

## **SEATING AND MOBILITY IN ESCOBAR SYNDROME**

*SUSAN HERSCOVITCH, OCCUPATIONAL THERAPIST*

**Centre de réadaptation Marie Enfant**

### **Client Background**

Catherine is a delightful 19 year old with a diagnosis of Escobar Syndrome. This rare syndrome is also known as multiple pterygium syndrome or arthrogryposis multiplex congenita. Escobar Syndrome is characterized by webbing causing severe contractures across flexion creases in the extremities including the neck, antecubital, digital, popliteal and intercrural areas. Associated anomalies may include scoliosis, vertical talus and atypical facial features. Intelligence is normal. Catherine resides with her parents and three younger male siblings in a suburb of Montreal in a fully accessible home. The family's van is adapted to accommodate her power chair. Catherine underwent spinal arthrodesis in June 1999 at the age of eleven. Persistent severe post-operative pain resulted in a second surgery to resect the distalmost section of the instrumentation in July 2000. Catherine is an active young woman attending school part-time and working during the summer months as a receptionist at a camp for the disabled.

### **Current Issues**

Catherine has severe muscle weakness and multiple joint contractures of the upper and lower extremities. She is not able to maintain a sitting posture independently and has difficulty remaining in the same position for a prolonged period of time. Fatigue and pain are significant problems. In order to access her joystick and computer, Catherine must be positioned with a zero or anterior tilt. Upper extremity support is critical for her independent mobility. Catherine is extremely sensitive to any variation from her perceived optimal position. The seating and mobility equipment required replacement to accommodate growth, comfort and updating to presently available technology.

### **Goals for Prescription**

Many criteria required consideration in the prescription and the adjustment of Catherine's equipment. The priority was to maintain independent mobility and computer access while improving comfort and increasing sitting tolerance. The objective was for Catherine to be able to participate in educational, leisure and work related activities without pain or discomfort limiting her autonomy.

### **Equipment / Solutions**

Catherine's seating which consisted of a modular backrest with a molded seat cushion in a power chair with power tilt was no longer adequate. A Quantum 1121HDRQ by Pride Mobility with integrated power tilt was chosen. A custom molded seat was made twice because Catherine lost 25 pounds between the measurement and the delivery of the first seat. Special attention was required in determining seat back angle and positioning of the left upper extremity for optimal access to joystick and of the right arm for comfort. Severe flexion and external rotation contractures of the lower extremities called upon some creative reorganizing of the footrest. Accommodations for Catherine's poodle were also considered.

### **Outcomes**

A balance between comfort and function was obtained after several post delivery visits to the technical aids department. Additional modifications to upper extremity support, headrest position and footrest cushion was required. Catherine is satisfied with her new chair and is presently working at a summer camp.

**References**

Escobar Syndrome Is a Prenatal Myasthenia Caused by Disruption of the Acetylcholine Receptor Fetal  $\gamma$  Unit, Hoffmann, K. et al, Am J Hum Genet, 2006 August;79(2): 303-312