American Journal of Medical Genetics 120A:447–458 (2003)

Living With Achondroplasia in an Average-Sized World: An Assessment of Quality of Life

Sarah E. Gollust,[1] Richard E. Thompson,[2] Holly C. Gooding,[1] and Barbara B. Biesecker[1]*

[1]*Medical Genetics Branch, National Human Genome Research Institute, National Institutes of Health, Bethesda, Maryland*

[2]*Johns Hopkins University Biostatistics Center, Baltimore, Maryland*

Mutations in the gene encoding fibroblast growth factor receptor 3 cause achondroplasia, the most common form of inherited skeletal dysplasia. Although there are more than 10,000 individuals with achondroplasia living in the United States, there has been little study of their quality of life (QOL). For this study, surveys were collected from 189 individuals affected with achondroplasia (ACH) and 136 unaffected first-degree relatives (FDRs) of affected individuals. Individuals affected with achondroplasia had lower annual income, less education, and were less likely to be married than FDRs. They also differed significantly in their perceptions of achondroplasia, with FDRs believing that achondroplasia is a more serious condition. Total QOL indices and indices in each of four QOL subdomains were significantly lower in affected individuals than in relatives. When controlling for demographic characteristics and affected status, having lower self-esteem scores and perceiving achondroplasia as more serious were the independent factors most highly associated with lower QOL. A qualitative analysis of open responses to questions about the advantages and disadvantages of achondroplasia revealed that individuals were as likely to cite disadvantages relating to social barriers as they were to cite those relating to health and functioning. We interpret the low QOL scores to reflect the social challenges that individuals with achondroplasia regularly experience in the average-sized world. Genetics professionals should considersources of lower QOL for affected individuals in their counseling sessions to acknowledge the relative importance of nonmedical contributions.

KEY WORDS: short stature; quality of life; achondroplasia; self-esteem; illness perception; social functioning

INTRODUCTION

Achondroplasia is a type of skeletal dysplasia occurring in approximately 1 in 25,000 live births [Gorlin et al., 1990]. It is an autosomal dominant condition, caused by a mutation in fibroblast growth factor receptor 3 (FGFR3) on human chromosome 4 [Francomano, 1995]. There are more than 10,000 individuals living with achondroplasia in the United States, of equal frequency in men and women and across all racial and ethnic groups, making it the most common skeletal dysplasia. Individuals with achondroplasia, in addition to having a characteristic appearance of disproportionate short stature, may experience other complications. These include delayed motor milestones as children, otitis media, respiratory difficulty, spinal stenosis, pain, and complications with aging. However, people with achondroplasia have normal cognition and overall physical development and are productive and independent adults [Castiglia, 1996]. While the medical issues and genetics of achondroplasia are well characterized, the ways in which the syndrome affects the lives of affected individuals have not been fully explored.

Individuals with achondroplasia tend to have an adult height of approximately four feet (120–130 cm) [Castiglia, 1996]. Such a notable reduction in height as compared to the average has been hypothesized to have a negative effect on one's quality of life (QOL). Research supports correlations between taller height and personal and professional accomplishments in American society [Wilson et al., 1986; Ablon, 1990]. Our Westernized societal emphasis on height suggests that this highly visible characteristic influences how individuals

*Correspondence to: Barbara B. Biesecker, MS, NHGRI/NIH, 10 Center Drive, Building 10, Room 10C101, Bethesda, MD 20892. E-mail: barbarab@nhgri.nih.gov

Received 23 April 2002; Accepted 2 September 2002

DOI 10.1002/ajmg.a.20127

are perceived and perceive themselves socially. Research over the last 40 years has examined this assumption, identifying the potential relationships between short height and QOL, intelligence, school performance, behavior problems, employment opportunities and other psychosocial factors [Pollitt and Money, 1964; Stace and Danks, 1981a; Gordon et al., 1982; Skuse, 1987; Rieser, 1992]. Such studies have indicated that children with short stature (having dysplasia, growth hormone deficiency, constitutional short stature or Turner syndrome) have significant deficits in social and academic settings [Clopper et al., 1986; Dean et al., 1986; Mitchell et al., 1986; Richman et al., 1986]. Introversion, delayed motor development, academic underachievement, social isolation, reduced self-esteem, and depression have been described in many studies of children with a variety of growth problems, some which included cohorts of children with achondroplasia [Pollitt and Money, 1964; Stace and Danks, 1981b; Gordon et al., 1982; Rieser, 1992]. Short adults and adults with various dysplasias have experienced reduced success in finding employment and in marriage [Stace and Danks, 1981c; Dean et al., 1986; Roizen et al., 1990; Sartorio et al., 1990]. It is widely agreed that the deficits experienced by children and adults with short stature are a result of social factors and psychological and emotional responses rather than medical complications [Stace and Danks, 1981a].

More recent studies of children and adults with short stature indicate that earlier conclusions attributing academic or psychosocial difficulties to cohorts of short children could be the result of referral and sampling bias, inappropriately attributing behavioral and academic problems to short stature [Skuse et al., 1994; Kranzler et al., 2000]. One study suggested that QOL deficits disappear when growth hormone deficient individuals are compared to their normal-statured siblings instead of a control group of unrelated normal-statured individuals [Sandberg et al., 1998]. A study examining the QOL of short adults found that the impact of short stature on QOL is small, not true for all conditions leading to short stature, and in particular cases, may be related to unsuccessful coping [Busschbach et al., 1998]. These contradictions in research findings suggest there is more to learn about the correlations between stature and dimensions of QOL.

Despite the abundance of research on the psychological and social effects of stature in general, there has been limited research on the particular experiences of living with achondroplasia. Some of the relatively few prior studies had methodological shortcomings, including using non-standardized QOL instruments, small numbers, and heterogeneous population groups, consisting of short-statured individuals with chondrodysplasias in addition to individuals with short stature of other causes [Haverkamp and Noeker, 1998]. This is problematic because individuals with chondrodysplasias, having disproportionate appearance as well as pain and other medical complications, might encounter very different psychosocial and health-related dimensions of their condition than individuals who have another type of short stature.

Several recent larger studies have examined psychosocial and health factors for individuals with chondrodysplasias. One study explored health-related QOL, a measure that limits the analysis to those elements likely to be considered within a medical interpretation of health, such as mobility, hearing, eating, pain, and sexual activity. Apajasalo and colleagues found that 121 adults with a variety of chondrodysplasias, including achondroplasia, had a significantly lower overall health-related quality of life (HRQOL) than normal controls [Apajasalo et al., 1998]. Another study examined psychosocial factors in 192 dysplasic individuals (of which 72 had achondroplasia) as well as many of their first-degree relatives (FDRs) [Hunter, 1998b; Hunter, 1998c]. Hunter found moderate increases in depression in adult individuals with chondrodysplasia as compared to their FDRs as well as a significantly lower self-esteem in affected adults as compared to their FDRs. Hunter also measured the satisfaction of various life domains of both individuals affected with dysplasias and unaffected individuals, using a 7-point Likert scale. He found somewhat higher levels of reported satisfaction among affected individuals as compared to parents of affected children [Hunter, 1998a]. Another study investigated the physical, as opposed to the psychosocial, impact of achondroplasia on health. In a population of 437 adults affected with achondroplasia, Mahomed et al. [1998] found that the functional health status of individuals with achondroplasia is not "drastically reduced" compared to the general United States population. This complements earlier data that suggest the negative life characteristics individuals with achondroplasia experience may as often be related to social factors as medical problems.

