TABLE 2. Complications of SMA

<table>
<thead>
<tr>
<th>Complication</th>
<th>Characteristics</th>
</tr>
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<tbody>
<tr>
<td>Respiratory</td>
<td>Proportional to general weakness</td>
</tr>
<tr>
<td>insufficiency</td>
<td>May occur during sleep before clinically apparent</td>
</tr>
<tr>
<td></td>
<td>Responds well to noninvasive ventilation</td>
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<tr>
<td>FTT</td>
<td>Particularly infants</td>
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<tr>
<td></td>
<td>Exacerbates weakness, fatigue</td>
</tr>
<tr>
<td>Constipation</td>
<td>Very common</td>
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<tr>
<td></td>
<td>Responds to dietary management</td>
</tr>
<tr>
<td>Orthopedic</td>
<td>Club foot</td>
</tr>
<tr>
<td>deformities</td>
<td>Scoliosis, kyphosis</td>
</tr>
<tr>
<td></td>
<td>Flexion contractures</td>
</tr>
<tr>
<td>Psychosocial</td>
<td>Inadequate intellectual challenge</td>
</tr>
<tr>
<td>dysfunction</td>
<td>Depression rare in patients,</td>
</tr>
<tr>
<td></td>
<td>common in siblings</td>
</tr>
<tr>
<td></td>
<td>Marital discord</td>
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SMA, spinal muscular atrophy; FTT, failure to thrive.

ability. Particularly in infants, but also older children, there may be an exacerbation of weakness and fatigue secondary to negative nitrogen balance. The mechanism for this effect is not well understood. At least some SMA patients suffer from chronic malnutrition that may manifest as easy fatigability and reduced reserve. Some cases of organic aciduria in SMA may have been caused by inadequate nutrition.

A feeding evaluation should be done by a team of occupational and speech therapists and dieticians. Such professionals can adjust the feeding schedule, positioning during feeding, or food textures to maximize caloric intake. If necessary, the child should be examined during a modified barium swallow using several food textures, including liquid, semiliquid, soft, and solid food. If aspiration occurs, a gastrostomy should be recommended. In some cases, supplemental gastrostomy feedings may be indicated in the absence of aspiration because the child cannot take in enough by mouth before fatiguing. Constipation is nearly universal in such patients because of immobility, but it responds easily to increasing fluid and fiber intake.

Scoliosis is the most serious orthopedic problem among patients with SMA (53). Nonwalkers tend to develop spinal deformities earlier than walkers. Most curves are thoracolumbar in location. Spinal orthoses usually do not prevent or retard scoliosis; they may, however, help the patients to sit. Pulmonary function tests (PFTs) should be monitored with the child wearing the orthosis and without it. Frequently, it is necessary to cut a window in the front to allow for movement of the abdominal wall with breathing. If the orthosis limits breathing, its use should be discontinued. The timing of spine surgery is crucial, because one would maximize the child’s growth and so wait until the curve is severe while wanting to do surgery when PFTs are relatively normal. Vigorous preoperative and postoperative physical therapy is required to prevent loss of strength or function after spinal fusion and to prevent respiratory complications. Marked improvement in the degree of scoliosis is often possible after fusion or instrumentation, with associated improvement in vital capacity, sitting, balance, and comfort (54–56). Patients who do not undergo correction of scoliosis will experience progressive deformity with discomfort, inability to position, and further decompensation of PFTs.

Club foot deformity, although not common, may be a presenting complaint for SMA during infancy. It is usually flexible and may not require surgical correction. More common deformities include flexion contractures secondary to immobility. They affect hips, knees, and ankles very quickly. Range of motion exercises are used to prevent such contractures, but they must be done daily and consistently, a feat most patients and families find impossible to accomplish. Splints and braces have been of little help in preventing deformities because of lack of compliance as well.

Walking may occasionally be facilitated by lightweight orthoses for the legs, although for SMA1 or 2 patients, it will likely be a temporary skill (15). Power chairs should be prescribed as close to the second birthday as possible to provide some independent mobility at an appropriate developmental age (57). Because SMA children have high cognitive function (58), they easily learn how to maneuver a joy stick. The motor speed can be adjusted as needed, and the parents should be encouraged to set consistent limits for behavior. As the child grows, pneumatic lifts, special mattresses, and bath accessories will be beneficial for many patients. An in-home occupational therapy consult will ensure that the patient receives appropriate equipment.

FIG. 3. A chest x-ray from a 14-year-old boy with spinal muscular atrophy type 2. The lung volume is markedly decreased, and the rib cage has collapsed into a bell-shape.
School-aged children may benefit from a full-time aid who can help with toileting and feeding and maintain the respiratory and physical therapy regimens during the school day. Parents can be encouraged to seek resources for such help and to communicate freely with school district officials to provide their child with an education that will maximize his or her abilities. A common hurdle for parents is a lack of adequate intellectual challenge for their child. Because of the structure of special education programs, many educators fail to recognize that the SMA child is actually gifted intellectually. It can be helpful to perform psychometric testing at age 4 years to document the level of functioning before entering school.

Families of SMA children suffer from all the common complications seen with chronic disease in childhood: financial stress, marital discord, and depression among siblings. The SMA child is rarely depressed, presumably because there is usually no discernible loss of function after mid-childhood. However, it is advisable to encourage the families to participate in formal counseling and support group activities soon after diagnosis.

REFERENCES

41. Roy N, Mahadevan MS, McLean M, et al. The gene for neuronal apop-