

Table 3: Clinical Manifestations of Neuroacanthocytosis Syndromes

Syndrome	Onset	Neuromuscular	Neurological	Neuropsychiatric	Additional symptoms
Chorea-acanthocytosis [2]	Mean age: 30	Chorea, parkinsonism, syndromes, oral dystonia, distal myopathy	Seizures (30% of cases), impaired upward gaze or saccades	Progressive behavioral changes resembling frontal lobe syndrome, apathy, depression, bradyphrenia, aggression, impulsivity	Dysarthria (increases the risk of aspiration and nutritional loss) Uncommonly cardiomyopathy
McLeod Syndrome [3]	Mean age: before 40	Sensorimotor neuropathy, neuropenic muscle atrophy, myopathy, absent DTRs	Progressive chorea with head drops, dystonia, gait abnormalities; seizures (40% of cases)	Cognitive deficits (20% of cases), psychiatric changes including personality disorders, anxiety, depression, OCD, bipolar disorder, schizoaffective disorder (30% of cases)	Cognitive or dilated cardiomyopathy, cardiac arrhythmias (e.g. atrial fibrillation); hepatosplenomegaly secondary to hemolysis (30% of cases); increased risk of aspiration and nutritional loss due to dysarthria
Huntington like-disease 2 [4]	Mean age: 41	Chorea affecting gait, speech, swallowing, oculomotor abnormalities, dystonia (late)	Progressive dementia, neuronal loss in striatum and cerebral cortex	Personality changes (e.g. apathy, irritability), depression	Retinal degeneration (2/3 of cases)
PKAN/Hallerorden Spatz Disease [7][8]	Typical form 1 st decade	Typical: extrapyramidal dysfunction (e.g. dystonia, spasticity, choreoathetosis); loss of ambulation (varies by type)	Extrapyramidal symptoms (e.g. dysarthria, dystonia), static dystonias secondary to long bone fractures and osteopenia, early seizures, corticospinal tract involvement, dementia	Typical: Intellectual impairment (severity correlated to age of onset)	Typical: restricted visual fields due to retinopathy
	Atypical form 2 nd , 3 rd decade	Atypical: slower progression with eventual spasticity, hyperreflexia, and limited ambulation w/ "freezing"; tennis syndrome		Atypical: personality changes (e.g. impulsivity, agitation, depression, emotional lability); verbal tic, obsessive-compulsive behavior, and psychotic symptoms	Atypical: multiple speech defects such as palilalia, tachyphasia, tachylogia, and dysarthria Retinopathy (rare)
Abetalipoproteinemia/Hereditary Hypobetalipoproteinemia [7][8]	Presents at birth with failure to thrive, vomiting, and fat malabsorption	Spinocerebellar ataxia and myopathy; progressive loss of DTRs, vibratory sense, and proprioception; muscle weakness, dysarthria	Friedrich's-like ataxia in untreated individuals Evaluate for hyporeflexia, proprioception, loss, and muscle strength		Failure to thrive, severe diarrhea, vomiting, fat malabsorption, loss of night or color vision; atypical retinal pigmentation Acanthocytosis Multiorgan involvement early including cardiomyopathy and hypothyroidism
Acanthoplasmiemia [9][10]	Age 30 to >70	Anxia, chorea, tremors, facial or neck dystonia, blepharospasm, grimacing	Parkinsonism (e.g. rigidity, akinesia)	Cognitive dysfunction (e.g. apathy, forgetfulness)	Iron-restricted microcyte (30%), Retinal degeneration (75%), Diabetes mellitus (70%)

Neuroanthocytosis Syndromes

Zhe Xu



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