For this study, we investigated QOL by comparing that of a large population of individuals with achondroplasia to that of a group of FDRs of other individuals with achondroplasia. Parents and siblings of individuals with achondroplasia have direct experience with the condition over the life span but likely perceive its impact differently than those affected. We sought to explore multidimensional QOL in order to investigate the contributions of social, psychological, and health-related factors to QOL. Although QOL instruments have been widely used in clinical research and health policy applications, they have not been used with frequency in medical genetics research.

Tools that examine QOL are heterogeneous and differ according to population and context; in addition, researchers' understanding of the meaning of QOL varies [Ferrans and Powers, 1985; Fayers and Machin, 2000]. Researchers intend to operationalize a variety of concepts when they explore QOL, including happiness, satisfaction, well-being, and health status [Brock, 1993; Smith et al., 1999]. Despite the inconsistencies in QOL instruments, there is general agreement that outside observers are inaccurate judges of patient QOL, sometimes overestimating and sometimes underestimating QOL [Epstein et al., 1989; Fayers and Machin, 2000]. QOL ratings based on public attitudes instead of affected patient measurements may offer a skewed interpretation of the experience of living with a disability [Ubel,

2000]. The tool used in this study was the Ferrans and Powers Quality of Life Index (QLI), which assesses quality of life information from the patients directly. It differs from other QOL instruments by acknowledging patient values, ranking individuals' satisfaction with various life domains along with the perceived importance of each factor [Anderson and Ferrans, 1997].

The specific aims of this study were threefold. First, we sought to determine how individuals with achondroplasia perceive their condition in comparison to FDRs. Affected individuals often collectively judge their condition differently than their FDRs or their health care professionals [Leung et al., 1997]. We hypothesized that individuals with achondroplasia perceive their condition as less serious than their FDRs. Second, we sought to understand the QOL of members of the affected population compared to unaffected FDRs. We hypothesized that the QOL of affected individuals would be lower than that of unaffected relatives. Finally, we sought to incorporate qualitative data reported by affected individuals and relatives into our analysis to supplement our understanding of the experience of living with achondroplasia.

METHODS

This was a cross-sectional survey administered within an intramural National Human Genome Research Institute protocol (#96-HG-0123) and approved by the NHGRI Institutional Review Board. The study was also reviewed and approved by the Medical Advisory Board of the Little People of America (LPA). The mailing list was generated from the membership list of the LPA. Only those members of LPA with achondroplasia and the FDRs were included.

Study Population

Fifteen hundred surveys were distributed in an initial mailing, with 750 sent to individuals with achondroplasia and 750 sent to parents and/or siblings of affected individuals. Surveys were not sent to more than one member of a given household; the respondents were unmatched by family and thus unrelated. One hundred eighty-two completed surveys were returned initially, and a follow-up mailing yielded 147 additional surveys. The total number of participants in the study was 189 affected individuals (ACH) and 136 FDRs (FDR), with response rates of 25.2% for ACH and 18.1% for FDR, for a total population response rate of 21.6%. See Gooding et al. [2002] for more information about this study population.

Study Instrument

The survey instrument included questions assessing participants' sociodemographic information, their perception of achondroplasia, self-esteem, and QOL. Perceptions of the seriousness of achondroplasia were queried using a previously developed measure that asked subjects whether they viewed achondroplasia as a lethal, serious, or not serious disorder [Wertz and Knoppers, 2002]. Four-point Likert scales were developed to assess how strongly participants agreed or disagreed that there are advantages and disadvantages associated with having achondroplasia. Open-ended response fields were provided to allow them to comment on specific advantages and disadvantages. Questions were field tested on a sample of 17 affected individuals for content validity, and several changes were made to questions based on these responses.

The self-esteem scale (SES) used was the Rosenberg self-esteem scale, a 10-item scale asking participants to rank feelings about self-worth, attitudes, and personal abilities on a 4-point Likert scale. The scale was scored by assigning values of 0 or 1 to each of the response choices, and individual scores were created by summing each response. Scores may range from 0 to 10. The SES has a Cronbach's alpha in various populations that ranges from 0.77 to 0.88 [Blascovich and Tomaka, 1993].

The Ferrans and Powers Quality of Life Index (QLI) was used to assess the participants' perceived QOL [Ferrans and Powers, 1985; Ferrans, 1996]. The QLI consists of two 34-item sections, rating items on a 5-point Likert scale ranging from "very satisfied" to "very dissatisfied" and "very important" to "very unimportant." The highest scores result from combinations of *high satisfaction* and *high importance* items, and the lowest result from *high dissatisfaction* and *high importance*. The QLI measures total QOL as well as the QOL in four subdomains: Health and Functioning, Social and Economic, Psychological and Spiritual, and Family. The range of scores for total QOL and each subdomain is 0 to 24, with higher scores indicating higher QOL. Cronbach's alphas for the entire instrument have been reported to range from 0.90 to 0.95 [Anderson and Ferrans, 1997].

Statistical Analysis

Data were analyzed using the Statistical Package for the Social Sciences 10.0 for Macintosh (SPSS, Inc., Chicago IL, 2000) and STATA 6.0 (Stata Corporation, College Park, TX, 1999). In preliminary univariate analysis, *t*-tests, Kruskall-Wallis, and chi-square tests were used to compare the two groups in terms of demographics, self-esteem, illness perception, and QOL. Significant differences in characteristics between the affected and unaffected groups were noted (listed in Tables I through III). Subsequent multivariate regression modeling was performed to assess the association of affected status with QOL outcomes after controlling for covariates, such as gender, education, income, religious participation, marital status, and particular outcomes, such as self-esteem and perception of illness. This multiple regression analysis allowed a determination of whether achondroplasia status has an effect on QOL beyond that of other covariates. If affected status is no longer significant after controlling for these covariates, then this suggests that QOL differences between affected individuals and FDRs may be explainable by group differences in other characteristics that are associated with QOL outcomes, instead of from the condition itself.

