 1, 1997 to July 31, 1998. Serum ferritin levels are routinely measured two or three times per year as a measure of iron loading and as an indirect measure of compliance with chelation therapy. Liver iron estimates are also made annually, and biopsy is done when extensive hepatocellular damage is suspected.

**Statistical Analysis**

For the purpose of analysis, the groups were divided into four subsets: men and women with β-thalassemia and male and female controls. In general, chi-square analysis based on multiway tables was used to assess associations between categorical values and the groups. When the sample was small, the Fisher exact test was used.

When comparing continuous variables (e.g., age or biochemical variables) between two groups, the two-sample t-test and its P value were computed to compare the two sample means. The Kruskal–Wallis test was computed for this data to check the P value computed by the t-test and agreed on all accounts.

One-way analysis of variance was used to compare the means between more than two groups when the response variable was continuous. To avoid problems with multiple comparisons, a P value ≤.01 was considered significant. Missing values were omitted in every statistical test.
RESULTS

Study Sample

One hundred and four (80%) of 130 persons with β-thalassemia who were invited to participate in this study consented to participate. Of this group, 56 (53.8%) were women (mean age, 27.31 ± 7.20 years) and 48 (46.2%) were men (mean age, 28.46 ± 7.09 years).

Twenty-two case-patients (16.9%) declined to participate (9 women and 13 men). Reasons for declining included: not being allowed to view the questionnaire before consenting, lack of interest, and issues related to privacy and confidentiality of medical records. Failure to return parental consent resulted in the exclusion of 4 (3.1%) patients (2 female and 2 male), and 1 (0.8%) female patient withdrew because she developed a severe headache while completing the questionnaire.

Of the 99 controls without β-thalassemia, 50 (50.5%) were women 18 to 43 years of age (mean age, 26.45 ± 6.28 years) and 49 (49.5%) were men 16 to 46 years of age (mean age, 27.53 ± 7.37 years).

Age in the four groups did not differ significantly in analysis of variance (P = .569). The case-patient and control groups were well matched for sex, ethnicity, employment status, marital and relationship status, and household composition (Table 1). Overall, case-patients reported a significantly lower level of formal education completed at the time of the survey compared with controls: Only 39% of the patient group had left school to pursue higher education at university or other institutions compared with 65% of the control group (P = .001) (Table 1).

Compliance with Treatment

Forty-five of the 56 (80.4%) female case-patients and 33 of 48 (69%) male case-patients reported that they complied with their physician’s recommendations for how often to use their subcutaneous desferrioxamine pump (P = .173). However, serum ferritin levels >2,000 μg/L were consistently recorded for the year in 13 case-patients, and these patients were therefore included in the poorly compliant group. Female case-patients cited pain, bruising, and reduced freedom associated with treatment equally as the most common reasons for poor compliance. Male case-patients identified sleep problems as the most common reason for poor compliance, followed by bruising and inconvenience. Mean serum ferritin levels were 761 μg/L in the compliant group and 2,473 μg/L in the poorly compliant group.

Female Gonadal Status and HRT

Thirty-five (62.5%) female case-patients were categorized as hypogonadal because they had estradiol levels <20 pmol/L on multiple testing over 12 months. Fourteen (25.0%) of these women had primary amenorrhea of these women and 21 (37.5%) had secondary amenorrhea. The remaining 21 (37.5%) female case-patients had normal go-
Nadal status (Figure 1A) and were currently menstruating regularly without the need for HRT. As part of routine care, all hypogonadal female patients at the Medical Therapy Unit are offered HRT. At the time of the survey, 28 (80%) of hypogonadal women were taking HRT or oral contraceptive pills, 6 (17.1%) had declined HRT, and estrogen replacement was contraindicated in 1 (2.8%) owing to hepatic disease (Figure 1B).

Menstrual Patterns

Fifty-two (91.1%) of the 56 female case-patients reported ever having a period (43 spontaneously and 9 HRT-induced) compared with 50 (100%) of the female controls (49 spontaneously and 1 HRT induced) ($P < .001$). The mean age at menarche was 15.7 ± 0.4 years in female case-patients and 12.8 ± 0.2 years in female controls ($P < .001$). The median age at menarche was 15.5 years (range, 12–25 years) in female case-patients and 12.6 years (range, 10–19 years) in female controls.

When menstrual history was analyzed according to age, primary and secondary amenorrhea were significantly more common in women currently >30 years of age ($P = .003$). This finding may reflect previous suboptimal treatment.

