

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

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Search parameters: "Pallister-Killian" & "PKS" in title, abstract, main journal.

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

Developmental and behavioral characteristics of individuals with Pallister-Killian syndrome.

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

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;55(8-9):480-4

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

Johnstone ED, Jones EA.

Ultrasound In Obstetrics & Gynecology : The Official Journal Of The International Society Of Ultrasound In Obstetrics And Gynecology Aug 2012,40(2):239-41

Occurrence and clinical features of epileptic and non-epileptic paroxysmal events in five children with **Pallister-Killian syndrome**.

Filloux FM, Carey JC, Krantz ID, Ekstrand JJ, Candee MS.

Eur J Med Genet. 2012 May;55(5):367-73.

Clinical and radiological findings in Pallister-Killian syndrome.

Jamuar S, Lai A, Unger S, Nishimura G.

Eur J Med Genet. 2012 Mar;55(3):167-72.

Array CGH on unstimulated blood does not detect all cases of Pallister-Killian syndrome: a skin biopsy should remain the diagnostic gold standard.

Hodge JC, Hulshizer RL, Seger P, St Antoine A, Bair J, Kirmani S.

Am J Med Genet A. 2012 Mar;158A(3):669-73

Array CGH on unstimulated blood does not detect all cases of Pallister-Killian syndrome: a skin biopsy should remain the diagnostic gold standard.

Hodge JC, Hulshizer RL, Seger P, St Antoine A, Bair J, Kirmani S.

Am J Med Genet A. 2012 Mar;158A(3):669-73.

Congenital diaphragmatic hernia.

Tovar JA.

Orphanet J Rare Dis. 2012 Jan 3;7:1. Review.

2011

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. 2011 Oct 20.

Anesthetic management of Pallister-Killian syndrome using a Bispectral Index monitor in a patient with severe seizures.

Kira S.

J Clin Anesth. 2011 Dec;23(8):674-6.

Amniocentesis can be useful during the third trimester of pregnancy for antenatal diagnosis of Pallister-Killian syndrome: a case report.

Murakami M, Iwasa T, Takahashi Y, Morine M.

Clin Exp Obstet Gynecol. 2011;38(3):269-71.

Diagnosis of Pallister-Killian syndrome by array comparative genome hybridization from a spleen sample.

Harrison V, Williams R, Connell L, Kini U.

Clin Dysmorphol. 2011 Jan;20(1):58-60.

Current World Literature

Current Opinion In Obstetrics And Gynecology Apr 2011,23(2):135-141

2010

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.

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.

Liehr T, Karamysheva T, Merkas M, Brecevic L, Hamid AB, Ewers E, Mrasek K, Kosyakova N, Weise A.
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.
Clin Dysmorphol. 2010 Aug 27.

Pallister-Killian syndrome with additional manifestations of cleft palate and sacral appendage.

Chaouachi S, Ben Hamida E, Ennine I, Chaabouni M, Sfar R, Chaabouni H, Marrakchi Z.
Tunis Med. 2010 Aug;88(8):614-6.

Trisomy 12p syndrome secondary to a balanced familial translocation.

Zen PR, Rosa RF, Rosa RC, Graziadio C, Paskulin GA.
Pediatr Int. 2010 Jun;52(3):e144-6

Mosaic tetrasomy 12p with discrepancy between fetal tissues and extraembryonic tissues: molecular analysis and possible mechanism of formation.

Chen CP, Su YN, Chern SR, Tsai FJ, Wu PC, Chen HE, Chiang SS, Wang W.
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.

Chen CP, Su YN, Hsu CY, Tsai FJ, Chien SC, Chern SR, Lee MS, Wu PC, Chen HE, Wang W.
Taiwan J Obstet Gynecol. 2010 Mar;49(1):124-8. No abstract available.

Pallister Killian syndrome: unusual significant postnatal overgrowth in a girl with otherwise typical presentation.

Frković SH, Durisević IT, Trečić RL, Sarnavka V, Gornik KC, Muzinić D, Letica L, Barić I, Begović D.
Coll Antropol. 2010 Mar;34(1):247-50.

Phenotypic overlapping of trisomy 12p and Pallister-Killian syndrome.

Inage E, Suzuki M, Minowa K, Akimoto N, Hisata K, Shoji H, Okumura A, Shimojima K, Shimizu T, Yamamoto T.
Eur J Med Genet. 2010 May-Jun;53(3):159-61.

Iris transillumination defects associated with Pallister-Killian syndrome.

Ticho BH.
J Pediatr Ophthalmol Strabismus. 2010 Jan-Feb;47(1):58-9.

Late-Onset Epileptic Spasms in Children With Pallister-Killian Syndrome: A Report of Two New Cases and Review of the Electroclinical Aspects.

Cerminara C, Compagnone E, Bagnolo V, Galasso C, Lo-Castro A, Brinciotti M, Curatolo P.
J Child Neurol. 2010 Feb;25(2):238-45.

2009

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

Park IY, Shin JC, Kwon JY, Koo BK, Kim M, Lim J, Kim Y, Han K.
Korean J Lab Med. 2009 Aug;29(4):366-70.

