Objective: Spinal muscular atrophy (SMA) is common. The prevalence of SMA in southern Chinese is 1 in 53,000. The clinical course is variable. The traditional classification of SMA includes age of onset, age of death, achievement of motor milestones, and ambulatory status as criteria. There was a lack of inclusion of the best lifetime functional status of any child with SMA. With the advances in medical care, the life expectancy and ambulatory status of patients with SMA have improved. The objective of this study was to assess the survival pattern, ambulatory status, and functional status of children with SMA. Methods: Patients with SMA were recruited from the neuromuscular clinic of the Duchess of Kent Children's Hospital, which is a university-affiliated hospital, and the Families of SMA in Hong Kong. By September 2002, 102 SMA cases had been registered in the Duchess of Kent Children's Hospital neuromuscular clinic and Families of SMA registry, and 83 patients were analyzed. Among them, 39 were recruited for the administration of Functional Independence Measure for Children (WeeFIM), an assessment tool for functional status that has been previously validated by us for Chinese children. The diagnosis of SMA was made from clinical history, serum muscle enzyme, electromyography, muscle biopsy, and, recently, by molecular studies. In Hong Kong, molecular tests of the survivor motor neuron gene was available since 1995. A total of 36 in our cohort of 83 patients had the diagnosis confirmed with molecular analyses. We adopted the classification of SMA from previous studies in which the criteria were based on the International SMA consortium (1992) with modifications according to the 59th European Neuromuscular Center International Workshops. As only SMA patients with childhood onset were studied, we did not include any type IV patients in our study. Parents were interviewed and records were reviewed for demographic and clinical data, including age of onset, gender, family history, motor milestones, disease progression, loss of motor function, and involvement of respiratory or bulbar muscles. We define the age of disease onset as the age in which the first abnormalities were obvious from the medical records or from the descriptions of the parents about the first signs of weakness, eg, age of achievement of certain motor milestones or loss of functions. For the ambulatory status, we define "being ambulatory" as having the ability to walk for 100 meters, either with assistance such as calipers or walkers or without assistance. Actuarial survival curves were obtained by using the Kaplan-Meier method for calculating survival probabilities and probabilities of remaining ambulatory. The parents or the chief caregivers were interviewed for functional status using WeeFIM at the last registered date in September 2002. The WeeFIM consists of 3 domains: 1) self-care, 2) mobility, and 3) cognition. The self-care domain consists of 8 items, namely eating, grooming, bathing, dressing (upper body), dressing (lower body), toileting, and bladder and bowel management. The mobility domain consists of 5 items: transfer from chair or wheelchair, transfer to toilet, transfer to tub or shower, walking/wheelchair/crawling distance, and moving up and down stairs. The cognition domain assesses comprehension, expression, social interaction, problem solving, and memory. A scoring scale from 1 to 7 was used (1 = total assistance, 2 = maximal assistance, 3 = moderate assistance, 4 = minimal contact assistance, 5 = supervision, 6 = modified independence, and 7 = complete independence). The maximum total WeeFIM score is 126, and the maximum score for self-care, mobility, and cognition are 56, 35, and 35, respectively. Results: For type I SMA (n = 22), the survival probabilities at 1, 2, 4, 10, and 20 years were 50%, 40%, 30%, 30%, and 30%, respectively. For type II SMA (n = 26), the survival probabilities at 1, 2, 4, 10, and 20 years were 100%, 100%, 100%, 92%, and 92%, respectively. Sixteen of the SMA I patients and 4 of the SMA II patients died of cardiorespiratory failure. The 5 surviving SMA I patients all were ventilator dependent. All SMA III patients were surviving at the time of study. The probability of remaining ambulatory at 2, 4, 10, and 20 years after onset was 100%, 100%, 81%, and 50% for type IIIa and 100%, 100%, 84%, and 68% for type IIIb (age of onset between 3 and 30 years), respectively. The interval between disease onset and inability to walk was 15.0 +/- 10.9 years (mean +/- standard deviation) and 21.2 +/- 11.7 years for patients with SMA IIIa and IIIb, respectively. Only 39 patients participated in the WeeFIM interview as 20 had already died at the time of study and 24 refused participation. No difference could be found in the age of onset, gender, or types of SMA between those who participated (n = 39) and those who did not (n = 24). The mean total WeeFIM quotients were 24% for SMA type 1, 57% for SMA type 11, 75% for SMA type IIIa, and 78% for SMA type IIIb. For the self-care domain, 100% SMA type I and 73% SMA type II patients required assistance, whereas 55% and 63% of SMA types IIIa and IIIb patients achieved functional independence. Bathing and dressing (upper and lower body) were items with which most SMA children required help or supervision. For the mobility domain, assistance was needed in >90% of SMA types I, II, and IIIa and in 63% of SMA type IIIb patients. Stair management was the major obstacle for independence in achieving mobility for all types of SMA. For the cognition domain, performance was the best among the 3 domains, and 60% of SMA type II, 78% of SMA type IIIa, and 90% of SMA type IIIb patients achieved functional independence. However, except for SMA type IIIb, a significant proportion of patients still need assistance or supervision in the area of problem solving. Statistically significant differences were found in the WeeFIM scores between type I and type II and between type IIIa and IIIb patients. However, no significant difference could be observed between type II and type IIIa SMA patients in the overall WeeFIM scores or performance in any of the 3 domains. Conclusion: We found that there was improvement in survival in SMA patients as compared with other studies. Assistance or supervision was needed for the majority of SMA patients for both mobility and self-care domains. With improvement in survival as a result of medical advances, assessment of the most current or the best-ever functional status at a designated age might be an important criterion for classification of SMA.