Phenotypes Associated with Recurrent Pathogenic Variants in STXBP1

Some STXBP1 pathogenic missense variants occurring in more than one affected individual were listed below, comparing phenotypes:

- **c.1217G>A** was found in eight unrelated individuals. All had severe to profound global developmental delay or intellectual disability (ID). Seizures required two or more anti-epileptic drugs (AEDs) or were drug resistant [Saitsu et al 2010, Mignot et al 2011, Allen et al 2013, Di Meglio et al 2015, Romaniello et al 2015, Stamberger et al 2016].

- **c.875G>A** was found in five unrelated individuals [Michaud et al 2014, Helbig et al 2016, Stamberger et al 2016, Trump et al 2016]. Age at seizure onset varied from 6 months to 8 years. Seizure types included infantile spasms and partial seizures. ID ranged from mild to severe. Two individuals had a good response to AEDs becoming seizure free [Stamberger et al 2016].

- **c.703C>T** was found in three unrelated individuals. All had severe to profound global developmental delay or ID. Two became seizure free [Saitsu et al 2010, Allen et al 2013, Boutry-Kryza et al 2015].

- **c.364C>T** was found in four unrelated individuals [Lemke et al 2012, Mercimek-Mahmutoglu et al 2015, Stamberger et al 2016]. Age of seizure onset varied between the neonatal period and age one year. One individual had no seizure history.

- **c.1439C>T** was found in three unrelated individuals. All had severe developmental delay. Two became seizure free and one was refractory to anti-seizure treatment [Milh et al 2011, Tso et al 2014, Di Meglio et al 2015].

- **c.703C>T** was found in three unrelated individuals. All had severe to profound global developmental delay or ID. Two became seizure free [Saitsu et al 2010, Allen et al 2013, Boutry-Kryza et al 2015].

References


