AP1S2 is a gene located on the X chromosome that encodes a protein involved in the assembly of adaptor protein complex 1 (AP-1). This complex is crucial for the trafficking of proteins between the trans-Golgi network and endosomes. Specifically, the AP1S2 gene encodes the sigma 2 subunit of the AP-1 complex.

Mutations in the AP1S2 gene are associated with a rare genetic disorder known as X-linked intellectual disability (XLID). This condition can sometimes present with other neurological features, including congenital hydrocephalus. The exact mechanism by which mutations in AP1S2 lead to hydrocephalus is not fully understood, but it is likely related to the gene's role in cellular trafficking and the development of the nervous system.

In the context of congenital hydrocephalus, AP1S2 mutations disrupt normal cellular functions that are critical during brain development, possibly leading to abnormalities in CSF flow or brain structure that result in hydrocephalus.