TABLE I. Demographic Characteristics of Respondents With ACH and FDRs

	ACH (N = 189)	FDR (N = 136)	*P*-value
Age,* mean (range), in years	40.5 (19–89)	43.5 (20–84)	<0.05
Female	127 (67%)	103 (76%)	0.097
Caucasian	172 (91%)	128 (94%)	0.337
Married*	91 (49%)	121 (89%)	<0.001
Completed college or graduate school*	86 (46%)	80 (59%)	0.02
Employed full-time	100 (53%)	65 (48%)	0.363
Income greater than $50,000*	55 (31%)	98 (73%)	<0.001
Religious attendance at least once/month*	89 (47%)	86 (63%)	0.004

*Affected individuals and relatives differed significantly in age, marital status, education level, income, and religious attendance ($P < 0.05$).

Some of the demographic variables with multiple response categories were dichotomized before univariate and multivariate analyses were performed. These included household income (greater than $50,000/year vs. less than or equal to $50,000 per year), education (completed college versus not completed college), marital status (currently married versus not currently married), and religious service attendance (attending at least once a month vs. attending less than once a month). The perceived severity of achondroplasia was dichotomized for the multivariate regression analysis into "not a serious condition" versus "serious or lethal (deadly) condition."

Because QOL scores tended to be skewed toward higher values, normality transformations were considered. A square transformation of the QOL scores gave data that were closer to a representation of normalized scores. Because self-esteem values were also skewed, self-esteem was dichotomized for univariate analysis into two groups, those having a higher self-esteem of 10 and those having a lower self-esteem of less than 10. There was a slight majority of scores of 10. In the regression analysis, self-esteem was treated as a continuous variable.

For the qualitative component of the study, themes of the reported advantages and disadvantages were identified, corresponding with the subdomains of the QOL instrument: advantages and disadvantages relating to individuals' health and functioning, social and economic factors, psychological and spiritual factors, and family issues and concerns. The responses were coded based on these themes, and the documents and coding were entered into the research software NUD*IST (QSR Pty Ltd., Melbourne, Australia) for further analysis. A second coder (B.B.B.) verified the coding system.

RESULTS

Characteristics of respondents with achondroplasia (ACH) and first-degree relatives of individuals with achondroplasia (FDR) are presented in Table I. Sex, ethnicity, and employment did not differ significantly among the two groups. The sample was predominantly Caucasian and approximately three-fourths of the survey participants were female. The mean age of affected individuals was younger than the FDRs, and the two groups also differed significantly with regards to marriage, education, annual income, and religious service attendance ($P < 0.05$). Of the total sample, 90% were members of the Little People of America (LPA), whereas 10% were not. Of the responding individuals with achondroplasia who were members of LPA, 14% never attended any LPA meetings or activities, 37% attended once or twice a year, and 40% attended LPA events with greater frequency.

Self-Esteem

Individuals with achondroplasia had significantly lower self-esteem (SE) values, with a mean SE of 8.36 ± 2.22 for individuals with achondroplasia and 9.42 ± 1.01 for FDRs ($P < 0.0001$). For individuals with achondroplasia, scores ranged from 0 to 10, while scores for FDRs ranged from 5 to 10. Categorical analysis (SE = 10 or SE < 10) also indicated a significant association between self-esteem and affected status ($X^2 = 15.14$, $P < 0.0001$).

Perception of the Condition

Individuals with achondroplasia were 4.01 times (95% CI = 2.09, 7.72) more likely than relatives to view their condition as "not serious" as opposed to "serious" or "lethal" ($P < 0.001$) when controlling for marital status, age, and opinions about prenatal testing use [Gooding et al., 2002]. Because of this significant difference, the individuals' view of the condition was considered in multivariate analysis along with the other factors that differentiate the FDR and ACH population.

Effect of Characteristics on QOL

All of the characteristics considered above were highly significant with respect to QOL (Table II). Those who were FDRs, had a high self-esteem, were female, were currently married, had at least a college degree, earned more than $50,000 per year, regularly attended religious services and viewed achondroplasia as a less serious condition had a higher QOL than those individuals who had achondroplasia, had a lower self-esteem, were unmarried, had less than a college degree, earned less than $50,000, and perceived achondroplasia as a serious or lethal illness ($P \leq 0.0005$).

Subdomains of QOL

Row 1 of Table II shows the significant differences in quality of life indices (QLI) for individuals with

TABLE II. Mean QOL Indices for Each Dichotomized Group in the Study Population

Characteristic	Dichotomized group	N	Mean QOL (tot) ± SD	*P*-value
Affected status	ACH	189	14.083 ± 3.248	0.0001
	FDR	135	16.149 ± 2.354	
Self-esteem	High (= 10)	153	16.407 ± 2.178	<0.0001
	Low (<10)	171	13.635 ± 3.182	
Gender	Male	94	13.996 ± 3.114	<0.0001
	Female	230	15.331 ± 2.986	
Marital status	Currently married	211	15.575 ± 2.833	0.0001
	Currently unmarried	113	13.766 ± 3.185	
Education	College degree or higher	166	15.498 ± 2.951	0.0004
	Some college or less	157	14.352 ± 3.120	
Annual income	Greater than $50,000	153	15.898 ± 2.475	0.0001
	Less than $50,000	157	13.983 ± 3.282	
Religious attendance	Attendance at least once/month	175	15.765 ± 2.720	0.0001
	Attendance less than once/month	149	13.980 ± 3.204	
Perception of seriousness	Not serious	189	15.500 ± 2.797	0.0005
	Serious or lethal	132	14.263 ± 3.181	

achondroplasia and FDRs, with individuals with achondroplasia having mean QLI of 14.08 ± 3.25 and FDRs having a mean QLI of 16.15 ± 2.35. Table III indicates that these between-group differences were sustained for all four subdomains of the QOL instrument. Individuals with achondroplasia had significantly lower QLI across the four subdomains: Health and Functioning, Social and Economic, Psychological and Spiritual, and Family ($P < 0.0001$). Both groups had the highest QLI in the Family subdomain and the lowest QLI in the Health and Functioning subdomain.

Associations With QOL by Subdomain

Table IV includes forced regression models for the outcomes of QOL total as well as for each of the four subdomains when controlling for all relevant characteristics. All outcomes are the squared QOL-indices (QLI^2). An examination of the first model for QOL total reveals that the variables for self-esteem and perception of severity are both highly significant in the model ($P < 0.005$). Those who viewed achondroplasia as serious or lethal had lower QLI than those who viewed achondroplasia as not as serious, and those with a lower self-esteem had lower QLI than those with a higher self-esteem. Self-esteem and perception of severity were the only characteristics with significant associations with QOL across all four subdomains ($P < 0.05$), with self-esteem having the highest significance.

