

Supplemental Appendix

Supplemental information on psychometric testing

The Medical Outcomes Study 36-item short-form health survey (SF-36) is the most widely used generic instrument to assess health-related subjective health status (17). The SF-36 consists of eight multi-item health domains. These include: limitations in physical activities because of health problems (physical functioning, PF), limitations in usual role activities because of physical health problems (role functioning physical, RP), bodily pain (BP), general health perception (GH), vitality (energy and fatigue) (VT), limitations in social activities because of physical or emotional problems (social functioning, SF), limitations in usual role activities because of emotional problems (role functioning emotional, RE) and general mental health (psychological distress and well-being) (mental health, MH). The domain scores range from 0-100, with higher values indicating a better subjective health status

The Hospital Anxiety and Depression Scale (HADS) is a 14-item rating scale designed to measure anxiety and depression in physically ill individuals (18;18). Anxiety and depression is assessed by seven items rated on a four-point scale from 0-3; thus the maximum score of each subscale is 21. Higher scores indicate more severe anxiety and depression. Scores >8 are considered as indicative of significant impairment and scores >11 indicate major impairment as in psychiatric disorders, e.g. major depression (Ref. HADS).

The CAH well-being questionnaire is a non-validated questionnaire specifically developed for this study, to address issues that might concern adult CAH patients. Patients were asked to rate how they felt about six statements (“I am unhappy with my height”, “I am concerned about my weight”, “I am worried about long-term health risks of CAH,” “I am worried about being able to have children”, “I am unhappy with my sex-life”, “I feel self-conscious about my body hair”) by referring to a four-level Likert scale (“All the time”, “Most of the time”, “Sometimes”, “Never”).

Sexual function in men was assessed employing the 5-item short form of the International Index of Erectile Function (IIEF-5) (19). Each of the five items is scored on a five-point scale where lower values represent poorer sexual function. Erectile dysfunction is classified into five categories based on the overall score: severe (1-7), moderate (8-11), mild to moderate (12-16), mild (17-21), and none (22-25).

Sexuality in women was assessed by the 9-item short version of the McCoy Female Sexuality Questionnaire (MFSQ-9) (20). MFSQ-9 measures four dimensions of female sexuality with all items rated on a 7-point scale ranging from 1 (absent, very low) to 7 (extremely high). Dimensions include sexual interest (4 items, min-max score range 4-28), vaginal lubrication (2 items, score 2-14), orgasm (1 item, score 1-7), and sex partner (2 items, score 2-14). Higher scores indicate better, more complete sexual function.

Patients with concomitant disease relevant to subjective health status were excluded from overall psychometric data analysis. Self-reported concomitant disease was independently scored as either relevant or irrelevant by two different investigators (W.A., R.J.R.). Concomitant disease was considered relevant when scored as such by either one or both physicians, resulting in exclusion of the patient’s data. This criterion applied to six patients and included the following diagnoses: chronic lymphocytic leukaemia, Turner’s syndrome, traumatic brain injury, thyrotoxicosis, total colectomy due to familial polyposis coli, and primary sclerosing cholangitis.