Подборка литературы по Скапулоперонеальному Синдрому тип Кайзер

(Fotopulos and Schulz 1966; Nagatani, Ito et al. 1967; Feigenbaum and Munsat 1970; Thomas, Calne et al. 1972; Steidl and Urbanek 1973; Urbanek, jansa et al. 1974; Kazakov, Bogorodinsky et al. 1975; Schwartz and Swash 1975; Calandre, Garcia Albea et al. 1977; Calandre, Garcia-Albea et al. 1978; Mercelis, Demeester et al. 1980; Chakrabarti and Pearce 1981; Sulaiman, McQuillen et al. 1981; Ronen, Lowry et al. 1986; Ansher, Smith et al. 1989; Tandan, Verma et al. 1989; Milanov, Georgiev et al. 1996; Masuzugawa, Kuzuhara et al. 1997; Milanov and Ishpekova 1997; Uyama 2001; Kazakov 2003; Verma 2005; Walter, Reilich et al. 2007)

Ansher, M., L. D. Smith, et al. (1989). "Autosomal dominant scapuloperoneal syndrome with sensorineural hearing loss." Otolaryngol Head Neck Surg **100**(3): 242-4.

Calandre, L., E. Garcia-Albea, et al. (1978). "[Scapuloperoneal syndrome. Report of a case with myopathic and neuropathic involvement]." Arch Neurobiol (Madr) **41**(1): 49-62.

Calandre, L., E. Garcia Albea, et al. (1977). "[Scapuloperoneal syndrome]." Arch Neurobiol (Madr) **40**(4): 291-306.

Chakrabarti, A. and J. M. Pearce (1981). "Scapuloperoneal syndrome with cardiomyopathy: report of a family with autosomal dominant inheritance and unusual features." J Neurol Neurosurg Psychiatry **44**(12): 1146-52.

 Four members of a family with scapuloperoneal syndrome were examined and investigated. The pattern of inheritance was autosomal dominant and the myopathic basis of the muscle atrophy was established by histological studies of muscle and spinal cord. The family illustrates an unusual combination of features which appear to be distinct from those of other families with myopathic scapuloperoneal syndrome and autosomal dominant inheritance. These include early age of onset and rapid progression in most cases; occurrence of early muscle contractures; and a high incidence of severe cardiomyopathy in three of the four cases. Some of these features resemble those seen in the x-linked form of the disease and the present family appeared to be a new variant of the autosomal dominant form of the scapuloperoneal syndrome.

Feigenbaum, J. A. and T. L. Munsat (1970). "A neuromuscular syndrome of scapuloperoneal distribution." Bull Los Angeles Neurol Soc **35**(2): 47-57.

Fotopulos, D. and H. Schulz (1966). "[Contribution to the pathogenesis of the scapuloperoneal syndrome]." Psychiatr Neurol Med Psychol (Leipz) **18**(4): 129-36.

Kazakov, V. (2003). "What is Davidenkov's scapuloperoneal amyotrophy: is it a myopathic entity or a neurogenic syndrome? What was Davidenkov's opinion concerning this knotty problem?" Neuromuscul Disord **13**(1): 91-2.

Kazakov, V. M., D. K. Bogorodinsky, et al. (1975). "Myogenic scapuloperoneal syndrome - muscular dystrophy in the K. kindred. Reexamination of the K. family described for the first time by Oransky in 1927." Eur Neurol **13**(4): 350-9.

 Additional study of six generations belonging to the K. kindred, which were previously investigated by Oransky in 1927 was carried out. 18 members of this kindred were studied. At the early stages of the disease a sharp dissimilarity of the phenotypes in the K. kindred resulting from the different rate of the development and intensity in the course of the disease was observed, varying from evident scapuloperoneal amyotrophy to the cases resembling Landouzy-Dejerine muscular dystrophy. At the late stages of the disease a considerable clinical simularity of those affected was noted. The clinical and genetic data allowed at the present time to consider the muscular dystrophy in the K. kindred as one of the varieties (namely, as a descending type with a "jump") of the facio-scapulo-limb (or facioscapulohumeral) muscular dystrophy. The scapuloperoneal syndrome could be a long stage in the development of the disorder in some members of the K. kindred.

Masuzugawa, S., S. Kuzuhara, et al. (1997). "Autosomal dominant hyaline body myopathy presenting as scapuloperoneal syndrome: clinical features and muscle pathology." Neurology **48**(1): 253-7.

