Abstract citation ID: bvac150.964

Genetics & Development
RF17 | PMON27

Genetic Subtype Differences in Relation to Health Problems Among Adults With Prader-Willi Syndrome
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Background: Prader-Willi syndrome (PWS) is a complex rare genetic disorder associated with hypothalamic dysfunction, pituitary hormone deficiencies, hyperphagia and (morbid) obesity. PWS is caused by loss of expression of paternally expressed genes on chromosome 15q11.2-q13. The most common genetic mechanisms leading to PWS are paternal deletion (DEL) and maternal uniparental disomy (mUPD). DELs can be subdivided in type 1 and (smaller) type 2 deletions (DEL-1, DEL-2). Most research has focused on behavioral, cognitive and psychological differences between patients with a DEL-1, DEL-2 or mUPD. However, little is known about the genetic subtype differences in relation to physical health problems.

Methods: We reviewed the medical files of all adults with genetically confirmed PWS who visited the outpatient...
Results: Twenty-eight adults had an mUPD and 65 a DEL (13 DEL-1, 27 DEL-2, 25 unspecified). Gender, age and BMI did not differ between the genetic subgroups. Although psychiatric problems (psychotic episodes) were significantly more often present in adults with an mUPD ($P < 0.001$) and scoliosis was more prevalent among patients with DEL ($P = 0.04$), there was only a slight difference in prevalence of other medical problems like hypertension, cold intolerance, edema, hyperphagia, skin picking, abdominal pain and fatigue. There were no significant differences between DEL-1 and DEL-2. However, fatigue, cold intolerance, edema and hyperphagia were slightly more prevalent among adults with a DEL-1, whereas osteopenia, constipation and skin picking were more prevalent among adults with a DEL-2.

Conclusion: The differences in health problems between PWS adults with DEL-1, DEL-2 and mUPD are mostly present in the psychological domain. Especially psychotic episodes were more frequent in adults with an mUPD. Apart from scoliosis, there were no significant differences in physical health outcomes between the genetic subtypes.

Presentation: Sunday, June 12, 2022 1:00 p.m. - 1:05 p.m., Monday, June 13, 2022 12:30 p.m. - 2:30 p.m.