Male Gonadal Status and HRT

Male case-patients were categorized as eugonadal or hypogonadal on the basis of their clinical records and biochemical results (Figure 2A). Twenty-eight (58.3%) of 48 male case-patients had normal gonadal status and 20 (41.7%) of 48 males were hypogonadal. Of the 20 hypogonadal case-patients, 18 (90%) were currently taking HRT; the remaining 2 (10%) had declined therapy because of such side effects as headache and lethargy (Figure 2B). Fifteen male case-patients were receiving intramuscular testosterone injections, 2 had received testosterone implants, and 1 was taking oral testosterone tablets. The mean age at start of testosterone therapy was 22.3 ± 1.9 years (range, 13–35 years).

Nine (20.0%) of 45 male case-patients and 7 (15%) of 48 male controls reported that they did not experience nocturnal or early morning erections ($P = .489$). Six of these 9 case-patients had normal gonadal status, 2 were hypogonadal but taking HRT, and 1 was hypogonadal but declined to take HRT.

Sexual Activity

Respondents were asked to rank their current level of sexual interest and libido on a five-point scale from very strong to none. Overall, a significantly greater percentage of men than women (case-patients and controls) reported very strong to moderately strong levels of sexual interest (91% vs. 58%, respectively) ($P < .001$). Weak to no sexual interest was reported most commonly by female case-patients. Both male gender and absence of thalassemia were significantly associated with an increased probability that a study participant had ever had sexual intercourse ($P = .004$).

More than 87% of case-patients and controls who said that they had ever had sexual intercourse also said that they had been sexually active in the preceding 12 months. Men reported a significantly lower age at first sexual intercourse than did women ($P < .001$). Almost 90% of the men had first had sexual intercourse before the age of 20 years. The proportions of male case-patients and male controls who had first sexual intercourse at an age younger than 16 years did not differ significantly ($P = .36$). Two-sample t-tests for within-differences sex showed no difference between male case-patients and male controls ($P > .1$) or between female case-patients and controls ($P > .05$).
Contraceptive Use

Contraceptive use and opinions about the effectiveness of contraception did not statistically differ across the four groups. The most commonly used form of contraception among sexually active participants, regardless of sex, was the condom, followed by oral contraceptive pills among female case-patients and female controls, the withdrawal method among male case-patients, and no contraception among male controls.

Fertility Experience

Table 2 shows the fertility experience of 10 women with \( \beta \)-thalassemia who reported ever being pregnant and had borne 10 children in total. Six of these women were hypogonadal at the time of conception and required fertility assistance to become pregnant. Four women, all of whom currently had normal gonadal status at the time of the study, reported at least one spontaneous unplanned pregnancy each; one woman had a termination for this reason. Two of these 4 women reported that at the time of the unplanned pregnancy, they were using condoms for contraception; one said that she was using the rhythm method; and one declined to answer the question. Despite their spontaneous pregnancies, all 4 women said that they believed women with thalassemia have reduced fertility. One woman with thalassemia had a miscarriage and termination of pregnancy because of fetal malformation (anecephaly).

Twelve male case-patients reported ever fathering children (Table 2). Six of these men were taking testosterone supplements, of whom 5 reported requiring fertility assistance for fertilization. Collectively, the 12 men had fathered 25 pregnancies, 3 of which ended in miscarriage and 2 in termination. One pregnancy was terminated because the pregnancy was unplanned, and one because prenatal diagnosis showed that the fetus was affected by thalassemia.

Understanding of Genetic Risk

Fewer than 50% of the patients could correctly describe the risk that their offspring would have homozygous \( \beta \)-thalassemia if their partner was a carrier of the \( \beta \)-thalassemia. More than 25% believed that every child born would be affected, and 13% believed that the risk was unpredictable.

DISCUSSION

The value of good reproductive and sexual health is increasingly acknowledged to be important, regardless of the presence or absence of chronic disease. Traditionally, adults with childhood-onset chronic illness have been considered asexual (18–20). Yet it is also acknowledged that reproductive health and sexuality are central to adult life, influencing identity, self-esteem, social roles, and family planning (21, 22).

Adolescents and young adults with \( \beta \)-thalassemia face the same sexual and reproductive issues as their peers, in addition to the specific complications of the disease and its treatment. These complications essentially arise from the chronic nature of the condition (1), the complexity of its treatment, and the prevalence of hypogonadotrophic hypogonadism (2, 3).

