PMS was initially described in the medical literature in 1985. Since that time, additional cases have been reported in the literature, with more than 1500 members in the Phelan-McDermid Syndrome Foundation membership. Males and females are equally likely to be affected. Based on limited statistical analysis, the occurrence rate has been estimated to fall in the range of 2.5-10 per million births, although this is likely to be a gross underestimate. Due to the subtle appearance of the deletion of chromosome 22 and the relatively mild physical features of affected individuals, diagnosis of PMS is often difficult. Over 30% of individuals with this deletion have required two or more chromosome studies before the deletion is detected. It is likely that there are many individuals who had “normal” chromosome studies at an earlier age but who actually carry this subtle chromosome abnormality.