Genetic considerations in the prenatal diagnosis of overgrowth syndromes.

Vora N, Bianchi DW.
Prenat Diagn. 2009 Jul 16;29(10):923-929.

The phenotypic spectrum of trisomy 2: report of two new cases.

Mihci E, Velagaleti GV, Ensenaer R, Babovic-Vuksanovic D.
Clin Dysmorphol. 2009 Oct;18(4):201-4.

aCGH detects partial tetrasomy of 12p in blood from Pallister-Killian syndrome cases without invasive skin biopsy.

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.

Kunz J, Schoner K, Stein W, Rehder H, Fritz B.
Arch Gynecol Obstet. 2009 Dec;280(6):1049-53.

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

Vogel I, Lyngbye T, Nielsen A, Pedersen S, Hertz JM.
Am J Med Genet A. 2009 Mar;149A(3):510-4.

Pallister-Killian syndrome caused by mosaicism for a supernumerary ring chromosome 12p.

Yeung A, Francis D, Giouzeppos O, Amor DJ.
Am J Med Genet A. 2009 Mar;149A(3):505-9.

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

Kolarski M, Joksić G, Beres M, Krstić A, Joksić I, Dobrojević B, Nikić S.
Arch Gynecol Obstet. 2009 Mar;279(3):377-9.

2008

Pallister-Killian Syndrome (PKS) as a Cause of Mental Retardation.

Shamdeen A, Meyer S, Gottschling S, Oehl-Jaschkowitz B, Gortner L, Shamdeen MG.
Klin Padiatr. 2008 Mar-Apr;221(2):97-9

The Pallister-Killian syndrome in a child with rare karyotype--a diagnostic problem.

R Smigiel, J Pilch, I Makowska, H Busza, R Slezak, MM Sasiadek
Eur J Pediatr. 2008 Sep;167(9):1063-5

Anesthesia for orthopedic surgery in Pallister-Killian syndrome.

J Knab_EW Heupel_D Steinmann
Paediatr Anaesth. 2008 Jul;18(7):682-4.

Valproate-associated reversible encephalopathy in a 3-year-old girl with Pallister-Killian syndrome.

T Gerstner, N Bell, SA Koenig
Therapeutics and clinical risk management. 2008 Jun;4(3):645-7

Pallister-Killian syndrome: a report of 2 cases and review of its surgical aspects.

M Baglaj, J King, R Carachi
J Pediatr Surg. 2008 Jun;43(6):1218-21.

Prenatal diagnosis of Pallister-Killian syndrome in two fetuses with increased nuchal translucency.

MH Kim, SY Park, MY Kim, BY Lee, MH Lee, HM Ryu
Prenat Diagn. 2008 May;28(5):454-6.

Fetal facial profile in Pallister-Killian syndrome.

M Liberati, K Melchiorre, I D'Emilio, PE Guanciali-Franchi, I Iezzi, S Rotmensch, C Celentano
Fetal Diagn Ther. 2008;23(1):15-7

2007

Mosaic tetrasomy 12p with triplication of 12p detected by array-based comparative genomic hybridization of peripheral blood DNA.
Z Powis, SH Kang, ML Cooper, A Patel, DA Peiffer, A Hawkins, R Heidenreich, KL Gunderson, SW Cheung, RP Erickson
Am J Med Genet A. 2007 Dec 15;143A(24):2910-5.

Pallister-Killian syndrome: tetrasomy of 12pter-->12p11.22 in a boy with an analphoid, inverted duplicated marker chromosome.
XL Huang, M Isabel de Michelena, E Leon, TA Maher, R McClure, A Milunsky
Clin Genet. 2007 Nov;72(5):434-40.

A case of Pallister-Killian syndrome associated with West syndrome.
H Yamamoto, M Fukuda, H Murakami, N Kamiyama, Y Miyamoto
Pediatr Neurol. 2007 Sep;37(3):226-8.

Pallister-Killian syndrome. Case report
MA Ramírez Fernández, R García Cavazos, HF Sánchez Martínez
Ginecol Obstet Mex. 2007 Jul;75(7):412-8. Spanish.

Dermatologic features in Pallister-Killian syndrome and their importance to the diagnosis.
E Guareschi, L Garavelli, S Pedori, V Di Lernia, L Grenzi, F Franchi, M Marinelli, GF Croci, E Pedretti, S Amarri, ...
Pediatr Dermatol. 2007 Jul-Aug;24(4):426-8

Chromosomal abnormalities associated with omphalocele.
CP Chen
Taiwan J Obstet Gynecol. 2007 Mar;46(1):1-8. Review.

2006

Detection of low-level mosaicism by array CGH in routine diagnostic specimens.

BC Ballif, EA Rorem, K Sundin, M Lincicum, S Gaskin, J Coppinger, CD Kashork, LG Shaffer, BA Bejjani
Am J Med Genet A. 2006 Dec 15;140(24):2757-67.

Pallister-Killian syndrome presenting with a complex congenital heart defect and increased nuchal translucency.