More than 80% of our case-patients were of Greek or Italian descent, reflecting both the high prevalence of

(Figure 2)
TABLE 2
Fertility experience among participants with homozygous \( \beta \)-thalassemia.

<table>
<thead>
<tr>
<th>Case-patient</th>
<th>No. of pregnancies achieved</th>
<th>Fertility assistance required?</th>
<th>Gonadal status</th>
<th>Unplanned pregnancy(^a)</th>
<th>Type of contraception when unplanned pregnancy occurred</th>
<th>Miscarriage</th>
<th>Pregnancy terminated</th>
<th>Reason for termination</th>
<th>No. of children</th>
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</tbody>
</table>

\(^a\) The number of unplanned pregnancies appears in parentheses.

\(^b\) It is unclear whether this man misunderstood the question or whether he developed hypogonadism subsequent to the birth of his children.

\(^c\) This man’s partner had suboptimal fertility.

In α-thalassemia in these countries and immigration patterns to Australia (23).

Hypogonadotrophic hypogonadism was present in 52.8% of our case-patients. Dysfunction of the pituitary gonadal axis results in failure of pubertal growth, delay or absence of sexual development, primary and secondary amenorrhea, infertility, sexual dysfunction, and osteoporosis (2, 3, 14–16, 24). In β-thalassemia, these problems primarily result from chronic anemia and iron deposition in the pituitary gland or in the gonad itself (2), particularly in persons with poor compliance with therapy or no access to optimal medical care. All hypogonadal patients at our medical therapy unit are offered HRT to optimize growth, fertility, self-esteem, and bone mineralization. As a result of increased education by staff at the unit, the use of HRT in these patients has increased substantially; more than 85% of hypogonadal patients currently take HRT, compared with 65% in 1995 (24). Persistence with patient education about the broad benefits of HRT is important to help motivate patients to remain compliant with this therapy.

Primary and secondary amenorrhea are common complications of thalassemia (16) and were observed in more than two thirds of our female case-patients. The high percentage of female case-patients (90%) who reported having regular periods reflects the widespread use of HRT in this group. One third of the female case-patients reported that they had their first period only after commencement of HRT. Medical intervention for primary amenorrhea is rarely done before 16 years of age, which in part explains the significant delay in mean age at menarche among case-patients (15.8 years) compared with controls (12.8 years). Some have advocated earlier use (<16 years) of HRT in patients with β-thalassemia to improve self-esteem and bone growth (2).

Smaller studies have shown that adolescents with β-thalassemia experience substantially more emotional problems, social isolation, and stigmatization than their peers, suggesting that these adolescents have limited opportunities for normal sexual experimentation and social development (25, 26). Limitations on sexual expression and experimentation have been described in adolescents with chronic illness whose parents and clinicians are overprotective (22) or among those whose parents doubt or deny the adolescent’s sexual and reproductive capacity (18). The consequences of these behaviors and attitudes often manifest as omnipotent and oppositional behavior, wherein adolescents may “act out” adult roles by having multiple sexual partners, not using contraception (18), or refusing to comply with essential medical treatment (27). However, we found that persons with β-thalassemia were just as likely as their unaffected peers to be in a relationship, be sexually active, use contraception, and have had children or be looking forward to having children in the future, all despite the demanding treatment regimen that these patients must follow to stay well.

Compliant patients use desferrioxamine therapy for 10 hours of every 24, and most do so only at night. The therapy involves the attachment of an infusion pump by subcutaneous needle to the abdominal wall; this may create problems with sexual intimacy at night, as indicated by a majority of the patients in our study. Patients and their partners should be counseled on strategies to maximize their compliance without compromising intimacy. Our results support those of other recent studies challenging the traditional view that chronic illness is always associated with psychosocial and, more specifically, sexual maladjustment (4, 18, 22). The multidisciplinary health care professionals involved with care of patients with thalassemia at our hospital encourage patients to take responsibility for their health and acknowledge the wholistic needs of each individual; we propose that our results from this study support this approach.

No type of contraception is contraindicated in patients with thalassemia, although some authorities suggest that thrombotic risk is increased in women taking the oral contraceptive pill who have had splenectomy (29). This idea is controversial, and in our clinic, splenectomy is not considered to be a contraindication to oral contraceptives, although platelet levels in these patients are carefully monitored. Use of HRT was contraindicated in one of our female patients because she had a hepatoma.

