Journal of Down Syndrome & Chromosome Abnormalities

OPEN ACCESS Freely available online

Opinion Article

Genetic Abnormalities Caused by Prader–Willi Syndrome

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DESCRIPTION

Prader-Willi syndrome (PWS) is a genetic disorder with a range of manifestations that impacts many different body systems which are hypotonia with poor weight gain in infancy, mild mental retardation, hypogonadism, a lack of growth hormone that results in short stature in the family, early childhood onset hyperphagia and an obesity recognisable appearance and behaviour which occurs in psychiatric disturbance among the numerous other minor characteristics. PWS is an illustration of a hereditary condition involving genomic imprinting paternal micro deletion, maternal uniparental disomy and imprinting deficit are the three primary potential causes. The lack of expression of genes passed down from parents in the 15q11.2-q13 region can be caused by any one of these reasons possesses an identifiable pattern of substantial neurologic, cognitive, endocrine, behavioural, and psychiatric abnormalities as well as dysmorphic traits. It was a usually sporadic condition that is rather prevalent. PWS was the first human disease connected to genomic imprinting that was demonstrated to be caused by uniparental disomy. It results from the failure of the paternally inherited genes to express themselves in the PWS region of chromosome 15. Major characteristics include hypogonadism (small external genitalia and pubertal insufficiency), hyperphagia, which if left unchecked can result in morbid obesity, infantile lethargy and hypotonia, which cause poor feeding and failure to thrive, developmental and intellectual disability, short stature, distinctive facial features and body habitus, and a typical behavioural phenotype that includes temper tantrums and compulsive traits. Key elements of symptomatic and supportive care include behaviour control, special education, sheltered employment, hormone replacement therapy, and special diets.

Hypotonia

Infantile hypotonia, which results in decreased movement, lowerspontaneous arousal, feeble cries etc. When carried out for diagnostic purposes, neuromuscular examinations, including muscle biopsy, are often normal or show hazy signs of disuse. The source of the hypotonia is central. In new-borns with central hypotonia, a muscle biopsy is typically not recommended unless PWS has been genetically ruled out. Early infancy fails as a result of laziness and necessitating gavage feeding from weeks to months. There has been a time when the child's feeding habits have been fairly average by the time the child is drinking from a cup or eating solids occurs though even adults still experience modest hypotonia and have decreased muscle mass and tone, the hypotonia improves over time. Other epigenetic markers at the PWS-ICR, which are also distinguished by allele-specific DNA methylation, are differentiated by parental origin. When the transcription factor ZNF274 works with the histone H3 lysine 9 H3K9 methyl transferases SETDB1 to bind to sites within the 5 cluster of SNORD116 repeats, maternal-specific H3K9 marks are specifically deposited. When both SETDB1 and ZNF274 were down regulated or inhibited, the normally silent maternal allele to produce a small amount of the SNORD116 mRNA.

Diagnosis

The mainstay of diagnosis is a DNA methylation test to identify abnormal parent-specific imprinting within the Prader-Willi Critical Region (PWCR) on chromosome 15. This test determines whether the region is solely inherited from the mother (i.e., the region contributed by the father is absent) and detects more than 99% of affected people. SNORD116 repeats. The normally quiet maternal allele was able to express a low level of the SNORD116 transcript when both SETDB1 and ZNF274 were knocked down or inhibited.

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Received: 02-Sep -2022, Manuscript No. JDSCA-22-19500; **Editor assigned:** 06- Sep -2022, Pre QC No. JDSCA-22-19500 (PQ); **Reviewed:** 20- Sep -2022, QC No. JDSCA-22-19500; **Revised:** 27-Aug-2022, Manuscript No. JDSCA-22-19500 (R); **Published:** 04-Oct 2022, DOI: 10.35248/2472-1115.22.08.205.

Citation: Hopkins A (2022) Genetic Abnormalities Caused by Prader–Willi Syndrome.J Down Syndr Chr Abnorm. 8:205.