DE Abad, JA Gabarre, AM Izquierdo, C López-Sánchez, V García-Martínez, AG Izquierdo
J Ultrasound Med. 2006 Nov;25(11):1475-80.

High cognitive functioning and behavioral phenotype in Pallister-Killian syndrome.

HJ Stalker, BA Gray, A Bent-Williams, RT Zori
Am J Med Genet A. 2006 Sep 15;140(18):1950-4.

Pallister-Killian syndrome: Multiband FISH of tetrasomy 12p.

AM Gerdes, LK Hansen, F Brandrup, K Soegaard, A Christoffersen, K Rasmussen
Pediatr Dermatol. 2006 Jul-Aug;23(4):378-81.

Pallister-Killian syndrome: report of one case.

HC Wu, LH Lin, LP Tsai, CH Huang, KL Hung, HT Liao
Acta Paediatr Taiwan. 2006 May-Jun;47(3):139-41.

Brain MRI findings of older patients with Pallister-Killian syndrome.

Y Saito, K Masuko, K Kaneko, Y Chikumaru, K Saito, H Iwamoto, A Matsui, N Aida, K Kurosawa, Y Kuroki, ...
Brain Dev. 2006 Jan;28(1):34-8.

Retrospective diagnosis of Pallister-Killian syndrome by CGH array.

A Delahaye, E Pipiras, C Delorme-Vincent, M Benkhalifa, S Kasakyan, L Devisme, JP Wolf, B Benzacken
Fetal Diagn Ther. 2006;21(6):485-8.

Pallister-Killian syndrome (PKS): clinical case report.

AL de Oliveira, Ade O Ortega, AL Ciamponi
J Clin Pediatr Dent. 2006 Spring;30(3):257-60.

Small supernumerary marker chromosomes--progress towards a genotype-phenotype correlation.

Liehr T, Mrasek K, Weise A, Dufke A, Rodríguez L, Martínez Guardia N, Sanchis A, Vermeesch JR, Ramel C, Polityko A, Haas OA, Anderson J, Claussen U, von Eggeling F, Starke H.
Cytogenet Genome Res. 2006;112(1-2):23-34.

2005

Forty-two supernumerary marker chromosomes (SMCs) in 43,273 prenatal samples: chromosomal distribution, clinical findings, and UPD studies.

O Bartsch, A Loitzsch, P Kozlowski, ML Mazauric, G Hickmann
European journal of human genetics : 2005 Nov;13(11):1192-204.

Pallister-Killian syndrome: an unusual cause of epileptic spasms.

R Sánchez-Carpintero, A McLellan, L Parmeggiani, AE Cockwell, RJ Ellis, JH Cross, S Eckhardt, R Guerrini
Developmental medicine and child neurology. 2005 Nov;47(11):776-9.

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.

HY Ahn, JC Shin, YH Kim, HS Ko, IY Park, SJ Kim, JG Rha, SP Kim
Journal of Korean medical science. 2005 Oct;20(5):895-8

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

JR Vermeesch, C Melotte, I Salden, M Riegel, V Trifnov, A Polityko, N Romyantseva, I Naumchik, H Starke, G Matthijs, ...
European journal of medical genetics. 2005 Jul-Sep ;48(3):319-27.

Pallister-Killian syndrome: rapid decrease of isochromosome 12p frequency during amniocyte subculturing. Conclusion for strategy of prenatal cytogenetic diagnostics.

AD Polityko, E Goncharova, L Shamgina, N Drozdovskaja, L Podleschuk, E Abramchik, E Jaroshevich, O Khurs, I Pizarik, O Pribushenya, ...
The journal of histochemistry and cytochemistry : 2005 Mar;53(3):361-4.

Cell death as a possible mechanism for tissue limited mosaicism in Pallister-Killian syndrome.

W Tang, SL Wenger
Journal of the Association of Genetic Technologists. 2005;31(4):168-9

2004

Post-zygotic origin of isochromosome 12p.

TJ de Ravel, K Keymolen, E van Assche, I Wittevronghel, P Moerman, I Salden, G Matthijs, JP Fryns, JR Vermeesch
Prenatal diagnosis. 2004 Dec 15;24(12):984-8.

[Anesthesia in child with Pallister-Killian syndrome: case report.]

Cruz JR, Videira RL.
Rev Bras Anesthesiol. 2004 Oct;54(5):677-80. Portuguese.

Assisted reproductive technology and congenital overgrowth: some speculations on a case of Pallister-Killian syndrome.

P Chiurazzi, J Bajer, E Tabolacci, MG Pomponi, R Lecce, M Zollino, G Neri
American journal of medical genetics. Part A. 2004 Oct 15 130A(3):315-6.

Genotype/phenotype analysis in a patient with pure and complete trisomy 12p.

W Zumkeller, M Volleth, P Muschke, H Tönnies, A Heller, T Liehr, P Wieacker, M Stumm
American journal of medical genetics. Part A. 2004 Sep 1 129A(3):261-4.

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

V Antonella, G Pantaleo, C Anna Irma, C Savino, L Selvaggi
Prenat Diagn. 2004 Mar;24(3):229-30.

Small supernumerary marker chromosomes (sSMC) in humans.