Affected status, on the other hand, was only modestly significant ($P < 0.05$) for the outcome of QOL total and Health and Functioning QOL, and failed to reach statistical significance for the other QOL subdomain outcomes. This indicates that factors other than whether or not the individual has achondroplasia were more highly associated with an individual's social and economic, psychological and spiritual, and family subdomain of QOL. In particular, affected status was of lowest significance for one's psychological and spiritual QOL ($P = 0.418$). This suggests that for members of the study population, having achondroplasia was not associated with individuals' satisfaction with and perceived importance of the psychological and spiritual dimensions of their lives.

TABLE III. Mean QOL for ACH and FDR Individuals by Subdomain

QOL domain	ACH	FDR	*P*-value
Health and functioning	13.3 ± 3.58	15.7 ± 2.54	<0.001
Social and economic	14.6 ± 3.46	16.2 ± 2.51	<0.001
Psychological and spiritual	14.6 ± 3.85	16.3 ± 2.90	<0.001
Family	15.3 ± 3.83	17.3 ± 2.60	<0.001

Gender was of moderate significance for QOL total ($P < 0.05$), with females having greater QLI than males. In particular, gender was highly significantly associated with an individual's health and fitness QOL and an individual's psychological and spiritual QOL ($P < 0.01$), but not with the other two QOL subdomains.

Interestingly, religious attendance was a covariate of significance across several models. Religious attendance was of moderate significance ($P < 0.05$) for QOL total and Social/Economic QOL, and of high significance ($P = 0.01$) for Psychological/Spiritual QOL. Individuals who attended religious services at least once a month had higher predicted QLI than individuals who attended religious services less than once a month. Not surprisingly, this relationship between religious participation and QOL was especially strong for an individual's QOL relating to psychological and spiritual factors.

Annual income approached significance ($P = 0.69$) for QOL total and was moderately significant ($P < 0.05$) for Health and Functioning QOL, with individuals in households who made more than $50,000 per year having higher QLI than those who made less than $50,000. From Table I, it is clear that FDRs were more likely to have incomes greater than $50,000 per year than individuals affected with achondroplasia, with over 70% of relatives earning more than $50,000 and only 30% of affected individuals earning a comparable amount.

TABLE IV. Regression Coefficient Values[†] (With 95% Confidence Intervals) and Corresponding *P*-Values for Characteristics Related to QOL Outcomes (QLI^2)

	QOL (total)		Health and functioning		Social and economic		Psychological and spiritual		Family	
Characteristics	Coefficient	*P*	Coefficient	*P*	Coefficient	*P*	Coefficient	*P*	Coefficient	*P*
Affected status	19.3 (1.0, 37.6)	0.039*	22.3 (3.0, 41.6)	0.024*	17.2 (−4.5, 39.0)	0.120	9.2 (−13.2, 31.6)	0.418	20.5 (−3.3, 44.3)	0.092
Self-esteem	21.0 (16.1, 26.0)	<0.001**	21.3 (16.0, 26.5)	<0.001**	15.9 (10.0, 21.8)	<0.001**	25.4 (19.3, 31.5)	<0.001**	20.1 (13.6, 26.6)	<0.001**
Gender	22.3 (4.6, 40.0)	0.014*	25.1 (6.4, 43.7)	0.009**	15.8 (−5.2, 36.9)	0.141	29.0 (7.4, 50.7)	0.009**	7.7 (−15.4, 30.8)	0.512
Marital status	1.9 (−17.8, 21.6)	0.847	2.24 (−18.6, 23.0)	0.832	−7.07 (−30.6, 16.4)	0.554	−0.2 (−24.4, 24.0)	0.987	22.1 (−3.7, 47.9)	0.093
Education	9.7 (−6.3, 25.7)	0.234	10.9 (−6.0, 27.8)	0.204	18.5 (−0.5, 37.6)	0.057	8.9 (−10.7, 28.5)	0.372	−10.1 (−31.0, 10.8)	0.341
Income	16.9 (−1.3, 35.2)	0.069	21.3 (2.0, 40.5)	0.030*	19.0 (−2.7, 40.8)	0.086	3.9 (−18.4, 26.3)	0.729	18.0 (−5.8, 41.8)	0.137
Religious attendance	18.1 (1.8, 34.4)	0.030*	11.6 (−5.6, 28.8)	0.186	22.6 (3.1, 42.0)	0.023*	26.5 (6.5, 46.5)	0.010**	14.5 (6.8, 35.8)	0.182
Perception of severity	−24.4 (−40.6, −8.2)	0.003**	−23.3 (−40.5, −6.24)	0.008**	−26.0 (−45.3, −6.7)	0.009**	−20.7 (−40.2, −0.4)	0.046*	−28.7 (−49.8, −7.5)	0.008**
R^2	0.4136		0.4006		0.2684		0.3532		0.2913	

*Indicates a significant association with the QOL outcome ($P < 0.05$).
**Indicates a highly significant association with the QOL outcome ($P \leq 0.01$).
[†]Coefficients represent the predicted difference in the square of the QOL score between variable categories, i.e., in column 1, FDRs have a mean QOL^2 that is 19.3 points higher than affected individuals' score.

The relationship of income with QOL seemed particularly important for an individual's QOL relating to health and functioning concerns ($P = 0.03$), and least important for that relating to psychological and spiritual concerns ($P = 0.729$).

The relationship between self-esteem and QOL deserves further exploration (Table V). Table II indicates that self-esteem is highly significant with respect to mean total QOL ($P < 0.0001$). Furthermore, the correlation coefficient for QOL-total and self-esteem indicates a strong correlation between the two measures ($r = 0.6051$). When self-esteem is removed from the regression analysis, as in model 2 in Table V, the percent of variability in QOL tot^2 explained by the independent variables (R^2) decreases from 41% to 27%, suggesting that self-esteem accounts for much of the variability in total QOL. When self-esteem is not controlled for, as in model 2, an individual's education level and income level become statistically significant factors associated with QOL. Most interestingly, when self-esteem is removed from the model, affected status becomes highly significant. These findings suggest that having a lower QOL is highly associated with having a lower self-esteem, which in turn is highly associated with having achondroplasia.

Advantages and Disadvantages

Individuals with achondroplasia were 2.06 times (95% CI = 1.19, 3.57) more likely than relatives to agree with the statement *"There are advantages that come with having achondroplasia"* when controlling for demographics, interest in prenatal diagnosis, and opinions about prenatal testing ($P < 0.05$) [Gooding et al., 2002]. Individuals with achondroplasia were 14.36 times (95% CI = 1.89, 108.93) more likely to disagree with the statement *"There are disadvantages that come with having achondroplasia"* ($P < 0.05$) when controlling for demographics, interest in prenatal diagnosis, and opinions about prenatal testing. In addition to these differences in their likelihood to perceive positive and negative aspects of the condition, affected individuals and FDRs differed in the particular aspects they described.

Figure 1 shows the different types of advantages and disadvantages that individuals with achondroplasia and FDRs described. Advantages and disadvantages were classified into four categories, paralleling the domains of the QOL instrument: health and functioning (HF), social and economic (SE), psychological and spiritual (PS), and family (FAM).

