Rosenberg-Chutorian syndrome is an extremely rare genetic disorder characterized by the triad of hearing loss, degeneration of the optic nerve (optic atrophy) and neurological abnormalities, specifically disease of the nerves outside of the central nervous system (peripheral neuropathy). The arms and legs are most often affected by peripheral neuropathy. Rosenberg-Chutorian syndrome is inherited as an X-linked disorder with occasional mild symptoms present in the female carrier. A diagnosis of Rosenberg-Chutorian syndrome is made based upon a thorough clinical evaluation, a detailed patient history and identification of characteristic findings. Molecular genetic testing for PRSP1 gene mutations is available to confirm the diagnosis. Carrier testing and prenatal diagnosis are available if a PRSP1 gene mutation has been identified in an affected family member.