Liehr T, Claussen U, Starke H.
Cytogenet Genome Res. 2004;107(1-2):55-67.

A newborn infant with left diaphragm agenesis, radial aplasia and preauricular appendices.

N Oner, UN Başaran, O Yalçın, U Vatansver, B Acunaş
Clin Dysmorphol. 2004 Jan;13(1):59-60.

Congenital diaphragmatic hernia and chromosomal anomalies: autopsy study.

D Borys, JB Taxy
Pediatr Dev Pathol. 2004 Jan-Feb;7(1):35-8.

2003

Clinical, cytogenetic, and molecular observations in a patient with Pallister-Killian syndrome with an unusual karyotype.

B Leube, F Majewski, J Gebauer, B Royer-Pokora
Am J Med Genet A. 2003 Dec 15;123A(3):296-300.

Clinical overlap of OFD type IX with Pallister-Killian syndrome (tetrasomy 12p).

D Geneviève, Y Sznajer, M Raoul, D Sanlaville, A Verloes, MF Portnoi, C Bauman
Am J Med Genet A. 2003 Oct 1;122A(2):180-2.

Anaesthetic management of a child with Pallister-Killian syndrome.

T Iacobucci, M Galeone, G De Francisci
Paediatr Anaesth. 2003 Jun;13(5):457-9.

[Behavioural phenotypes. Biologically determined neuropsychological patterns]

Ruggieri VL, Arberas CL.
Rev Neurol. 2003 Aug 1-15;37(3):239-53. Review. Spanish.

Tetrasomy 12p--unusual presentation in CVS.

L Dong, RE Falk, J Williams, M Kohan, RR Schreck
Prenat Diagn. 2003 Feb;23(2):101-3.

Mild phenotype in a 15-year-old boy with Pallister-Killian syndrome.

D Genevieve, V Cormier-Daire, D Sanlaville, L Faivre, P Gosset, L Allart, M Picq, A Munnich, S Romana, M de Blois, ...
Am J Med Genet A. 2003 Jan 1;116A(1):90-3. Review.

A rapid and noninvasive method for detecting tissue-limited mosaicism: detection of i(12)(p10) in buccal smear from a child with Pallister-Killian syndrome.

GV Velagaleti, JK Tapper, BA Rampy, S Zhang, JC Hawkins, LH Lockhart
Genet Test. 2003 Fall;7(3):219-23.

2002

Severe hearing loss in Pallister-Killian syndrome.

M Schuster, U Hoppe, U Eysholdt, F Rosanowski
ORL J Otorhinolaryngol Relat Spec. 2002 Sep-Oct;64(5):343-5.

Bilateral congenital diaphragmatic hernia: Differentiation between Pallister-Killian and Fryns syndromes.

A Veldman, R Schlösser, A Allendorf, D Fischer, K Heller, B Schaeff, S Fuchs
Am J Med Genet. 2002 Jul 22;111(1):86-7.

Tissue-limited mosaicism in Pallister-Killian syndrome -- a case in point.

S Choo, SH Teo, M Tan, MH Yong, LY Ho
J Perinatol. 2002 Jul-Aug;22(5):420-3.

Pallister-Killian syndrome: difficulties of prenatal diagnosis.

B Doray, F Girard-Lemaire, B Gasser, JJ Baldauf, B De Geeter, M Spizzo, C Zeidan, E Flori
Prenat Diagn. 2002 Jun;22(6):470-7.

2001

Partial tetrasomy 12pter-12p12.3 in a girl with Pallister-Killian syndrome: extraordinary finding of an analphoid, inverted duplicated marker.

A Dufke, C Walczak, T Liehr, H Starke, V Trifonov, N Rubtsov, M Schöning, H Enders, T Eggermann
Eur J Hum Genet. 2001 Aug;9(8):572-6.

Ultrastructural examination of the axillary skin biopsy in the diagnosis of metabolic diseases.

CM Abramovich, RA Prayson, JT McMahon, BH Cohen
Hum Pathol. 2001 Jun;32(6):649-55.

Pallister-Killian mosaic syndrome

H Kawame
Ryoikibetsu Shokogun Shirizu. 2001;(34 Pt 2):464-5. Review. Japanese.

2000

Prospective ultrasound diagnosis of Pallister-Killian syndrome in the second trimester of pregnancy: the importance of the fetal facial profile.

D Paladini, A Borghese, M Arienzo, A Teodoro, P Martinelli, C Nappi
Prenat Diagn. 2000 Dec;20(12):996-8.

Pallister-Killian syndrome: case report with pineal tumor.

L Mauceri, G Sorge, G Incorpora, L Pavone
Am J Med Genet. 2000 Nov 6;95(1):75-8.

Pallister-Killian syndrome presenting through nuchal translucency screening for trisomy 21.

K Langford, S Hodgson, M Seller, D Maxwell
Prenat Diagn. 2000 Aug;20(8):670-2.

The Pallister-Killian syndrome is reliably diagnosed by FISH on buccal mucosa.

BF Manasse, N Lekgate, WM Pfaffenzeller, TJ de Ravel
Clin Dysmorphol. 2000 Jul;9(3):163-5.

