

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

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(updated: 20 June 2017)

## 2017

### Pallister-Killian syndrome in a two-year-old boy

Leigh Stone, Ramya Tripuraneni, Michelle Bain, Claudia Hernandez  
Clin Case Rep. 2017 Jun; 5(6): 774–777.

### Unclassifiable pattern of hypopigmentation in a patient with mosaic partial 12p tetrasomy without Pallister-Killian syndrome.

Alesi V, Dentici ML, Restaldi F, Orlando V, Liambo MT, Calacci C, Capolino R, Digilio MC, Hachem ME, Novelli A, Diociaiuti A, Dallapiccola B.  
Am J Med Genet A. 2017 May 10.

### Targeted prenatal diagnosis of Pallister-Killian syndrome.

Kucińska-Chahwan A, Bijok J, Dąbkowska S, Józwiak A, Ilnicka A, Nowakowska B, Jakiel G, Roszkowski T.  
Prenat Diagn. 2017 May;37(5):446-452.

### Using Array-Based Comparative Genomic Hybridization to Diagnose Pallister-Killian Syndrome

Mi-Na Lee, Jiwon Lee, Hee Joon Yu, Jeehun Lee, Sun-Hee Kim  
Ann Lab Med. 2017 Jan; 37(1): 66–70.

## 2016

### Pallister-Killian syndrome: Cytogenetics and molecular investigations of mosaic tetrasomy 12p in prenatal chorionic villus and in amniocytes. Strategy of prenatal diagnosis.

Libotte F, Bizzoco D, Gabrielli I, Mesoraca A, Cignini P, Vitale SG, Marilli I, Gulino FA, Rapisarda AM, Giorlandino C.  
Taiwan J Obstet Gynecol. 2016 Dec;55(6):863-866.

### Oro-dental features of Pallister-Killian syndrome: Evaluation of 21 European probands.

Bagattoni S, D'Alessandro G, Sadotti A, Alkhamis N, Rocca A, Cocchi G, Krantz ID, Piana G.  
Am J Med Genet A. 2016 Sep;170(9):2357-64.

### Rare case of Killian-Pallister syndrome associated with idiopathic short stature detected with fluorescent in situ hybridization on buccal smear.

Sukarova-Angelovska E, Kocova M, Ilieva G, Angelkova N, Kochova E.  
Mol Cytogenet. 2016 May 3;9:38.

### [Prenatal and clinicopathological study of 6 cases of Pallister-Killian syndrome and review].

Desseauve D, Legendre M, Dugué-Maréchaud M, Vequeau-Goua V, Pierre F.  
Gynecol Obstet Fertil. 2016 Apr;44(4):200-6. French.

### Dental Treatment of a Child with Pallister-Killian Syndrome.

Didinen S, Atabek D, Kip G, Patr Münevveroğlu A, Tulunoğlu Ö.  
Case Rep Dent. 2016;2016:4130961.

Prenatally identified Pallister-Killian syndrome: Ultrasound pattern and diagnostic considerations.

Santamaria A, Laganà AS, Barresi V, Triolo O, Fondacaro NS, Corrado F.  
J Obstet Gynaecol. 2016 Apr;36(3):406-7.

Methylation and expression analyses of Pallister-Killian syndrome reveal partial dosage compensation of tetrasomy 12p and hypomethylation of gene-poor regions on 12p.

Davidsson J, Johansson B.  
Epigenetics. 2016 Mar 3;11(3):194-204.

Mosaic ratio quantification of isochromosome 12p in Pallister-Killian syndrome using droplet digital PCR.

Fujiki K, Shirahige K, Kaur M, Deardorff MA, Conlin LK, Krantz ID, Izumi K.  
Mol Genet Genomic Med. 2016 Jan 20;4(3):257-61.

## 2015

Small supernumerary marker chromosomes and their correlation with specific syndromes.

Jafari-Ghahfarokhi H, Moradi-Chaleshtori M, Liehr T, Hashemzadeh-Chaleshtori M, Teimori H, Ghasemi-Dehkordi P.  
Adv Biomed Res. 2015 Jul 27;4:140.

Pallister-Killian syndrome: a study of 22 British patients.

Blyth M, Maloney V, Beal S, Collinson M, Huang S, Crolla J, Temple IK, Baralle D.  
J Med Genet. 2015 Jul;52(7):454-64.

Cytogenomic delineation and clinical follow-up of 10 Brazilian patients with Pallister-Killian syndrome.

Costa LS, Zandona-Teixeira AC, Montenegro MM, Dias AT, Dutra RL, Honjo RS, Bertola DR, Kulikowski LD, Kim CA.  
Mol Cytogenet. 2015 Jun 26;8:43.

Elevation of insulin-like growth factor binding protein-2 level in Pallister-Killian syndrome: implications for the postnatal growth retardation phenotype.

Izumi K, Kellogg E, Fujiki K, Kaur M, Tilton RK, Noon S, Wilkens A, Shirahige K, Krantz ID.  
Am J Med Genet A. 2015 Jun;167(6):1268-74.

[Prenatal diagnosis of a case of Pallister-Killian syndrome].

Xi H, Wang H, Jia Z, Zhou Y, Yu H, Liu J, Wu L.  
Zhonghua Yi Xue Yi Chuan Xue Za Zhi. 2015 Feb 10;32(1):73-6. Chinese.

Mosaicism and clinical genetics.

Spinner NB, Conlin LK.

Am J Med Genet C Semin Med Genet. 2014 Dec;166C(4):397-405.

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-Casanelles 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.

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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.

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

Kostanecka A, Close LB, Izumi K, Krantz ID, Pipan M.

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Pallister-Killian syndrome: historical perspective and foreword.

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

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, Sári Z.  
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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, Altiock Clark O, Cetin Z, Keser I, Berker S, Luleci G.  
Balkan J Med Genet. 2012 Jun;15(1):61-4.

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.

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.

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.

Congenital diaphragmatic hernia.  
Tovar JA.  
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## 2011

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.

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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.

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. No abstract available.

### 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.

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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.  
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### Iris transillumination defects associated with Pallister-Killian syndrome.

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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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J Child Neurol. 2010 Feb;25(2):238-45.

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.  
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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.  
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Am J Med Genet A. 2009 Mar;149A(3):505-9.

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Valproate-associated reversible encephalopathy in a 3-year-old girl with Pallister-Killian syndrome.  
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M Baglaj, J King, R Carachi  
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Fetal facial profile in Pallister-Killian syndrome.  
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E Guareschi, L Garavelli, S Pedori, V Di Lernia, L Grenzi, F Franchi, M Marinelli, GF Croci, E Pedretti, S Amarri,...  
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## 2006

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

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

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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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### DEVELOPMENT AND EDUCATION IN CHILDHOOD

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