 Hyaline bodies are rare subsarcolemmal aggregates in type 1 fibers of the skeletal muscle, stain pale pink with hematoxylin-eosin and pale green with the modified Gomori trichrome, and lack reactivity for glycogen and oxidative enzymes. We report clinical findings of autosomal-dominant hyaline body myopathy in seven members in four generations and muscle biopsy findings in two of them. Slowly progressive muscle weakness and atrophy developed with scapuloperoneal distribution; age at onset was from the first to the fifth decade. Muscle biopsy showed subsarcolemmal hyaline bodies in approximately 20% of type 1 fibers. Hyaline bodies showed myofibrillar ATPase activity after acid pre-incubation. Immunohistochemically, they stained intensely with myosin heavy chain (slow), but not with myosin heavy chain (fast). Ultrastructurally, they consisted of granules sometimes in linear array, filaments, and amorphous materials. These findings suggest that hyaline bodies may be products of degeneration of myosin heavy chain (slow).

Mercelis, R., J. Demeester, et al. (1980). "Neurogenic scapuloperoneal syndrome in childhood." J Neurol Neurosurg Psychiatry **43**(10): 888-96.

 Two brothers presented with a slowly progressive scapuloperoneal syndrome starting in early childhood. Initially there were myopathic EMG changes, but these changed to those of denervation. Neuromuscular biopsies at an interval of five years confirmed the neurogenic character of the muscle atrophy.

Milanov, I., D. Georgiev, et al. (1996). "Scapuloperoneal muscular atrophy: Davidenkow's syndrome. Family report." Electromyogr Clin Neurophysiol **36**(5): 259-63.

 The nosology of scapuloperoneal syndrome remains controversial. Is it a variant of Charcot-Marie-Tooth's disease, a form of myopathy, or of spinal muscular atrophy is still unknown. A family with a scapuloperoneal syndrome caused by anterior horn cell involvement is described. Data for sensory involvement were also found. In addition one member of the family had motor nerves and corticospinal tract involvement. The distribution of weakness and muscle wasting was unusual--only lower limbs were involved. However, electromyographic data for both upper and lower limbs involvement were evident. The combination of scapuloperoneal syndrome with sensory loss has been described by Davidenkow, but was not proved by neurographic investigation. Later only one case with proved sensory disturbances was reported. Patients with motor nerves involvement in addition were reported, but not received satisfactory explanation. Probably in some patients the disease could be manifested only with anterior horn cell involvement, while in other both motor and sensory nerves could be involved. In conclusion we suppose that the scapuloperoneal muscular atrophy is a form of spinal muscular atrophy which could be manifested by different symptoms.

Milanov, I. and B. Ishpekova (1997). "Differential diagnosis of scapuloperoneal syndrome." Electromyogr Clin Neurophysiol **37**(2): 73-8.

 The nosology of scapuloperoneal syndrome remains controversial. It may be only a stage in the development of facioscapulohumeral dystrophy. The aim of this investigation was to re-establish the clues for distinguishing between scapuloperoneal syndrome and facioscapulohumeral muscular dystrophy. Twelve patients with a scapuloperoneal syndrome were investigated and compared with seventeen patients with facioscapulohumeral dystrophy. The distribution of weakness for both groups of patients was quite different. The electromyographic examination of patients with scapuloperoneal syndrome revealed typical mixed myogenic and neurogenic features and peroneal nerves involvement. The electromyographic findings in patients with facioscapulohumeral muscular dystrophy were typical for myogenic involvement. In conclusion we can suppose that the scapuloperoneal muscular atrophy is not a stage in the development of facioscapulohumeral dystrophy. It is a separate nosological entity with typical clinical and electromyographic findings.

Nagatani, M., K. Ito, et al. (1967). "[Case of muscular atrophy in scapuloperoneal syndrome]." Naika **20**(7): 1353-7.

Ronen, G. M., N. Lowry, et al. (1986). "Hereditary motor sensory neuropathy type I presenting as scapuloperoneal atrophy (Davidenkow syndrome) electrophysiological and pathological studies." Can J Neurol Sci **13**(3): 264-6.

 A 14 year old boy with scapuloperoneal muscular atrophy, pes cavus, areflexia and distal sensory loss (Davidenkow syndrome) is described. Nerve conduction velocities were diminished. Sural nerve biopsy demonstrated a reduction in the number of myelinated fibers and early "onion-bulb" formation. These observations support the hypothesis that the scapuloperoneal amyotrophy associated with distal sensory loss may represent a variant of type I hereditary motor sensory neuropathy.

Schwartz, M. S. and M. Swash (1975). "Scapuloperoneal atrophy with sensory involvement: Davidenkow's syndrome." J Neurol Neurosurg Psychiatry **38**(11): 1063-7.

 A patient with scapuloperoneal atrophy of neurogenic type, in whome there was also distal sensory impairment, has been studied with conventional EMG, single fibre EMG, and muscle biopsy. This disorder, described by Davidenkow, may be a distinct entity.

