

# Individual publication results for: **Pallister-Killian Syndrome**

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(updated: 10 January 2015)

2014

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

Chen CP, Peng CR, Chern SR, Kuo YL, Wu PS, Town DD, Pan CW, Yang CW, Wang W.  
Taiwan J Obstet Gynecol. 2014 Dec;53(4):566-71.

Pallister-Killian syndrome.

Izumi K, Krantz ID.  
Am J Med Genet C Semin Med Genet. 2014 Dec;166C(4):406-13.

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

Izumi K, Zhang Z, Kaur M, Krantz ID.  
Chromosome Res. 2014 Dec;22(4):453-61.

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

Kaur M, Izumi K, Wilkens AB, Chatfield KC, Spinner NB, Conlin LK, Zhang Z, Krantz ID.  
PLoS One. 2014 Oct 16;9(10):e108853.

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

Ozlu T, Ocak Z, Ozyurt O.  
Taiwan J Obstet Gynecol. 2014 Sep;53(3):404-5.

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

Moro MA, Sanna R, Cambosu F, Soro G, Dessole S, Montella A, Capobianco G.  
Clin Exp Obstet Gynecol. 2014;41(4):483-5.

The genetics of common disorders - congenital diaphragmatic hernia.

Slavotinek AM.  
Eur J Med Genet. 2014 Aug;57(8):418-23.

Pallister-Killian syndrome.

Srinivasan A, Wright D.  
Am J Case Rep. 2014 May 7;15:194-8.

Cardiac manifestations of Pallister-Killian syndrome.

Tilton RK, Wilkens A, Krantz ID, Izumi K.  
Am J Med Genet A. 2014 Feb 6.

[Three cases of Pallister-Killian syndrome].

Toledo-Bravo de Laguna L, del Campo-Casanellas M, Santana-Rodriguez A, Santana-Artiles A, Sebastian-Garcia I, Cabrera-Lopez JC.  
Rev Neurol. 2014 Jan 16;58(2):63-8. Spanish.

## 2013

Pallister-Killian syndrome: a common yet under-recognised cause of epileptic spasms.  
Kapoor S.  
Epileptic Disord. 2013 Dec;15(4):466-7.

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.  
Case Rep Genet. 2013;2013:857926. 2013 Sep 15.

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.  
Am J Med Genet A. 2013 Jun;161A(6):1520-1.

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.  
Am J Med Genet A. 2013 Jun;161A(6):1517-9.

Supernumerary ring chromosome: an etiology for Pallister-Killian syndrome?  
Lloveras E, Canellas A, Cirigliano V, Català V, Cerdan C, Plaja A.  
Fetal Diagn Ther. 2013;34(3):172-5.

## 2012

Utility of SNP arrays in detecting, quantifying, and determining meiotic origin of tetrasomy 12p in blood from individuals with Pallister-Killian syndrome.  
Conlin LK, Kaur M, Izumi K, Campbell L, Wilkens A, Clark D, Deardorff MA, Zackai EH, Pallister P, Hakonarson H, Spinner NB, Krantz ID.  
Am J Med Genet A. 2012 Dec;158A(12):3046-53.

Novel clinical manifestations in Pallister-Killian syndrome: comprehensive evaluation of 59 affected individuals and review of previously reported cases.  
Wilkens A, Liu H, Park K, Campbell LB, Jackson M, Kostanecka A, Pipan M, Izumi K, Pallister P, Krantz ID.  
Am J Med Genet A. 2012 Dec;158A(12):3002-17.

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Kostanecka A, Close LB, Izumi K, Krantz ID, Pipan M.  
Am J Med Genet A. 2012 Dec;158A(12):3018-25.

Pallister-Killian syndrome: historical perspective and foreword.  
Pallister P.  
Am J Med Genet A. 2012 Dec;158A(12):2999-3001.

Seizure characteristics in Pallister-Killian syndrome.

Candee MS, Carey JC, Krantz ID, Filloux FM.  
Am J Med Genet A. 2012 Dec;158A(12):3026-32.

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

Izumi K, Conlin LK, Berrodin D, Fincher C, Wilkens A, Haldeman-Englert C, Saitta SC, Zackai EH, Spinner NB, Krantz ID.  
Am J Med Genet A. 2012 Dec;158A(12):3033-45.

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

Giordano L, Viri M, Borgatti R, Lodi M, Accorsi P, Faravelli F, Ferretti MC, Grasso R, Memo L, Prola S, Pruna D, Santucci M, Savasta S, Verrotti A, Romeo A.  
Eur J Paediatr Neurol. 2012 Nov;16(6):636-41.

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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.

Pauli S, Schmidt T, Funke R, Zoll B, Burfeind P, Dybowski U, Shoukier M, Bartels I.  
Eur J Med Genet. 2012 Aug-Sep;55(8-9):480-4.

Ultrasound presentation of Pallister-Killian syndrome with a prominent sacral appendage.

Johnstone ED, Jones EA.  
Ultrasound Obstet Gynecol. 2012 Aug;40(2):239-41.

Mosaic Intrachromosomal Triplication of (12)(p11.2p13) in a Patient with Pallister-Killian Syndrome.

