Individual publication results for: Pallister-Killian Syndrome
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2014

Interphase fluorescence in situ hybridization characterization of mosaicism using uncultured amniocytes and cultured stimulated cord blood lymphocytes in prenatally detected Pallister-Killian syndrome.

Pallister-Killian syndrome.

12p microRNA expression in fibroblast cell lines from probands with Pallister-Killian syndrome.

Genome-wide expression analysis in fibroblast cell lines from probands with Pallister Killian syndrome.

Prenatal diagnosis of Pallister Killian Syndrome in a fetus with congenital diaphragmatic hernia, short limbs, and increased nuchal translucency.

Pallister-Killian syndrome in a preterm newborn who died soon after precipitous delivery: cytogenetic analysis.

The genetics of common disorders - congenital diaphragmatic hernia.

Pallister-Killian syndrome.

Cardiac manifestations of Pallister-Killian syndrome.

[Three cases of Pallister-Killian syndrome].
2013

Pallister-Killian syndrome: a common yet under-recognised cause of epileptic spasms.
Kapoor S.

Central auditory development in children with hearing loss: clinical relevance of the P1 CAEP biomarker in hearing-impaired children with multiple disabilities.
Sharma A, Glick H, Campbell J, Biever A.
Hearing Balance Commun. 2013 Sep;11(3).

Persistent mosaicism for 12p duplication/triplication chromosome structural abnormality in peripheral blood.
Shackelford AL, Conlin LK, Hummel M, Spinner NB, Wenger SL.

Response to Cobben et al. "Array CGH on unstimulated blood does not detect all cases of Pallister-Killian syndrome: buccal smear analysis should remain the diagnostic procedure of first choice".
Hodge JC, Kirmani S.

Array CGH on unstimulated blood does not detect all cases of Pallister-Killian syndrome: buccal smear analysis should remain the diagnostic procedure of first choice.
Cobben JM, Engelen M, Polstra A.

Supernumerary ring chromosome: an etiology for Pallister-Killian syndrome?
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Utility of SNP arrays in detecting, quantifying, and determining meiotic origin of tetrasomy 12p in blood from individuals with Pallister-Killian syndrome.

Novel clinical manifestations in Pallister-Killian syndrome: comprehensive evaluation of 59 affected individuals and review of previously reported cases.

Developmental and behavioral characteristics of individuals with Pallister-Killian syndrome.
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Pallister-Killian syndrome: historical perspective and foreword.
Pallister P.
Seizure characteristics in Pallister-Killian syndrome.
Candee MS, Carey JC, Krantz ID, Filloux FM.

Duplication 12p and Pallister-Killian syndrome: a case report and review of the literature toward defining a Pallister-Killian syndrome minimal critical region.

Seizures and EEG patterns in Pallister-Killian syndrome: 13 new Italian patients.

Gonadoblastoma: Case report of two young patients with isochromosome 12p found in the dysgerminoma overgrowth component in one case.
Changchien YC, Haltrich I, Micsik T, Kiss E, Fónyad L, Papp G, Sápi Z.

An Indian boy with additional features in Pallister-Killian syndrome.
Shah K, George R, Balla ES, Oommen SP, Padankatti CS, Srivastava VM, Danda S.
Indian J Pediatr. 2012 Sep;79(9):1238-40

Discordant phenotype in monozygotic twins with mosaic trisomy 12p in lymphocytes.

Ultrasound presentation of Pallister-Killian syndrome with a prominent sacral appendage.
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Mosaic Intrachromosomal Triplication of (12)(p11.2p13) in a Patient with Pallister-Killian Syndrome.
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2011


2010

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Liehr T, Karanysheva T, Merkas M, Brecevic L, Hamid AB, Ewers E, Mrasek K, Kosyakova N, Weise A. 

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Abnormally flat facial profile on two- and three-dimensional ultrasound and array comparative genomic hybridization for the diagnosis of Pallister-Killian syndrome. 
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Tetrasomy 12p (Pallister-Killian syndrome): difficulties in prenatal diagnosis.
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Prenatal diagnosis of Pallister-Killian syndrome in young woman: ultrasound indicators and confirmation by FISH.
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Pallister-Killian Syndrome (PKS) as a Cause of Mental Retardation.

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Anesthesia for orthopedic surgery in Pallister-Killian syndrome.
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Pallister-Killian syndrome: a report of 2 cases and review of its surgical aspects.
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High cognitive functioning and behavioral phenotype in Pallister-Killian syndrome.
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Pallister-Killian syndrome: report of one case.
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Brain MRI findings of older patients with Pallister-Killian syndrome.
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Small supernumerary marker chromosomes (sSMC) in humans.
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Mild phenotype in a 15-year-old boy with Pallister-Killian syndrome.
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Parental origin of the isochromosome 12p in Pallister-Killian syndrome: molecular analysis of one patient and review of the reported cases.  
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DEVELOPMENT AND EDUCATION IN CHILDHOOD
The constructive theory on subject-object activity in human development.
Part 1
A case study of a profoundly handicapped infant
Shinsuke TANAKA
ISBN 4-9900355-2-6

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