Most commonly cited disadvantages for both groups were health/functioning- and social/economic-related, with a similar frequency of responses given for disadvantages related to difficulties with health and daily functioning and those related to difficulties with a social dimension. Individuals with achondroplasia cited health and functioning issues as disadvantages more frequently than did FDRs.

The pattern of advantages cited by individuals with achondroplasia and FDRs was more variable than that of the disadvantages. More individuals affected with achondroplasia stated advantages related to social

TABLE V. Regression Coefficient Values (With 95% Confidence Intervals) and Corresponding *P*-Values for Two Regression Models of QOL Outcomes (Total QOL[2]), Controlling for Self-Esteem (Model 1), and Not Controlling for Self-Esteem (Model 2)

	Model 1		Model 2	
Variable	Coefficient	*P*	Coefficient	*P*
Affected status	19.3 (1.0, 37.6)	0.039*	37.0 (17.3, 56.6)	<0.001**
Self-esteem	21.0 (16.1, 26.0)	<0.001**		
Gender	22.3 (4.6, 40.0)	0.014*	27.5 (8.6, 46.3)	0.004**
Marital status	1.9 (−17.8, 21.6)	0.847	13.6 (−7.1, 34.4)	0.198
Education	9.7 (−6.3, 25.7)	0.234	19.4 (2.4, 36.4)	0.025*
Income	16.9 (−1.3, 35.2)	0.069	23.0 (3.3, 42.7)	0.022*
Religious attendance	18.1 (1.8, 34.4)	0.030*	22.4 (4.8, 40.0)	0.013*
Perception of severity	−24.4 (−40.6, −8.2)	0.003**	−36.1 (−53.3, −18.9)	<0.001**
R^2	0.4136		0.2690	

*Indicates a significant association with the QOL outcome ($P < 0.05$).
**Indicates a highly significant association with the QOL outcome ($P < 0.01$).

interactions and friendships than did FDRs. Relatives, on the other hand, were more likely to state advantages related to psychological or spiritual traits such as having a special outlook on life, a unique perspective on diversity, personal strength, and a heightened sense of compassion.

A summary of the most common responses given by both affected individuals and FDRs is provided in Table VI. The frequency of responses is given rather than the number of individuals who gave the responses, as individuals cited more than one advantage and disadvantage.

There was great variability in respondents' perceptions of advantages and disadvantages of living with achondroplasia. There was a common thread of ambivalence expressed by individuals: social factors had both positive and negative impacts on the daily lives of affected individuals.

Many individuals (n = 37) expressed the sentiment that disadvantages arise not intrinsically from the condition of achondroplasia, but from when the "average-sized world" presents conflicts:

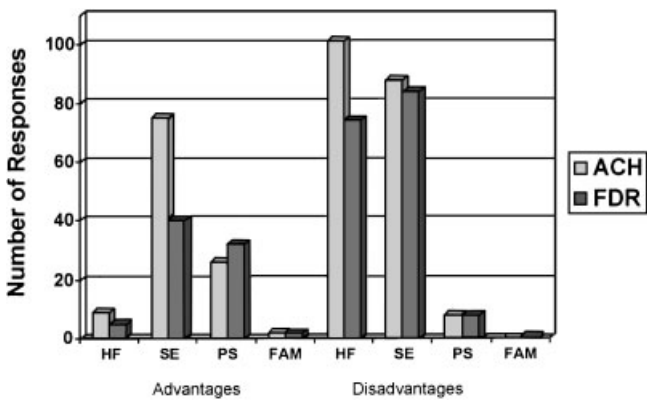

Fig. 1. Number of responses given by individuals with ACH and FDRs for Each Category† of Advantages and Disadvantages. †HF, health and functioning; SE, social and economic; PS, psychological and spiritual; FAM, family.

- *"The only disadvantage I see is that people with achondroplasia live in a world that is built for the average-sized."* (ACH female)
- *"People with achondroplasia are limited in society from having any advantages . . . simply because the 'normal' people's world is unsustainable for little people."* (ACH male)
- *"You have to make accommodations because the world is made for the average person."* (ACH female)
- *"Society creates circumstances that make short stature into a disadvantage"* (ACH female)
- *"The world is not built for me."* (ACH female)
- *"People with achondroplasia must live in an average size world."* (FDR female)
- *"[I'm] living in a world that doesn't fit."* (ACH female)

A few (n = 8) individuals used downward social comparison to cope with their condition [Festinger, 1954; Wood et al., 1985]. Individuals stated that their condition was "not as bad" as a variety of other, more debilitating conditions:

- *"It's not the worst thing in the world."* (FDR male)
- *"There are many, many worse conditions than achondroplasia."* (ACH female)
- *"In regards to other kind of dwarfism, health-wise, this is the best."* (ACH female)

Other individuals (n = 11) expressed the normalizing statement that every life experience has both advantages and disadvantages:

- *"There are disadvantages to life for average-sized people as well."* (ACH female)
- *". . . Almost every trait/condition has disadvantages and most people have or get something, and achondroplasia has pluses too."* (ACH female)
- *"Everyone is born with some kind of disadvantage."* (ACH female)

Responses, in general, provide data to understand factors that may contribute to the measured lower levels

TABLE VI. Common Advantages and Disadvantages of Achondroplasia Stated by FDRs and Affected Individuals, Number of Times Cited

	N
Advantages	
Strength of spirit or character	25
Memorable	25
Friendships improved, allows for meeting more people	24
Make people more aware, sensitive, tolerating of diversity	21
Center of attention/noticeable	19
Can fit into small places, reach things lower	14
Gains priority, favoritism, from being different	11
Possesses patience and grace	10
Occupation helped by distinction, humor	6
Disadvantages	
Medical problems, general	66
Difficulty reaching objects	56
Teasing, staring, too much attention	37
Limited career choices, issues of unemployment and reduced income	32
General discrimination or prejudice	32
General physical functioning impaired	30
General deficits in standard of living relating to public accommodations	26
Specific limited activities (sports, etc.)	26
Specific difficulties finding clothes and shoes	26
Inaccurate expectations by others or stigma	21
Difficulties in personal relationships, dating, socializing	18
Inappropriate comments or rudeness	13
Pain, specific reference to	7

of QOL. Furthermore, they provide insight into the use of healthy coping mechanisms, such as downward social comparison and normalization, that may help affected individuals achieve a higher QOL than they would otherwise achieve.

