Pontocerebellar hypoplasias (PCH) are a group of rare heterogeneous conditions characterized by prenatal development of an abnormally small cerebellum and brain stem, which is usually associated with profound psychomotor retardation. Although the clinical features vary widely, pontocerebellar hypoplasias are usually associated with profound intellectual disability and delayed or absent psychomotor milestones. In most cases, the disease is uniformly fatal early in life. Life span has ranged from death in the perinatal period to about 20-25 years of age. Only a few individuals-usually patients with PCH type 2-have survived to the second and third decades of life. At least 6 types of PCH have been described and a few rare variants are now being identified. The disease affects both males and females without a predilection for either sex. More than 100 cases have been reported in the medical literature. The exact incidence of pontocerebellar hypoplasia is unknown.