Basic knowledge about contraception was not significantly different across the groups. Public health messages about the benefits of using condoms to guard against sexually transmitted diseases appeared to be effective; more than 80% of the sample considered condoms to be the most effective form of protection. Only 44% of the sample appropriately chose oral contraception and condoms in combination as the most effective form of birth control on our list; no difference was observed between men and women or between case-patients and controls.

Disappointingly, even though the sample was small, the rate of unplanned pregnancy was similar between the case-patient and control groups (Table 2). This finding highlights the need for further education about contraceptive use to minimize unwanted pregnancy or pregnancy at a time of compromised maternal health. Of note, all male and female case-patients who reported experience with unplanned pregnancy believed that women and men with β-thalassemia had reduced fertility, despite their own spontaneous pregnancies; this erroneous attitude may explain why these patients used suboptimal or no contraceptive methods.

Consistent with the findings of Zani et al. (22), we found that persons with thalassemia were as likely as their unaffected peers to desire marriage and children. Furthermore, case-patients were as likely as controls to have had children, supporting the consensus that parenthood is now practical for women and men with β-thalassemia who are well chelated and who may or may not have to undergo fertility assistance (29).
Uncertainty currently exists about the teratogenic effects of desferrioxamine. In six case reports of seven women with β-thalassemia who became pregnant while receiving desferrioxamine and who continued its use to various stages of pregnancy, including two women who used it throughout pregnancy, no teratogenic effects were evident at birth (29, 30). However, it is the current practice of most physicians to cease desferrioxamine use during the first trimester in light of studies showing skeletal deformity in the offspring of rats given desferrioxamine. The uncertainty about the teratogenic effects of desferrioxamine further highlights the need for preconception counseling among women who want to become pregnant and adequate contraception among those who do not (29).

The possible transmission of bloodborne viruses from mother to baby is an important consideration in patients with β-thalassemia. Approximately 25 patients (16%) at Monash Medical Centre are currently infected with hepatitis C virus. Many of these patients reported being highly anxious about the possibility that they may infect their partner; however, to date, no transmission to partners or vertical transmission of the virus by our case-patients has been observed.

Although our case-patients almost unanimously thought it very important that they know the carrier status of their partner before they have children, only 42% of the women considered that very important that they know the carrier status of their partner. A possible explanation for this finding may be re-aggregated by the media about sexual health and development, sexuality with patients and, if necessary, provide accurate reproductive information (31). Clinicians need to raise issues of reproductive health experience of persons with homozygous β-thalassemia, further work is required. Areas of concern include education and employment experiences and aspirations, relationship experience, and management of patient fears and anxieties. Further studies are needed to provide substantial direction for future counseling by health care workers who work with such patients.

CONCLUSIONS

Our findings suggest that persons with thalassemia achieve the same goals as their unaffected peers; our case-patients were as likely as “healthy” controls to have full-time employment, be in a relationship, and be sexually active. As patients with thalassemia enter adulthood, the traditional reasons for poor compliance with therapy must be reconsidered. Such factors as sexual activity should be specifically addressed in general consultations to help motivate patients to remain compliant with iron chelation therapy and therefore stay healthy. Assisting patients to accept, adapt to, or overcome these complications and encouraging patients to consider themselves as normal and healthy yet treatment dependent is one of the most challenging aspects of care of adolescents and young adults with thalassemia.

Although our study provides some insights into the reproductive health experience of persons with homozygous β-thalassemia, further work is required. Areas of concern include education and employment experiences and aspirations, relationship experience, and management of patient fears and anxieties. Further studies are needed to provide substantial direction for future counseling by health care workers who work with such patients.

Acknowledgments: The authors thank Professor Henry Burger, A.O., F.A.A., M.D., B.S., F.R.A.C.P., F.R.A.C.O.G., Prince Henry’s Institute of Reproductive Health, Clayton, Australia, and Professor David De Kretzer, A.O., F.A.A., M.D., F.R.A.C.P., Institute Reproduction and Development, Clayton, Australia for valuable advice in questionnaire development; the Nursing Staff Medical Therapy Unit for support and encouragement in patient recruitment; Dr. Jim Doery, M.Sc., M.D., F.R.C.P.A., M.A.A.C.B., Biochemistry Department, Monash Medical Centre, Clayton, Australia for assistance with biochemistry results; and Monash University Faculty of Medicine for salary support of Vicki Psihogios as this work was carried out for her degree of Bachelor of Medical Science.

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