DISCUSSION

The disproportionate short stature and characteristic facial features of individuals with achondroplasia cause them to differ noticeably in appearance from individuals of average stature. This study reveals differences between individuals with achondroplasia and average-statured FDRs in other, less visible, attributes as well. Individuals with achondroplasia who participated in this study have lower self-esteem, less education, lower annual incomes, and are less likely to have a spouse, findings that support earlier studies of adults with growth deficiencies [Stace and Danks, 1981c; Dean et al., 1986; Roizen et al., 1990; Sartorio et al., 1990; Hunter, 1998c]. The lower QOL observed, however, contrasts with an earlier study which suggested that affected individuals actually have higher levels of life satisfaction than FDRs [Hunter, 1998a]. The significantly lower QOL indices across all subdomains as well as significantly lower self-esteem levels identified in this study sample suggest that these individuals with achondroplasia experience life quite differently than do the average-statured relatives. This study sought to examine the presence of these differences, investigate the variability in perception between those affected and those unaffected but informed, and analyze how physical factors related to achondroplasia—such as short stature, shortened limbs, and medical problems—interact with psychosocial factors and demographics to influence the overall QOL for individuals with achondroplasia.

Individuals with achondroplasia experience stigma beyond that of their short stature. As anthropologist Joan Ablon suggests, dwarf individuals are associated with a mystique not attributable to other types of physical and mental differences [Ablon, 1990]. The public, influenced by mythical media representations of dwarfs, has developed inaccurate expectations of dwarf individuals' abilities and limitations. Confronting these expectations presents a major challenge for dwarf individuals and their families. The qualitative data yielded in this study reflect the predominance of these social challenges. Many individuals commented on disadvantages of achondroplasia that related to public perceptions of their condition: teasing, staring, inaccurate expectations, rudeness, and discrimination. As one participant phrased his most palpable challenge: "we are thought to have minds in range of our size; on occasions we are not considered to have thoughts or opinions or feelings." Another stated, "the biggest disadvantage I experience are people's reactions to me." Dramatic short stature and a disproportionate appearance, compounded by inaccurate culturally derived characteristics of dwarfs, might translate into differences between how the public versus affected individuals perceive the condition. This study suggests that such differences in perception exist even between individuals affected with achondroplasia and FDRs, individuals who have greater experience with the condition than the general public.

Self-Esteem

Individuals with achondroplasia, dealing with the many social and physical disadvantages related to their

condition, seemed to have significantly lower values of self-esteem as well as lower QOL than relatives. In this study, it is not possible to uncouple the concepts of self-esteem and QOL: having a lower self-esteem might lead to a lower QOL; on the other hand, a low QOL might lead to a lower self-esteem. The regression analysis suggests that the variability in QOL in the total study population is highly influenced by self-esteem. Self-esteem has been suggested to relate to successful adaptation to a disability. Li and Moore [1998] found that self-esteem was among the most important predictors of disability acceptance when controlling for demographics, disability characteristics including onset, chronic pain, and presence of multiple disabilities, and psychosocial factors including both self-esteem and emotional support. Greater acceptance of one's disability, as indicated by higher self-esteem, might reflect an individual's ease and ability to integrate into society [Li and Moore, 1998]. Alternatively, those who have managed to adapt with greater ease might experience higher self-esteem and have a more positive perception of their condition or disability. This study offers additional evidence to support the concept that self-esteem has an important role in shaping and reflecting the QOL experienced by individuals with disabilities.

Perception of Achondroplasia

Beyond self-esteem, perception of seriousness was the second most significant characteristic associated with QOL outcomes. Even unaffected individuals who viewed the condition as "serious or lethal" had a lower QOL than those unaffected relatives who viewed the condition as "not serious." One explanation for the correlation between a more severe outlook on achondroplasia and a lower QOL is that individuals who judge the condition as more serious may actually face a greater magnitude of physical problems either personally or in their close relatives with achondroplasia, thus lowering their perception of their QOL. Alternatively, individuals who perceive their QOL to be low might attribute this to having achondroplasia; as a result, these individuals might have a more serious perception of the condition due to its impact on their life. Capelli et al. [1989] offer one explanation of the factors relating to illness perception. They found that an adolescent's perception of his/her physical health and the reaction of family members were important sources of discord in the family. Research supports the idea that a family member's chronic disease influences the QOL of other family members [Rees et al., 2001; Frank, 2002]. Family members who perceive their affected siblings or children as having more serious problems may have a more negative perception of their own QOL as a result. Indeed, characterization of achondroplasia as more serious had a significant association ($P = 0.008$) with a lower Family QOL, more significant than its associations with the social/economic or psychological/spiritual domains of QOL. This suggests that individuals' perceptions of a chronic condition affecting a member of the family carry over to others in the family and have an impact on the entire system.

Religious Participation

Religious attendance was significant in three of the five models of QOL outcomes. Recent literature reviews suggest that individuals with physical chronic illnesses may use religion to cope, positively affecting adjustment [Dein, 1997] and that religious participation leads to healthier individuals and may even extend the life span [Koenig, 2000]. The results of the present study suggest a relationship between religious attendance and QOL, as people who attended services with greater frequency had a higher QOL. Frequent religious service attendance might also represent an individual's level of involvement in a community. Community involvement may be associated with greater social acceptance of differences, likely to facilitate a higher QOL as we observed.

Interpretation of Affected Status and QOL

Although individuals with achondroplasia in this study population have significantly lower QOL than relatives, one cannot simply assert that having achondroplasia predicts or explains lower QOL indices. The characteristics discussed above—self-esteem, perception of the condition, and even religious attendance—?were all more significantly associated with QOL domains than was affected status for all QOL outcomes other than health and functioning QOL. Although these identified psychosocial and demographic factors were likely to be correlated with those individuals affected with achondroplasia, it is of significance that affected status alone is not the strongest indicator of a reduced QOL. The qualitative data enhance this interpretation. The cited disadvantages that accompany a lower QOL were often social in nature, arising not simply from health or functioning concerns attributable to achondroplasia, but to issues that may be interpreted as resulting from social dimensions. These data add evidence to support an idea common in disability theory: that disability is defined by social prejudice, not by physical or medical facts [Koch, 2001].

The implications of the finding that reductions in QOL are not entirely explained by affected status are numerous. An individual's physical status of having or not having achondroplasia is unchangeable. However, factors that are socially-based are more plastic. There may be factors that can be identified and changed to directly improve an individual's QOL. Some cited disadvantages such as being teased, discriminated against, or stigmatized might be mediated with public education. If the general public were more familiar with the condition in an accurate way, discrimination and negative stereotyping might be reduced, potentially leading to improvements in affected individuals' life experiences. Further, individuals with achondroplasia may benefit from assertiveness training or psychotherapy to develop means to successfully ward off the impact of others' negative responses. Ideally, such resources would be offered to the family and begin in childhood, involving the school environment.