Pallister-Killian syndrome

T Hasegawa
Ryoikibetsu Shokogun Shirizu. 2000;(30 Pt 5):26-9. Review. Japanese.

1999

Retinal pigment mosaicism in Pallister-Killian syndrome (mosaic tetrasomy 12p).

W Graham, SM Brown, F Shah, VS Tonk, MK Kukulich
Arch Ophthalmol. 1999 Dec;117(12):1648-9.

Parental origin of the isochromosome 12p in Pallister-Killian syndrome: molecular analysis of one patient and review of the reported cases.

JL Struthers, CD Cuthbert, MM Khalifa
Am J Med Genet. 1999 May 21;84(2):111-5. Review.

Chromosome instability limited to the aneuploid clone in the Pallister-Killian syndrome: a pitfall in prenatal diagnosis.

M Zollino, J Bajer, G Neri
Prenat Diagn. 1999 Feb;19(2):184-5.

1998

Pallister-Killian syndrome [i(12p)]: first pre-natal diagnosis using cordocentesis in the second trimester confirmed by in situ hybridization.

J Chiesa, M Hoffet, O Rousseau, JM Bourgeois, P Sarda, P Mares, JP Bureau
Clin Genet. 1998 Oct;54(4):294-302. Review.

Pallister-Killian syndrome: the first reported case in Hong Kong.

IF Lo, LY Cheung, FW Lam, ST Lam
Zhonghua Min Guo Xiao Er Ke Yi Xue Hui Za Zhi. 1998 Sep-Oct;39(5):333-5.

Prenatal diagnosis of genetic syndromes may be facilitated by serendipitous findings at fetal blood sampling.

F Lalatta, S Salmona, R Fogliani, T Rizzuti, U Nicolini
Prenat Diagn. 1998 Aug;18(8):834-7.

1997

A case of mosaic tetrasomy 12p (Pallister-Killian Syndrome) diagnosed prenatally: comparison of chromosome analyses of various cells obtained from the patient.

K Takakuwa, I Hataya, M Arakawa, M Tamura, N Sekizuka, K Tanaka
Am J Perinatol. 1997 Nov;14(10):641-3.

Report of two new cases of Pallister-Killian syndrome confirmed by FISH: tissue-specific mosaicism and loss of i(12p) by in vitro selection.

R Schubert, R Viersbach, T Eggermann, M Hansmann, G Schwanitz
Am J Med Genet. 1997 Mar 17;69(2):166-8

Prezygotic origin of the isochromosome 12p in Pallister-Killian syndrome.

V Cormier-Daire, M Le Merrer, N Gigarel, N Morichon, M Prieur, S Lyonnet, M Vekemans, A Munnich
Am J Med Genet. 1997 Mar 17;69(2):166-8.

The use of interphase FISH for prenatal diagnosis of Pallister-Killian syndrome.

PA Mowery-Rushton, MP Stadler, SJ Kochmar, E McPherson, U Surti, WA Hogge
Prenat Diagn. 1997 Mar;17(3):255-65

Clinical variability of tetrasomy 12p.

GB Schaefer, A Jochar, R Muneer, WG Sanger
Clin Genet. 1997 Feb;51(2):102-8.

Collaborative study of mosaic tetrasomy 12p or Pallister-Killian syndrome (nineteen fetuses or children).

M Mathieu, C Piussan, F Thepot, A Gouget, D Lacombe, JM Pedespan, F Serville, D Fontan, M Ruffie, A Nivelon-Chevallier,...
Ann Genet. 1997;40(1):45-54.

1996

Pallister-Killian syndrome: a mild case diagnosed by fluorescence in situ hybridization. Review of the literature and expansion of the phenotype.

MM Bielanska, MM Khalifa, AM Duncan
Am J Med Genet. 1996 Oct 16;65(2):104-8.

Genetics casebook. Pallister-Killian syndrome.

J Smulian, E Guzman, C Mohan, B Weinberger, ML Hanley, R Richardson
J Perinatol. 1996 Sep-Oct;16(5):406-12. Review.

Parental origin and mechanisms of formation of three cases of 12p tetrasomy.

C Turleau, B Simon-Bouy, E Austruy, MC Grisard, F Lemaire, D Molina-Gomes, JP Siffroi, J Boué
Clin Genet. 1996 Jul;50(1):41-6.

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

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1995

Prenatal diagnosis of mosaic tetrasomy 12p/trisomy 12p by fluorescent in situ hybridization in amniotic fluid cells: a case report of Pallister-Killian syndrome.

FJ Los, D Van Opstal, MP Schol, JL Gaillard, H Brandenburg, AM Van Den Ouweland, PA in 't Veld
Prenatal diagnosis. 1995 Dec 15(12):1155-9.

Pallister-Killian syndrome detected by fluorescence in situ hybridization.

MG Bulter, VG Dev
American journal of medical genetics. 1995 Jul 3 57(3):498-500.

A 10-year survey, 1980-1990, of prenatally diagnosed small supernumerary marker chromosomes, identified by FISH analysis.

