Angelman Syndrome: the emerging genotype/phenotype

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The purpose of this presentation is to review the characteristics of Angelman syndrome in relation to the growing knowledge about its differing genetic profiles. The variation in the behavioral phenotype will be outlined and, in particular, the communication phenotype will be discussed following the findings of a current study by the author.

There are four major genetic groups which have been recognized in this syndrome: maternal deletions of chromosome 15q11-13, paternal uniparental disomy, imprinting defects, and single point mutations (UBE3A). The aim of the communication study was to establish the difference in communication study was to extablish the difference in communication phenotype, which parallel a difference in genetic pattern.

Seven pairs of young people with a classic deletion were matched for age with those who had either an imprinting defect or uniparental disomy. A subsidiary aim was to determine whether children with more severe epilepsy have lower levels of communication skill. Assessments and questionnaires were used to establish developmental ability in a number of domains but most particularly in verbal comprehension, modes of expressive communication, pragmatic use of language, and eating and drinking skills. The results between the two groups were significant and will be discussed in detail. This study has advanced the knowledge of the differing phenotypes in the genetic groups and contributes to a clearer understanding of the long-term outcome of young people with the syndrome.

Understanding the prognosis for speech and communication will have implications for teaching and therapeutic management. These will be discussed. A videotape will be used to illustrate the key points of the presentation.