

AP-4-associated HSP affects males and females of ethnic groups from around the world. The prevalence of AP-4-HSP is unknown. AP-4-HSP is likely under-recognized since the symptoms (phenotypic spectrum) largely overlap with that of cerebral palsy and, in the absence of genetic testing, patients may be misdiagnosed as having cerebral palsy. Many of the initial clinical manifestations of AP-4-associated HSP are nonspecific and may resemble other disorders characterized by spasticity, developmental delay / intellectual disability, and seizures. Patients may be misdiagnosed as having cerebral palsy. The diagnosis of AP-4-associated HSP is based on clinical characteristics and testing that may include a brain MRI showing characteristic features such as a thin corpus callosum, wide lateral ventricles and changes in the white matter. A definitive diagnosis is reached by genetic testing.