Outcome and follow-up of 14 cases diagnosed in a series of 12,699 prenatal samples.

K Brøndum-Nielsen, M Mikkelsen
Prenatal diagnosis. 1995 Jul 15(7):615-9.

Hypopigmentation of the fundi associated with Pallister-Killian syndrome.

M Birch, A Patterson, A Fryer
Journal of pediatric ophthalmology and strabismus. 1995 Mar-Apr 32(2):128-31.

Failure of amniotic-fluid-cell growth: is it related to fetal aneuploidy?

WH Persutte, RR Lenke

Lancet. 1995 Jan 14 345(8942):96-7.

The Pallister-Killian syndrome in an African individual.

BF Woodman, MA Jordan, LI Moller, JD Cartwright, TJ De Ravel

Genetic counseling (Geneva, Switzerland). 1995 6(1):33-6. Review

Pallister-Killian syndrome: normal karyotype in prenatal chorionic villi, in postnatal lymphocytes, and in slowly growing epidermal cells, but mosaic tetrasomy 12p in skin fibroblasts.

D Horn, F Majewski, B Hildebrandt, H Körner

Journal of medical genetics. 1995 Jan 32(1):68-71

1994

Lethal Pallister-Killian syndrome: phenotypic similarity with Fryns syndrome.

JJ Rodríguez, I Garcia, J Alvarez, A Delicado, J Palacios

American journal of medical genetics. 1994 Nov 1 53(2):176-81.

Tetrasomy 12p (Pallister-Killian syndrome): ultrasound indicators and confirmation by interphase fish.

RD Wilson, K Harrison, LA Clarke, SL Yong

Prenatal diagnosis. 1994 Sep 14(9):787-92.

Pallister-Killian syndrome: an unusual presentation.

M el-Naggar, M Hawthorne

The Journal of laryngology and otology. 1994 Aug 108(8):669-70.

Cytogenetic study of a severe case of Pallister-Killian syndrome using fluorescence in situ hybridization.

SM Gamal, T Hasegawa, H Satoh, T Watanabe, K Endo, Y Satoh

The Japanese journal of human genetics. 1994 Jun 39(2):259-67. Review.

Pallister-Killian and Fryns syndromes.

RF Stratton, CM Moore, CS Popham, BR DuPont, VL Mattern

American journal of medical genetics. 1994 May 15 51(1):90.

1993

Prenatal diagnosis of Pallister-Killian syndrome by chorionic villus sampling--its diagnostic problems.

AH Boyle, R Kulkarni, JS Smoleniec, T Davies, A McDermott

Prenatal diagnosis. 1993 Dec 13(12):1160-1

Pallister-Killian and Fryns syndromes: nosology.

EW McPherson, DM Ketterer, DJ Salsburey

American journal of medical genetics. 1993 Aug 15 47(2):241-5.

Pallister-Killian syndrome in older children and adolescents.

G Horneff, F Majewski, B Hildebrand, T Voit, HG Lenard
Pediatric neurology. 1993 Jul-Aug 9(4):312-5.

Diaphragmatic hernia in tetrasomy 12p mosaicism.

J Bergoffen, H Punnett, TJ Campbell, AJ Ross, E Ruchelli, EH Zackai
The Journal of pediatrics. 1993 Apr 122(4):603-6.

A case of neonatal Pallister-Killian syndrome (tetrasomy 12p)

J González de Dios, A García-Alix Pérez, A Díaz de Bustamante, A Delicado Navarro, S Arés Segura, S Salas Hernández, J Quero Jiménez
Anales españoles de pediatría. 1993 Mar 38(3):277-9. Spanish.

Molecular cytogenetic study of patients with Pallister-Killian syndrome.

M Larramendy, M Heiskanen, M Wessman, A Ritvanen, P Peltomäki, K Simola, H Kääriäinen, H von Koskull, M Kähkönen, S Knuutila
Human genetics. 1993 Mar 91(2):121-7.

Prenatal diagnosis of tetrasomy 12p (Pallister-Killian syndrome)

JH Priest
Prenatal diagnosis. 1993 Feb 13(2):152

New diagnostic method for Pallister-Killian syndrome: detection of i(12p) in interphase nuclei of buccal mucosa by fluorescence in situ hybridization.

H Ohashi, S Ishikiryama, Y Fukushima
American journal of medical genetics. 1993 Jan 1, 45(1):123-8.

1992

Prenatal diagnosis of tetrasomy 12p by in situ hybridization: varying levels of mosaicism in different fetal tissues.

JK Blancato, M Hunt, J George, J Katz, JM Meck
Prenatal diagnosis. 1992 Dec ;12(12):979-83..

Prenatal diagnosis of Pallister-Killian syndrome: resolution of cytogenetic ambiguity by use of fluorescent in situ hybridization.

S McLean, W Stanley, H Stern, J Fonda-Allen, G Devine, T Ellingham, K Rosenbaum
Prenatal diagnosis. 1992 Dec 12(12):985-91

A further prenatal diagnosis of mosaic tetrasomy 12p (Pallister-Killian syndrome)

MI Tejada, A Uribarren, P Briones, MA Vilaseca
Prenatal diagnosis. 1992 Jun;12(6):529-34.

