

2. Title: A Clinical Study of Hereditary - Familial Spino - Cerebellar Degeneration in a Thai Family

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Objective: This study describes a spectrum of spino-cerebellar degenerations occurring in a Thai family.

Description: Introduction. The hereditary - familial spino - cerebellar degenerations cover a wide - spectrum of syndromes. These include Friedreich's ataxia, Levy - Roussy, Marie's ataxia, oliv - cerebellar and olivo-pontocerebellar degenerations, and the Bassen - Kornzweig syndrome. Friedreich's ataxia is characterized clinically by the appearance in the first or second decades of life of ataxia of the trunk and extremities, intention tremor, absence of deep reflexes, loss of vibration and position sense in the lower extremities, and the presence of an extensor plantar response. Skeletal and cardiac abnormalities are common; blub feet are present in 75% of the cases and scoliosis in 80%. Levy - Roussy is clinically, similar to Friedreich's and includes the appearance of both an ataxia and muscle wasting. Marie's ataxia differs from Friedreich's in the later onset of symptoms, exaggerated stretch reflexes and the frequent occurrence of optic atrophy and oculomotor palsies, and absence of skeletal deformities with no evidence of posterior funicular disease. The olivo - cerebellar and olivo - ponto - cerebella atrophies are characterized by the appearance in adult life of a progressive cerebellar ataxia of extremities and trunk, dysarthria, myastgmus and extra - pyramidal symptoms.

The Bassen - Kornzweig syndrome is an ataxia in which a metabolic disorder has been demonstrated. The syndrome is characterized by acanthocytosis, steatorrhea, retinitis pigmentosa, decrease in total serum lipids and an absolute deficiency of lipoproteins. Formes frustes of all the above syndromes are common in the families of fully affected individuals. A strict differentiation of all the syndromes is rarely possible clinically and only histological examination of the brain and spinal cord will serve to classify them.

Method. All twelve family members were examined and interviewed at Phrabudhabut Hospital. Peripheral visual field mapping and a pneumoencephalogram (done on the propositus), X - rays of thoracic and lumbosacral spines and EKG's (done on four of the affected family members), examination of peripheral blood for red cell acanthocytosis (done on the entire family), determination of total serum lipids (done on seven patients including four affected individuals) were done when possible.

All the above mentioned laboratory examinations were normal. Table 1 lists the pertinent information on all affected members of the family.

Four of the twelve individuals in this family have clinical evidence of spino - cerebellar involvement. The propositus, a 48 year old male and father of the family, clinically appears to have an olivo - ponto - cerebellar degeneration. An 18 year old male, son of the propositus by his first wife has either a Friedreich's ataxia or an olivo - ponto - cerebellar degeneration. Two affected females, aged 12 and 17, who are also children of the propositus but by his second wife have either Friedreich's ataxia or Marie's ataxia.

Summary: A Thai family of twelve has four members who have progressive spino - cerebellar ataxias. Clinically, the four cases range from Friedreich's ataxia to olivo - ponto - cerebellar degeneration. This is the first report in Thailand of an entire family which clinically demonstrates at least three types of the spino - cerebellar ataxias.

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Table 1

Clinical Data on a Thai Family with
Spino-Cerebellar Degeneration
A List of Affected Individuals

| Designation | Age | Sex | Clinical Status | Age at Occurrence of First Symptoms | Cerebellar Signs | Bulbar and Pontine Signs | Stretch Reflexes | Remarks |
|-------------|-----|-----|--|-------------------------------------|------------------|--------------------------|--------------------|--|
| 1 | 48 | M | Olivo-ponto-cerebellar degeneration | 35 | X | X | X | Propositus. Normal pneumo-encephalogram |
| 4 | 18 | M | Friedreich's ataxia or olivo-ponto-cerebellar degeneration | 12? | X | X | | This patient is by the propositus' first wife. |
| 6 | 17 | F | Friedreich's ataxia or Marie's Ataxia | 5 | X | | X (hyperactive) | |
| 7 | 12 | F | Friedreich's ataxia | 5 | X | X (dysarthria) | | |