

2 or 3.<sup>167</sup> In addition, nutritional modifications are promising (Chapter 3). In the future, gene therapy may be useful to correct for the absence of SMNI and to restore superoxide dismutase type 1 for people with SMA and this defect.

*A pulmonologist who considers the airways but who ignores the respiratory muscles is like a cardiologist who considers the blood vessels but who ignores the heart.*

• Peter Maclem, M.D.

## Nutrition

With Contributions from Irving Haber, M.D., and Jill Gaydos, B.S.

*It is the character of the true philosopher to hope all things not impossible, and believe all things not unreasonable, so is it the character of the physician to hope and believe that the term vis medicatrix naturae (the healing force of nature) does not represent a nonentity; and that there is in the animal body, when in a state of disease, a tendency to return to its healthy state, and that he has means and appliances to assist this curative process.*

• Edward Meryon (1809–1880), the physician who first described, accurately and comprehensively, what came to be known as Duchenne muscular dystrophy<sup>6</sup>

People with neuromuscular diseases (NMDs) have little lean body mass with small protein and mineral reserves and are, therefore, highly susceptible to periods of undernutrition. Malnutrition can exacerbate muscle weakness and decrease lung and immunologic function. Malnutrition and weight loss are independent and significant determinants of morbidity and mortality from respiratory causes.

### Nutritional Requirements

Recommended daily allowances (RDAs) are used to estimate nutrient needs.<sup>168,169</sup> They are designed to provide a margin of safety. Even in the general population, however, RDAs may not be adequate. The elderly often have inadequate gastrointestinal absorption of specific nutrients. Younger patients can also develop malabsorption abnormalities, especially with the presence of gastrointestinal or certain other complicating conditions. In addition, many people eating overly processed foods and few vegetables and fruit may simply not receive enough of many nutrients. It has also been estimated that up to 70% of all children do not receive adequate calcium from their daily diets.

Nutrient requirements of the normal infant and child are determined by taking into consideration rate of growth and physical activity as well as basal energy expenditure. The RDAs for children were estimated using intakes of normally growing infants and the nutrient content of human milk (Table 1). Nitrogen balance studies were used to establish amino acid requirements. The needs for children with NMD or generalized medical conditions can be quite different. The best way to measure adequacy of nutrient intake is to monitor growth.

Fluid needs are determined by the amount of water lost through the skin, lungs, urine,