Killian-Teschler-Nicola syndrome (Pallister-Killian syndrome, mosaic tetrasomy 12p)

R Pankau, U Diebold, J Jenderny, M Kautza, K Dörner
Monatsschr Kinderheilkd. 1992 Jun;140(6):340-2. German.

Failure of PHA-stimulated i(12p) lymphocytes to divide in Pallister-Killian syndrome.

SL Reeser, SL Wenger
American journal of medical genetics. 1992 Apr 1 ;42(6):815-9.

Tissue specificity and stability of mosaicism in Pallister-Killian +i(12p) syndrome: relevance for prenatal diagnosis.

JH Priest, JM Rust, PM Fernhoff

American journal of medical genetics. 1992 Apr 1 ;42(6):820-4.

Prenatal diagnosis of the Pallister-Killian mosaic aneuploidy syndrome by CVS.

J Bernert, I Bartels, G Gatz, I Hansmann, M Heyat, PD Niedmann, H Rehder, C Waldenmaier, B Zoll

American journal of medical genetics. 1992 Mar 1;42(5):747-50.

Pericardial agenesis and focal aplasia cutis in tetrasomy 12p (Pallister-Killian syndrome).

MF Zakowski, Y Wright, A Ricci

American journal of medical genetics. 1992 Feb 1;42(3):323-5.

Sister chromatid differentiation and chromosomal in situ suppression hybridization: a combined methodology for analyzing cell proliferation and SCEs in individual chromosomes.

S Knuutila, M Larramendy

Cytogenetics and cell genetics. 1992;61(2):99-102

1991

Pallister-Killian syndrome: characterization of the isochromosome 12p by fluorescent in situ hybridization.

F Speleman, JG Leroy, N Van Roy, A De Paepe, R Suijkerbuijk, H Brunner, L Looijenga, MR Verschraegen-Spae, E Orye

American journal of medical genetics. 1991 Dec 1;41(3):381-7.

Pallister-Killian syndrome: additional manifestations of cleft palate and sacral appendage.

DR McLeod, LR Wesselman, DI Hoar

Journal of medical genetics. 1991 Aug;28(8):541-3. Review

Pallister-Killian syndrome diagnosed by chorionic villus sampling.

M Sharland, L Hill, R Patel, M Patton

Prenatal diagnosis. 1991 Jul;11(7):477-9.

Pallister-Killian syndrome and 12p tetrasomy: increased LDH-B activity

J Antich Femenias, MP Briones Godino, MA Vilaseca Busca, ML Girós Blasco, J Campos Castelló, B Jaume Roig, N Clusellas Casals

An Esp Pediatr. 1991 Jun;34(6):459-62. Review. Spanish.

Tetrasomy 12p (Pallister-Killian syndrome).

A Schinzel

Journal of medical genetics. 1991 Feb 28(2):122-5.

Molecular cytogenetics of alpha satellite DNA from chromosome 12: fluorescence in situ hybridization and description of DNA and array length polymorphisms.

GM Greig, S Parikh, J George, VE Powers, HF Willard

Cytogenetics and cell genetics. 1991 ;56(3-4):144-8.

Pallister Killian--mosaic tetrasomy 12 p syndrome. Another prenatally diagnosed case.

Bresson JL, Arbez-Gindre F, Peltie J, Gouget A.

Prenat Diagn. 1991 Apr;11(4):271-5.

1990

Prenatal diagnosis of Pallister-Killian syndrome.

S Soukup, K Neidich
American journal of medical genetics. 1990 Apr ;35(4):526-8.

Mosaicism in Pallister i(12p) syndrome.

Wenger SL, Boone LY, Steele MW.
Am J Med Genet. 1990 Apr;35(4):523-5.

Trisomy 12p mosaicism syndrome.

Karki CB, Walters RM.
J Ment Defic Res. 1990 Feb;34 (Pt 1):75-80.
(possibly *PKS* related)

1989

Molecular analysis of the isochromosome 12P in the Pallister-Killian syndrome. Construction of a mouse-human hybrid cell line containing an i(12p) as the sole human chromosome.

J Zhang, P Marynen, K Devriendt, JP Fryns, H Van den Berghe, JJ Cassiman
Human genetics. 1989 Nov;83(4):359-63.

Tetrasomy 12p (Pallister-Killian syndrome): possible diagnosis before the age of a year

K Chrzanowska, JP Fryns
Journal de génétique humaine. 1989 Sep;37(3):259-61. French.

Lethal presentation of mosaic tetrasomy 12p (Pallister-Killian) syndrome.

ID Young, DP Duckett, KM O'Reilly
Annales de génétique. 1989;32(1):62-4.

1988

Isochromosome 12p mosaicism (Pallister-Killian syndrome): newborn diagnosis by direct bone marrow analysis.

BE Ward, MW Hayden, A Robinson
American journal of medical genetics. 1988 Dec;31(4):835-9.

Pallister-Killian mosaic syndrome with emphasis on the adult phenotype.

OW Quarrell, MA Hamill, HE Hughes
American journal of medical genetics. 1988 Dec ;31(4):841-4.