Policy Implications

These findings have further implications specifically for policy, as public education and counseling services cannot mitigate all negative attributes of achondroplasia. Daily functioning was severely hampered in affected individuals, and was a persistent comment by those personally affected by achondroplasia. Specific concerns cited frequently related to public access to supermarkets, restaurants, ATMs, gas stations, etc. This information could be used to yield better ways to increase public accessibility for a range of individuals with disabilities. As these results suggest that such social factorscontribute to a lower QOL, it is imperative to work toward more equitable access to public facilities. In addition, the demographic inequities observed, such as the lower income, lower levels of education, and difficulties with employment, may signify the existence of inaccurate cognitive and practical expectations that have been placed on individuals with achondroplasia. Such misunderstandings of an individual's potential might translate into their lower annual income, which has a significant relationship with lower QOL. The discrepancies revealed by this study suggest that there is significant work to be done in the policy setting to ensure equal access to employment and educational opportunities to individuals who are physically different from average individuals, promoting less discrepant incomes between affected and unaffected groups, and potentially improving QOL.

Clinical Implications

These findings have implications for the clinical setting as well. It has been suggested that health care professionals view disabilities as undesirable, tending to underestimate the QOL of individuals with disabilities and attributing this reduction to medical factors as opposed to social [Asch, 1998; Kirschner et al., 2000]. The present study ascertains affected individuals' QOL from the individuals themselves, and can be used to directly inform clinical practice to meet client needs. Although genetics health care providers should advise patients of the frequent physical limitations and medical problems associated with a diagnosis of achondroplasia, it is important for them to recognize that a number of difficulties are social in nature. Parents may seek advice about how best to handle the social conflicts they are very likely to encounter as their children's differences clash with those of individuals in the average-statured world. In addition, as self-esteem appears to play an important role in an individual's general QOL, intervention to help foster a more positive self-concept early in life might prove to increase an individual's QOL. Health care professionals have a responsibility to have a more accurate understanding of achondroplasia and other disabilities, incorporating physical, medical, and social factors, and to refer families to appropriate counseling resources even in the absence of obvious dysfunction.

Study Limitations

The overall study response rate was 21.6%, suggesting that respondents may not be representative of individuals with achondroplasia or unaffected relatives, either within the Little People of America or outside this organization. Secondly, as the two groups were not matched by age, between-group comparisons are speculative. In addition, over 90% of the participants were Caucasian. Since achondroplasia occurs in all ethnic groups, respondents represent a subset of one ethnically biased perspective. Individuals of other ethnic backgrounds may have different concerns relating to QOL and their social interactions. Finally, this study relied on self-reporting of diagnosis, and thus may have included respondents who do not have achondroplasia.

By only surveying those individuals from the Little People of America database, this study may be biased toward people who are more likely to have greater knowledge about achondroplasia and greater access to the community of dwarf individuals. It is important to note that membership in LPA does not necessarily indicate active participation, as individuals may be added to their mailing list by their health care providers; 14% of respondents with achondroplasia were not involved at all in LPA activities. Those who actively participate in LPA are generally middle-class, Caucasian, and are predominantly female [Ablon, 1984], reinforcing the limitation that neither this study population nor the LPA at large reflect all individuals with achondroplasia in the United States. Furthermore, LPA may have an influence on QOL as a result of the distinct cultural features of LPA. Ablon suggests that individuals who have exposure to other individuals who have dwarfing syndromes experience a destigmatization process that could lead to a greater degree of acceptance and successful coping and lead to a higher QOL [Ablon, 1981]. This process is very difficult, however, and involves an emotionally-laden "confrontation" with the individual's identity, appearance, and difficulties with functioning. Thus, individuals who have more exposure to other similarly affected individuals might exaggerate the effect that their physical difference has on their social and physical functioning, actually downplaying their QOL as a result.

Summary

Despite the limitations, this study suggests that short stature, along with other social and physical factors, constrains life with achondroplasia. Society's perception of individuals with achondroplasia, combined with the physical and medical hardships experienced daily in trying to adjust to a world that "doesn't fit," create significant challenges for affected individuals. This study identifies several areas of needed future research. There is much work to be done exploring the relationships between self-esteem and QOL and understanding how the two concepts interact. In addition, additional empirical work should be pursued to examine the differential perceptions of disabilities of affected individuals, family members, health care professionals, and the general public as well as to examine evidence of the social contributions of disabilities. Additional research about the influence of support groups on QOL would be

valuable. Finally, future research might investigate potential clinical or social interventions that could improve the QOL experienced by individuals with achondroplasia or other disabilities.

ACKNOWLEDGMENTS

We thank Karina Boehm for her work developing the survey instrument, Clair Franomano for facilitating approval from the Medical Advisory Board of the LPA, and Shelley Niwa of Westat, Inc. for her assistance with data management. Our sincere gratitude goes to those individuals with achondroplasia and relatives who completed the survey.

REFERENCES

Ablon J. 1981. Dwarfism and social identity: Self-help group participation. Soc Sci Med 15B:25–30.

Ablon J. 1984. Little people in America: The social dimension of dwarfism. New York: Praeger.

Ablon J. 1990. Ambiguity and difference: Families with dwarf children. Soc Sci Med 30:879–887.

Anderson JS, Ferrans CE. 1997. The quality of life of persons with chronic fatigue syndrome. J Nerv Ment Dis 185:359–367.

Apajasalo M, Sintonen H, Rautonen J, Kaitila I. 1998. Health-related quality of life of patients with genetic skeletal dysplasias. Eur J Pediatr 157:114–121.

Asch A. 1998. Distracted by disability. Camb Q Healthc Ethics 7:77–87.

Blascovich J, Tomaka J. 1993. Measures of self-esteem. In: Robinson JP, Shaver PR, Wrightsman LS, editors. Measures of personality and social psychological attitudes. Ann Arbor: Institute for Social Research. pp 115–160.

Brock D. 1993. Quality of life measures in health care and medical ethics. In: Nussbaum MC, Sen A, editors. The quality of life. Oxford: Clarendon Press.

Busschbach JJ, Rikken B, Grobbee DE, De Charro FT, Wit JM. 1998. Quality of life in short adults. Horm Res 49:32–38.

Capelli M, McGrath P, Heick C, Macdonald N, Feldman W, Rowe P. 1989. Chronic disease and its impact. J Adolesc Health Care 10:283–288.

Castiglia PT. 1996. Achondroplasia. J Pediatric Health Care 10:180–182.

Clopper RR, MacGillivray MH, Mazur T, Voorhess ML, Mills BJ. 1986. Post-treatment follow-up of growth hormone deficient patients: Psychosocial status. In: Stabler B, Underwood LE, editors. Slow grows the child: Psychosocial aspects of growth delay. Hillsdale: Lawrence Erlbaum Associates. pp 83–96.

Dean HJ, McTaggert TL, Fish DG, Friesen HG. 1986. Long-term social follow-up of growth hormone deficient adults treated with growth hormone during childhood. In: Stabler B, Underwood LE, editors. Slow grows the child: psychosocial aspects of growth delay. Hillsdale: Lawrence Erlbaum Associates. pp 73–82.