Steidl, L. and K. Urbanek (1973). "[The dystrophic type of scapuloperoneal syndrome]." Cesk Neurol **36**(3): 147-50.

Sulaiman, A. R., M. P. McQuillen, et al. (1981). "Scapuloperoneal syndrome. Report on two families with neurogenic muscular atrophy." J Neurol Sci **52**(2-3): 305-25.

 Scapuloperoneal syndrome is a more or less clinically distinct neurologic entity with predominant involvement of scapular and peroneal muscles. The disease shows a variable mode of inheritance. Electromyography and muscle biopsy has shown the presence of denervation and dystrophic changes, sometimes both in the same patient. Cardiac manifestations when present add a graver prognosis to an otherwise relatively benign disease. Study of two cases in this report, one with significant sensory changes and another with cardiopathy, showed degeneration of peripheral nerve and mixed features in muscle biopsy. It is postulated that the myopathic or dystrophic features in the muscle of these cases and other patients with scapuloperoneal syndrome is likely to be secondary to slow denervation and reinnervation.

Tandan, R., A. Verma, et al. (1989). "Adult-onset autosomal recessive neurogenic scapuloperoneal syndrome." J Neurol Sci **94**(1-3): 201-9.

 We report a brother and sister who showed weakness, atrophy and fasciculations starting in adulthood, predominantly affecting the muscles of the shoulder girdles and distal lower extremities. Mild ptosis and facial weakness were present in both. Electrophysiological studies and muscle histology supported a neurogenic basis for the autosomal recessive scapuloperoneal syndrome in this sibship.

Thomas, P. K., D. B. Calne, et al. (1972). "X-linked scapuloperoneal syndrome." J Neurol Neurosurg Psychiatry **35**(2): 208-15.

Urbanek, K., P. jansa, et al. (1974). "The course of a model cutaneous inflammation in the scapuloperoneal syndrome and in systemic neuromuscular diseases." J Neurol **208**(2): 139-46.

Uyama, E. (2001). "[Scapuloperoneal syndrome]." Ryoikibetsu Shokogun Shirizu(36): 462-6.

Verma, A. (2005). "Neuropathic scapuloperoneal syndrome (Davidenkow's syndrome) with chromosome 17p11.2 deletion." Muscle Nerve **32**(5): 668-71.

 The nosologic boundary of neuropathic scapuloperoneal syndrome (Davidenkow's syndrome) remains ill defined and its genetic basis is unknown. A case of Davidenkow's syndrome with the monochromosomic 17p11.2 deletion that often is associated with hereditary neuropathy with liability to pressure palsies (HNPP) is described. The other allele at chromosome 17p11.2 locus was of normal length, and direct sequencing of the coding region of the peripheral nerve protein-22 gene in this allele revealed no additional mutation. The deleted allele in the proband was inherited from the paternal line in which the affected members had a late onset Charcot-Marie-Tooth type 1 clinical phenotype. This observation suggests that the rare Davidenkow's syndrome is clinically related to HNPP and its genotype could be a chromosome 17p11.2 deletion.

Walter, M. C., P. Reilich, et al. (2007). "Scapuloperoneal syndrome type Kaeser and a wide phenotypic spectrum of adult-onset, dominant myopathies are associated with the desmin mutation R350P." Brain **130**(Pt 6): 1485-96.

 In 1965, an adult-onset, autosomal dominant disorder with a peculiar scapuloperoneal distribution of weakness and atrophy was described in a large, multi-generation kindred and named 'scapuloperoneal syndrome type Kaeser' (OMIM #181400). By genetic analysis of the original kindred, we discovered a heterozygous missense mutation of the desmin gene (R350P) cosegregating with the disorder. Moreover, we detected DES R350P in four unrelated German families allowing for genotype-phenotype correlations in a total of 15 patients carrying the same mutation. Large clinical variability was recognized, even within the same family, ranging from scapuloperoneal (n = 2, 12%), limb girdle (n = 10, 60%) and distal phenotypes (n = 3, 18%) with variable cardiac (n = 7, 41%) or respiratory involvement (n = 7, 41%). Facial weakness, dysphagia and gynaecomastia were frequent additional symptoms. Overall and within each family, affected men seemingly bear a higher risk of sudden, cardiac death as compared to affected women. Moreover, histological and immunohistochemical examination of muscle biopsy specimens revealed a wide spectrum of findings ranging from near normal or unspecific pathology to typical, myofibrillar changes with accumulation of desmin. This study reveals that the clinical and pathological variability generally observed in desminopathies may not be attributed to the nature of the DES mutation alone, but may be influenced by additional genetic and epigenetic factors such as gender. In addition, mutations of the desmin gene should be considered early in the diagnostic work-up of any adult-onset, dominant myopathy, even if specific myofibrillar pathology is absent.