Abstract

The Prader-Willi syndrome (PWS) and Angelman syndrome (AS) are neurobehavioural syndromes caused by loss of paternally or maternally expressed imprinted genes chromosom region 15q11-q13. Differential *gene expression* leads to differential phenotype. AS is caused by a lack of expression of gene *UBE3A.* The PWS is caused by changes in multiple genes, mainly in *NDN, MAGEL2, SNURF-SNRPN* a *SNORD116.* Both syndromes have unique morphological, physiological and behavioral manifestations. The most consistent features of PWS are neonatal hypotonia, hyperphagia, obesity and short stature. Typical features of AS are ataxia and epileptic seizure. Mental retardations are characteristic for both syndromes. Current knowledge of relationship between gene mutations and phenotypes in PWS and AS, their genetic background, diagnosis and treatment of these serious diseases are summarized in this Bachelor thesis.