

**NAME:** Sample, Darlene  
**CASE #:** s103  
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**BIRTHDATE:** 11/06/1994  
**LAST REVISED:** 06/19/2007

**SEX:** F  
**BY:** mv

**AGE:** 13

### Questions for Dr. Spinet

The following issues and questions have been developed from research based upon a limited review of selected records, review of video provided by Darlene's family, several phone interviews with Darlene's family, Dr. Walton (Darlene's primary physician/internist), Dr. Fanite (the orthopedist who helped with the tight heel cord and who has consulted on the scoliosis problem), Dr. Aldo Gregson (pediatric spine specialist at Yale University), Dr. John Q. Spinet (pediatric spine specialist at Center for Excellence Medical Center, Los Angeles) and others.

#### Neuromuscular disorder

- Darlene apparently has muscular weakness, muscle atrophy and ligamentous laxity dating from infancy.
- Despite medical investigation, this condition, which may be primarily of connective tissue, muscle, nerve or combined, remains undiagnosed. At one point, an opinion was voiced that this might be the result of poliovirus infection. The family has received conflicting information about this speculation.
- It is not clear whether this problem is progressing.
- Some degree of review of family history apparently did not identify similarly affected cases.
- The relationship between this general condition to Darlene's specific problems of leg spasticity and scoliosis does not seem completely clear.
- Research in connective tissue and neuromuscular disorders progresses steadily. Conditions that may not have been identifiable a few years ago may now be characterized. If a specific diagnosis could be made, it might be possible to have more informed predictions about prognosis, surgical risk, etc.

#### QUESTIONS

- Is it possible to have further genetic, neuromuscular and connective tissue evaluation when Darlene is being treated in Los Angeles?
- Can more clarity be gotten about the theory that polio was involved?

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### Spasticity

- Darlene was treated for a tight heel cord on her right ankle, around age 6. She had a phenol injection, which was followed by further tightening and gait disturbance.
- A fascial release around age 8 resulted in improvement.
- She has used a variety of orthotic devices. Currently, this problem appears to be stable.
- It has not been determined whether this condition is related to a general neuromuscular disorder, or some other cause.

### QUESTIONS

- What additional approaches might be beneficial, in addition to physical therapy and orthopedic monitoring?

### Scoliosis

- Around age 8, Darlene was noticed to be developing thoracic scoliosis. This has progressed to a significant curve, which now impairs respiration and eating. She has had casting therapy, and is now being evaluated for surgery, including possible VEPTR.
- The family apparently has been told that the scoliosis may be at least partly a result of abnormal gait secondary to her heel cord problem. They are also aware that it may be related to an underlying connective tissue disorder.
- Some investigations of respiratory function have been carried out, including pulmonary function testing and a sleep study. There seem to be components of both restrictive and obstructive (reversible) airway disease. It has been difficult to obtain valid pulmonary function testing, and her actual pulmonary status is not clear.
- Darlene may be a candidate for spinal fusion; however the timing is a matter of current discussion. The immediate question revolves around whether a transitional intervention, specifically some form of VEPTR procedure, would improve the outcome of a later fusion procedure (or even make it unnecessary).
- One strategy that has been proposed is to treat Darlene for an interval in traction (HALO, etc.) during which her nutritional and respiratory status might hopefully improve.
- The question has also been raised whether interim treatment, by delaying fusion, might ultimately contribute to a less satisfactory outcome.

### QUESTIONS

- Does VEPTR potentially add benefit? How does the equation of benefit vs. risk work out in Darlene's case?
- Likewise, what are the risks and benefits of simply proceeding directly to fusion? What would be sacrificed by omitting VEPTR? What gained?

- What is the benefit of an interval in HALO traction, and how can it be known how long this should be continued?
- Is there a way to determine Darlene's pulmonary status before intervention? Is there a way to predict it after fusion?
- What is the probability that this scoliosis was caused by abnormal gait induced by the heel cord problem? How likely is it that scoliosis could occur from such events? How likely is the scoliosis to be from underlying neuromuscular disorder? How likely is this to progress in other ways or forms?

### **Nutritional status**

- Before cast therapy, Darlene had an excellent appetite, and was noted to be a voracious eater despite her thin constitution. During the period she was in thoracic casts, she developed difficulty eating. After casting was discontinued, this problem persisted.
- Darlene recently has been evaluated by dietary consultants because of failure to thrive. She was found to have a mild caloric deficit. A recent metabolic survey apparently showed no signs of major nutritional deficits, otherwise. An attempt at nasogastric supplementation was not tolerated because of airway impingement. Currently, Darlene is taking oral supplements.

### **QUESTIONS**

- Is it possible to carry out a nutritional evaluation as part of Darlene's pre-operative assessment in Los Angeles?

### **Surgical risk**

- The family has expressed significant concern about surgical risks, for all the potential procedures under consideration for scoliosis treatment (VEPTR, HALO, fusion).
- Darlene has been noted to have significant muscle atrophy and lack of subcutaneous tissue. This may be partly the result of a caloric deficit, but may also be a constitutional effect of an underlying disorder.
- Because of her muscular weakness, body habitus, respiratory problems and possible underlying disorder, Darlene may be at increased risk during surgery, of whatever sort.
- Dr. Spinet has mentioned that some patients become permanently ventilator dependent after spine surgery.

### **QUESTIONS**

- What are the details of surgical risk for each procedure contemplated?
- Please amplify the details about the possibility of respiratory failure after surgery? What factors affect this risk? Can a prediction be made in Darlene's case? What is this based on?
- Are there interventions (pulmonary rehabilitation, etc.) that can be undertaken before surgery, to improve chances of a good outcome?