Case of Pallister-Killian syndrome with imperforate anus.

AE Lin, M Clemens, KL Garver, SL Wenger, MW Steele
American journal of medical genetics. 1988 Nov 31(3):705-7.

Prenatal diagnosis of tetrasomy 47,XY,+i(12p) confirmed by in situ hybridization.

Shivashankar L, Whitney E, Colmorgen G, Young T, Munshi G, Wilmoth D, Byrne K, Reeves G, Borgaonkar DS, Picciano SR, et al.
Prenat Diagn. 1988 Feb;8(2):85-91.

Risk effect of maternal age in Pallister i(12p) syndrome.

Wenger SL, Steele MW, Yu WD..

Clin Genet. 1988 Sep;34(3):181-184.

Pallister-Killian syndrome: cytogenetic and biochemical studies.

K Narahara, Y Wakita, K Kikkawa, K Hiramoto, H Namba, M Murakami, R Kasai, H Kimoto

Jinrui idengaku zasshi. 1988 Sep;33(3):339-47.

Pallister-Killian syndrome--an update of a clinical case.

PR Wyatt

American journal of medical genetics. 1988 Jan;29(1):229.

1987

Pallister-Killian syndrome: cytogenetic and molecular studies.

P Peltomäki, S Knuutila, A Ritvanen, I Kaitila, A de la Chapelle

Clinical genetics. 1987 Jun;31(6):399-405.

Isochromosome 12p mosaicism (Pallister mosaic aneuploidy or Pallister-Killian syndrome): report of 11 cases.

JF Reynolds, A Daniel, TE Kelly, SM Gollin, MJ Stephan, J Carey, WN Adkins, MJ Webb, F Char, JF Jimenez

American journal of medical genetics. 1987 Jun;27(2):257-74

Skeletal anomalies in a patient with the Pallister/Teschler-Nicola/Killian syndrome.

Kawashima H.

Am J Med Genet. 1987 Jun;27(2):285-9. Erratum in: *Am J Med Genet* 1987 Oct;28(2):527.

Mosaic tetrasomy 12p: four new cases, and confirmation of the chromosomal origin of the supernumerary chromosome in one of the original Pallister-Mosaic syndrome cases.

Warburton D, Anyane-Yeboah K, Francke U.

Am J Med Genet. 1987 Jun;27(2):275-83.

[Mosaic tetrasomy 12p. Identical nature of the Pallister syndrome, the Teschler-Nicola/Killian syndrome and mosaic tetrasomy 21]

Gilgenkrantz S, Fryns JP, Droulle P, Schweitzer M, Chadeaux B, Prieur M.

J Genet Hum. 1987 Jan;35(1):51-61. French.

1986

Chromosomal mosaicism in the Killian/Teschler-Nicola syndrome.

Raffel LJ, Mohandas T, Rimoin DL.

Am J Med Genet. 1986 Aug;24(4):607-11.

1985

The characteristic physiognomy and tissue specific karyotype distribution in the Pallister-Killian syndrome.

AG Hunter, B Clifford, DM Cox
Clinical genetics. 1985 Jul;28(1):47-53.

Mosaic tetrasomy 12p.

Gilgenkrantz S, Droulle P, Schweitzer M, Foliguet B, Chadefaux B, Lombard M, Chery M, Prieur M.
Clin Genet. 1985 Dec;28(6):495-502.

1983

"Killian Syndrome", Pallister mosaic syndrome, or mosaic tetrasomy 12P? - an analysis.

Buyse ML, Korf BR.
J Clin Dysmorphol. 1983 Fall;1(3):2-5.

Abnormal hair, craniofacial dysmorphism, and severe mental retardation - a new syndrome?

Killian W, Zonana J, Schroer RJ.
J Clin Dysmorphol. 1983 Fall;1(3):6-13.

First report of mosaic trisomy 12 in a liveborn individual.

Patil SR, Bosch EP, Hanson JW.
Am J Med Genet. 1983 Mar;14(3):453-60.

1979

Trisomy 12p syndrome: de novo occurrence of mosaic trisomy 12p in a mentally retarded boy.

Kondo I, Hamaguchi H, Haneda T.
Hum Genet. 1979 Jan 25;46(2):135-40.

1977

Syndrome +12p. Case report and review.

Tenconi R, Piovani E, Preto A, Magnabosco R, Baccichetti C.
Hum Genet. 1977 Nov 2;39(1):97-101.

The pallister mosaic syndrome.

**Pallister PD, Meisner LF, Elejalde BR, Francke U, Herrmann J, Spranger J, Tiddy W, Inhorn SL, Opitz JM.
Birth Defects Orig Artic Ser. 1977;13(3B):103-110.**

12P trisomy: a syndrome?

Hoo JJ.
Ann Genet. 1976 Dec;19(4):261-3.
(*Unsure if this is PKS related*)

The 12p trisomy syndrome.

Armendares S, Salamanca F, Nava S, Ramirez S, Cantu JM.
Ann Genet. 1975 Jun;18(2):89-94.
(*Unsure if this is PKS related*)