Extended Abstract Journal of Congenital Disorder 2020 Vol.2 Issue.2

# Peters Plus Syndrome: Another Way to See a Known Syndrome

Elisabetta Grande
University of Siena, viale Bracci, Italy.

**Abstract**

Peters Plus Syndrome is a rare autosomic recessive disorder, clinically characterized by abnormal formation of various structures including anterior eye chamber, genitourinary tract, skeletal system and central nervous system. PPS is due to defective B3GALTL gene encoding for a glycosyl-transferase that plays a crucial role during embryogenesis. Here we report on a 12-year old boy affected by Peters Plus syndrome who showed peculiar additional features such as absence epilepsy and recurrent bacterial infections.

Peters plus syndrome (PPS) is a rare autosomal recessive disorder mainly characterized by anterior chamber eye abnormalities (Peter's anomaly) and rhizomelic limb shortening. Genitourinary tract and central nervous system (CNS) anomalies are also commonly reported. PPS is due to defective B3GALTL gene encoding a glycosyltransferase which plays a crucial role in embryogenesis. Here we report a boy affected by PPS associated with peculiar additional features such as absence seizures and recurrent bacterial meningitis. Clinical History First son of a primigravid woman, he was born at 40 weeks. Intrauterine growth restriction was observed during pregnancy. At birth, apgar indices were 8I /10V . Weight, length and head circumference were respectively at the 10th-25th centile, below the 3rd centile and at 25th-50th centile. Physical examination showed dysmorphic features and bilateral corneal leukoma. Genetic and metabolic testing, including karyotyping, urinary organic acids and mucopolisaccharides, were normal.

Abdominal ultrasound (US) was normal except for the presence of testes in inguinal canals. Brain US showed a mild enlargement of lateral ventricles with calcifications of small thalamic arteries. Brain MRI imaging revealed widening of the 4th ventricle, enlarged cisterna magna, vermian hypoplasia, enlarged and dysmorphic lateral ventricles, increased signal intensity in the periventricular white matter and small bilateral areas of abnormal hyperintense signal at the level of the corona radiata. Ophthalmological evaluation showed large bilateral central leukoma associated with residuals of hyaloid artery, colobomatous appearance of right papilla and increased eye pressures. In infancy, peculiar somatic traits became more evident. The child showed a round face, short upslanting palpebral fissures, thick eyebrows, small and low set ears, micrognathia, maxillary hypoplasia (Figure 1), incomplete cleft lip, scoliosis, rhizomelic shortnening of the arms and brachydactyly. Motor developmental milestones were reached timely while a mild language delay was observed. On the basis of the physical appearance a diagnosis of Peters Plus Syndrome was formulated, subsequently confirmed by genetic analysis which detected a homozygous mutation c660+1G>A in B3GALTL gene. Both parents were found to be heterozygote for that mutation.

Peters Plus Syndrome: Another Way to See a Known Syndrome Elisabetta Grande1 *, Serena Ciabattoni2 , Elena Andreucci3 , Chiara Romano1 , Gianluca Capecchi1 , Silvia Ferranti1 and Salvatore Grosso1 1University of Siena, viale Bracci, Italy 2Medical Genetics Unit, Department of Clinical and Experimental Biomedical Sciences, "Mario Serio" University of Florence, Florence, Italy 3Medical Genetics Unit, Meyer Children's University Hospital, viale Gaetano Pieraccini, Florence, Italy Abstract Peters Plus Syndrome is a rare autosomic recessive disorder, clinically characterized by abnormal formation of various structures including anterior eye chamber, genitourinary tract, skeletal system and central nervous system. PPS is due to defective B3GALTL gene encoding for a glycosyl-transferase that plays a crucial role during embryogenesis. Here we report

on a 12-year old boy affected by Peters Plus syndrome who showed peculiar additional features such as absence epilepsy and recurrent bacterial infections. During childhood the patient developed three major infective episodes of CNS complicated by hearing loss with subsequent cochlear device implantation. The first CNS infection episode occurred at the age of 4 years, when he presented pneumococcal meningitis. Immunological tests were normal. In that occasion an electroencephalogram (EEG) was performed which was proved normal. Three years later, he developed a second meningitis.

**References**

1. Lesnik Oberstein SA, Kriek M, White SJ, Kalf ME, Szuhai K, et al. (2006) Peters Plus syndrome is caused by mutations in B3GALTL, a putative glycosyltransferase. Am J Hum Genet 79: 562-566.

2. Sato T, Sato M, Kiyohara K, Sogabe M, Shikanai T, et al. (2006) Molecular cloning and characterization of a novel human beta1,3-glucosyltransferase, which is localized at the endoplasmic reticulum and glucosylates O-linked fucosylglycan on thrombospondin type 1 repeat domain. Glycobiology 16: 1194-1206.

3. Lesnik Oberstein SAJ, van Belzen M, Hennekam R (2007) Peters Plus Syndrome. Gene Reviews, Seattle (WA), University of Washington, Seattle.

4. Heinonen TY, Maki M (2009) Peters'-plus syndrome is a congenital disorder of glycosylation caused by a defect in the beta1, 3-glucosyltransferase that modifies thrombospondin type 1 repeats. Ann Med 41: 2-10.

grande.elisabetta@yahoo.it