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Clinical description of infantile Facioscapulohumeral Muscular Dystrophy (FSHD)

Background
The diagnosis of infantile FSHD has historically been based upon two symptoms: facial weakness before the age of five and shoulder weakness before the age of ten. Patients diagnosed with infantile FSHD often have hearing loss and problems with retinal tissue. There is very little information available on the symptoms, progression of the disease over time, and other characteristics of infantile FSHD.

Results
The National Registry maintains a large sample of clinical and genetic information collected from patients diagnosed with infantile FSHD. The Registry currently contains 53 infantile FSHD patients. The average age at the onset of symptoms of FSHD was 5 years, although the average age at diagnosis was 19.8 years. Muscle pain occurred in 78% of patients and was most frequently experienced in the back/hips and shoulder/arms. The majority of patients reported some degree of muscle weakness in their arms and/or legs. Eight patients used wheelchairs on a regular basis. There were no reports of retinal vascular disorders in this group of patients, however approximately ¼ of the group reported hearing loss and 12% described respiratory problems related to FSHD.

Conclusions
Our report involves one of the largest samples of infantile FSHD. A similar and broad phenotype occurs in infantile FSHD symptoms compared to adult-onset FSHD. A subset of infantile patients may require greater use of wheelchairs by the age of 18 yrs and have greater hearing loss. Additional research is needed to better understand infantile FSHD.