Yakut S, Mihci E, Altioek Clark O, Cetin Z, Keser I, Berker S, Luleci G.  
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Occurrence and clinical features of epileptic and non-epileptic paroxysmal events in five children with Pallister-Killian syndrome.  
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Hodge JC, Hulshizer RL, Seger P, St Antoine A, Bair J, Kirmani S.  
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Clinical and radiological findings in Pallister-Killian syndrome.  
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Congenital diaphragmatic hernia.  
Tovar JA.  
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Anesthetic management of Pallister-Killian syndrome using a Bispectral Index monitor in a patient with severe seizures.  
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Amniocentesis can be useful during the third trimester of pregnancy for antenatal diagnosis of Pallister-Killian syndrome: a case report.  
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Pallister-Killian syndrome: meiosis II non-disjunction may be the first step in the formation of isochromosome 12p.  
Shen JD, Liang DS, Zhou ZM, Xia Y, Long ZG, Wu LQ.  
Chin Med J (Engl). 2010 Dec;123(23):3482-5. No abstract available.

First trimester diagnosis of Pallister-Killian Syndrome in a fetus with suggestive abnormalities.  
Mourali M, El Fekih C, Dimassi K, Fatnassi A, Ben Zineb N, Oueslati B.  
Tunis Med. 2010 Sep;88(9):666-9.

Somatic mosaicism in cases with small supernumerary marker chromosomes.  
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Curr Genomics. 2010 Sep;11(6):432-9.

Diagnosis of Pallister-Killian syndrome by array comparative genome hybridization from a spleen sample.  
Harrison V, Williams R, Connell L, Kini U.  
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Taiwan J Obstet Gynecol. 2010 Jun;49(2):235-8.

Contribution of 3D ultrasound and fetal face studies to the prenatal diagnosis of Pallister-Killian syndrome.  
Sananes N, Guigue V, Vayssiere C, Kohler M, Girard-Lemaire F, Flori E, Carelle-Calmels N, Boehm N, Samama B, Doray B, Favre R.  
J Matern Fetal Neonatal Med. 2010 Jun;23(6):558-62

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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Pallister Killian syndrome: unusual significant postnatal overgrowth in a girl with otherwise typical presentation.  
Frkovi SH, Durisevi IT, Trci RL, Sarnavka V, Gornik KC, Muzini D, Letica L, Bari I, Begovi D.  
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Phenotypic overlapping of trisomy 12p and Pallister-Killian syndrome.  
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Ticho BH.  
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Late-Onset Epileptic Spasms in Children With Pallister-Killian Syndrome: A Report of Two New Cases and Review of the Electroclinical Aspects.

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Prenatal diagnosis of Pallister-Killian syndrome associated with pulmonary stenosis and right ventricular dilatation.

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Genetic considerations in the prenatal diagnosis of overgrowth syndromes.

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Prenat Diagn. 2009 Jul 16;29(10):923-929.

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Mihci E, Velagaleti GV, Ensenaer R, Babovic-Vuksanovic D.  
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Theisen A, Rosenfeld JA, Farrell SA, Harris CJ, Wetzel HH, Torchia BA, Bejjani BA, Ballif BC, Shaffer LG.  
Am J Med Genet A. 2009 May;149A(5):914-8.

Tetrasomy 12p (Pallister-Killian syndrome): difficulties in prenatal diagnosis.

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Pallister-Killian syndrome in a girl with mild developmental delay and mosaicism for hexasomy 12p.

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Prenatal diagnosis of Pallister-Killian syndrome in young woman: ultrasound indicators and confirmation by FISH.

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The Pallister-Killian syndrome in a child with rare karyotype--a diagnostic problem.  
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Anesthesia for orthopedic surgery in Pallister-Killian syndrome.  
J Knab, EW Heupel, D Steinmann  
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Valproate-associated reversible encephalopathy in a 3-year-old girl with Pallister-Killian syndrome.  
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M Liberati, K Melchiorre, I D'Emilio, PE Guanciali-Franchi, I Iezzi, S Rotmensch, C Celentano  
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Mosaic tetrasomy 12p with triplication of 12p detected by array-based comparative genomic hybridization of peripheral blood DNA.  
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Pallister-Killian syndrome: tetrasomy of 12pter-->12p11.22 in a boy with an analphoid, inverted duplicated marker chromosome.  
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MA Ramírez Fernández, R García Cavazos, HF Sánchez Martínez  
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Dermatologic features in Pallister-Killian syndrome and their importance to the diagnosis.  
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Brain MRI findings of older patients with Pallister-Killian syndrome.

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Retrospective diagnosis of Pallister-Killian syndrome by CGH array.

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R Sánchez-Carpintero, A McLellan, L Parmeggiani, AE Cockwell, RJ Ellis, JH Cross, S Eckhardt, R Guerrini  
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Prenatal diagnosis of congenital diaphragmatic hernia in a fetus with 46.XY/46.X,-Y,+der(Y)t(Y;1)(q12;q12) mosaicism: a case report.

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Tetrasomy 12pter-12p13.31 in a girl with partial Pallister-Killian syndrome phenotype.

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Pallister-Killian syndrome presenting through nuchal oedema: cytogenetic investigation and parental origin by molecular analysis in a new case.

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[Behavioural phenotypes. Biologically determined neuropsychological patterns]  
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Partial tetrasomy 12pter-12p12.3 in a girl with Pallister-Killian syndrome: extraordinary finding of an analphoid, inverted duplicated marker.

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Pallister-Killian syndrome

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### DEVELOPMENT AND EDUCATION IN CHILDHOOD

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