Dein S. 1997. Does being religious help or hinder coping with chronic illness? A critical literature review. Palliat Med 11:291–298.

Epstein A, Hall J, Tognetti J, Son L, Conant L. 1989. Using proxies to evaluate quality of life. Med Care 27:S91–S98.

Fayers PM, Machin D. 2000 Quality of life: Assessment, analysis and interpretation. West Sussex: Wiley.

Ferrans CE. 1996. Development of a conceptual model of quality of life. Sch Inq Nurs Pract 10:293–304.

Ferrans CE, Powers MJ. 1985. Quality of life index: Development and psychometric properties. Ans-Adv Nurs Sci 8:15–24.

Festinger L. 1954. A theory of social comparison processes. Hum Relat 7:117–140.

Francomano CA. 1995. The genetic basis of dwarfism. N Engl J Med 332: 58–59.

Frank A. 2002. Impact of chronic illness on the family. J R Soc Med 9:57–58.

Gooding HC, Boehm K, Thompson R, Hadley D, Francomano C, Biesecker B. 2002. Issues surrounding prenatal diagnosis for achondroplasia. Prenat Diagn 22:933–940.

Gordon M, Crouthamel C, Post EM, Richman RA. 1982. Psychosocial aspects of constitutional short stature: Social competence, behavior problems, self-esteem, and family functioning. J Pediatr 101:477–480.

Gorlin RJ, Cohen MM, Levin LS. 1990. Syndromes of the head and neck. New York: Oxford University Press. pp 171–174.

Haverkamp F, Noeker M. 1998. Short stature in children—A questionnaire for parents: A new instrument for growth disorder-specific psychosocial adaptation in children. Qual Life Res 7:447–455.

Hunter AG. 1998a. Some psychosocial aspects of nonlethal chondrodysplasias: I. Assessment using a Life-Styles Questionnaire. Am J Med Genet 78:1–8.

Hunter AG. 1998b. Some psychosocial aspects of nonlethal chondrodysplasias: II. Depression and anxiety. Am J Med Genet 78:9–12.

Hunter AG. 1998c. Some psychosocial aspects of nonlethal chondrodysplasias: III. Self-esteem in children and adults. Am J Med Genet 78: 13–16.

Kirschner K, Ormond K, Gill C. 2000. The impact of genetic technologies on perceptions of disability. Qual Manage Health Care 8:19–26.

Koch T. 2001. Disability and difference: Balancing social and physical constructions. J Med Ethics 27:370–376.

Koenig H. 2000. Religion, spirituality, and medicine: Application to clinical practice. JAMA 284:1708.

Kranzler JH, Rosenbloom AL, Proctor B, Diamond FB, Jr., Watson M. 2000. Is short stature a handicap? A comparison of the psychosocial functioning of referred and nonreferred children with normal short stature and children with normal stature. J Pediatr 136:96–102.

Leung SS, Steinbeck KS, Morris SL, Kohn MR, Towns SJ, Bennett DL. 1997. Chronic illness perception in adolescence: Implications for the doctor-patient relationship. J Paediatr Child Health 33:107–112.

Li L, Moore D. 1998. Acceptance of disability and its correlates. J Soc Psychol 138:13–25.

Mahomed NN, Spellmann M, Goldberg MJ. 1998. Functional health status of adults with achondroplasia. Am J Med Genet 78:30–35.

Mitchell CM, Johanson AJ, Joyce S, Libber S, Plotnick L, Migeon CJ, Blizzard RM. 1986. Psychosocial impact of long-term growth hormone therapy. In: Stabler B, Underwood LE, editors. Slow grows the child: Psychosocial aspects of growth delay. Hillsdale: Lawrence Erlbaum Associates. pp 97–110.

Pollitt E, Money J. 1964. Studies in the psychology of dwarfism. I. Intelligence quotient and school achievement. Pediatrics 64:415–421.

Rees J, O'Boyle C, MacDonagh R. 2001. Quality of life: Impact of chronic illness on the partner. J R Soc Med 94:563–566.

Richman RA, Gordon M, Tegtmeyer P, Crouthamel C, Post EM. 1986. Academic and emotional difficulties associated with constitutional short stature. In: Stabler B, Underwood LE, editors. Slow grows the child: Psychosocial aspects of growth delay. Hillsdale: Lawrence Erlbaum Associates. pp 13–26.

Rieser PA. 1992. Educational, psychologic, and social aspects of short stature. J Pediatr Health Care 6:325–332.

Roizen N, Ekwo E, Gosselink C. 1990. Comparison of education and occupation of adults with achondroplasia with same-sex sibs. Am J Med Genet 35:257–260.

Sandberg DE, MacGillivray MH, Clopper RR, Fung C, LeRoux L, Alliger DE. 1998. Quality of life among formerly treated childhood-onset growth hormone-deficient adults: A comparison with unaffected siblings. J Clin Endocrinol Metab 83:1134–1142.

Sartorio A, Morabito F, Peri G, Conti A, Faglia G. 1990. The social outcome of adults with consitutional growth delay. J Endocrinol Invest 13:593–595.

Skuse D. 1987. The psychological consequences of being small. J Child Psychol Psychiatry 28:641–650.

Skuse D, Gilmour J, Tian CS, Hindmarsh P. 1994. Psychosocial assessment of children with short stature: A preliminary report. Acta Paediatrica Suppl 406:11–16 (Discussion 17).

Smith KW, Avis NE, Assmann SF. 1999. Distinguishing between quality of life and health status in quality of life research: A meta-analysis. Qual Life Res 8:447–459.

Stace L, Danks DM. 1981a. A social study of dwarfing conditions. I. The reactions and questions of parents of children with bone dysplasias. Aust Paediatr J 17:167–171.

Stace L, Danks DM. 1981b. A social study of dwarfing conditions. II. The experience of children with bone dysplasias, and of their parents. Aust Paediatr J 17:172–176.

Stace L, Danks DM. 1981c. A social study of dwarfing conditions. III. The social and emotional experiences of adults with bone dysplasias. Aust Paediatr J 17:177–182.

Ubel PA. 2000. Societal value, the person trade-off, and the dilemma of whose values to measure for cost-effectiveness analysis. Health Econ 9:127–136.

Wertz DC, Knoppers BM. 2002. Serious disorders: can or should they be defined? Am J Med Genet 108:29–35.

Wilson DM, Duncan PM, Dornbusch SM, Ritter PL, Rosenfeld RG. 1986. The effects of growth on intellectual function in children and adults. In: Stabler B, Underwood LE, editors. Slow grows the child: Psychosocial aspects of growth delay. Hillsdale: Lawrence Erlbaum Associates. pp 139–150.

Wood J, Taylor SE, Lichtman R. 1985. Social comparison in adjustment to breast cancer. J Pers Soc Psychol 49:1169–1183.