Close

Web of Science
Page 1 (Records 1 -- 48)

[1]

Record 1 of 48

Title: Postzygotic telomere capture causes segmental UPD, duplication and deletion of chromosome 8p in a patient with intell

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Source: EUROPEAN JOURNAL OF MEDICAL GENETICS **Volume:** 60 **Issue:** 9 **Pages:** 445-450 **DOI:** 10.1016/j.ejmg.2

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Abstract: Using SNP array and FISH analysis, a patient with moderate intellectual disability and obesity was found to harbou 8p23.1, directly flanked by a distally located interstitial deletion of 2.3 Mb and a terminal segmental uniparental disomy. The c between the two segmental duplication regions.

These segmental duplications on chromosome 8p23.1 are known to be involved in chromosomal rearrangements because of m genomic regions. Genomic instability mediated by these segmental duplications is generally caused by non-allelic homologous reciprocal duplications, inversions and translocations.

Additional analysis of the parental origin of the fragments of this atypical inverted duplication/interstitial deletion shows pater chromosome 8. Combined with the finding that the normal chromosome 8 carries an inversion in 8p23.1 we hypothesize that a chromosome was postzygotically repaired with the paternal inverted copy resulting in a duplication, deletion and segmental ur of the 8p23.1 segmental duplication regions in recombination. (C) 2017 Elsevier Masson SAS. All rights reserved.

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Language: English

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Record 2 of 48

Title: Analysis of Invdupdel(8p) Rearrangement: Clinical, Cytogenetic and Molecular Characterization

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Source: AMERICAN JOURNAL OF MEDICAL GENETICS PART A **Volume:** 167 **Issue:** 5 **Pages:** 1018-1025 **DOI:** 10.1

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Total Times Cited: 5

Abstract: Inverted duplication 8p associated with deletion of the short arms of chromosome 8 (invdupdel[8p]) is a relatively u rearrangement, with an estimated incidence of 1 in 10,000-30,000 live borns. The chromosomal rearrangement consists of a de an inverted duplication of the 8p11.2-p22 region. Clinical manifestations of this disorder include severe to moderate intellectua most cases, there are also CNS associated malformations and congenital heart defects. In this work, we present the cytogenetic children with invdupdel(8p) rearrangements. Subsequently, we have carried out genotype-phenotype correlations in these seve similar deletion but different size of duplications; the latter probably explaining the phenotypic variability among them. We rec detailed chromosomal microarray studies should be undertaken, enabling appropriate genetic counseling. (c) 2015 Wiley Perio

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PubMed ID: 25712135

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Record 3 of 48

Title: Three cases of isolated terminal deletion of chromosome 8p without heart defects presenting with a mild phenotype

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Source: AMERICAN JOURNAL OF MEDICAL GENETICS PART A **Volume:** 161A **Issue:** 4 **Pages:** 822-828 **DOI:** 10.1
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Abstract: Individuals with isolated terminal deletions of 8p have been well described in the literature, however, molecular cha
deletion in most instances is lacking. The phenotype of such individuals falls primarily into two categories: those with cardiac
8p has been demonstrated to contain two inversely oriented segmental duplications at 8p23.1, flanking the gene, GATA4. Hapl
in cardiac defects seen in numerous individuals with terminal 8p deletion. Current microarray technologies allow for the precis
the deleted region. We present three individuals with isolated terminal deletion of 8p distal to the segmental duplication telome
relatively mild and nonspecific phenotype including mildly dysmorphic features, developmental delay, speech delay, and early
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Record 4 of 48

Title: Proteome Atlas of Human Chromosome 8 and Its Multiple 8p Deficiencies in Tumorigenesis of the Stomach, Colon, an

Author(s): Zhang, Y (Zhang, Yang); Yan, GQ (Yan, Guoquan); Zhai, LH (Zhai, Linhui); Xu, SH (Xu, Shaohang); Shen, HL (S Feifei); Xie, LQ (Xie, Liqi); Tang, HL (Tang, Hailin); Yu, HX (Yu, Hongxiu); Liu, MQ (Liu, Mingqi); Yang, PY (Yang, Pengy Chengpu); Li, LW (Li, Liwei); Chang, C (Chang, Cheng); Li, N (Li, Ning); Wu, SF (Wu, Songfeng); Zhu, YP (Zhu, Yunping); Bo); Lin, L (Lin, Liang); Wang, YZ (Wang, Yinzhu); Zheng, GY (Zheng, Guiyan); Zhou, LP (Zhou, Lanping); Lu, HJ (Lu, Ha Zhong, F (Zhong, Fan)

Source: JOURNAL OF PROTEOME RESEARCH **Volume:** 12 **Issue:** 1 **Pages:** 81-88 **DOI:** 10.1021/pr300834r **Publishe**

Times Cited in Web of Science Core Collection: 11

Total Times Cited: 13

Abstract: Chromosome 8, a medium-length euchromatic unit in humans that has an extraordinarily high mutation rate, can be multiple mutant diseases, such as tumorigenesis, and further invasion/metastasis. The Chromosome-Centric Human Proteome proteomes of three digestive organs (i.e., stomach, colon, and liver) and their corresponding carcinoma tissues/cell lines accor By rigorous standards, we have identified 271 (38.7%), 330 (47.1%), and 325 (4-6.4%) of 701 chromosome 8-coded proteins respectively, in Swiss-Prot and observed a total coverage rate of up to 58.9% by 413 identified proteins. Using large-scale labe some 8p deficiencies, such as the presence of 8p21-p23 in tumorigenesis of the above-described digestive organs, which is in best knowledge, this is the first study to have verified these 8p deficiencies at the proteome level, complementing genome and

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Shanghai Health Bureau	2010Y005
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Record 5 of 48

Title: Genotype-Phenotype Association Studies of Chromosome 8p Inverted Duplication Deletion Syndrome

Author(s): Fisch, GS (Fisch, Gene S.); Davis, R (Davis, Ryan); Youngblom, J (Youngblom, Janey); Gregg, J (Gregg, Jeff)

Source: BEHAVIOR GENETICS **Volume:** 41 **Issue:** 3 **Special Issue:** SI **Pages:** 373-380 **DOI:** 10.1007/s10519-011-9447

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Total Times Cited: 8

Abstract: Individuals diagnosed with chromosome 8p inverted duplication deletion (invdupdel(8p)) manifest a wide range of purpose of this study is to employ array CGH technology to define more precisely the cytogenetic breakpoints and regions of c individuals with invdupdel(8p), and compare these results with their neuropsychological characteristics. We examined the cogn female children, ages 3-15 years, with invdupdel(8p). We noted cognitive deficits that ranged from mild to severe, and adaptiv significantly to substantially lower than adequate levels. CARS scores, a measure of autistic behavior, identified three children the four children exhibited attention deficits and hyperactivity consistent with a DSM-IV-TR diagnosis of ADHD. One child sh intellectual disability was not correlated with deletion size, nor was the deletion location associated with the autistic phenotype 8p21.1/8p22 was associated with cognitive deficit. In addition, a small locus of over-expression in 8p21.3 was common for all limitation of the study is its small sample size. Further analyses of the deleted and over-expressed regions are needed to ascerta and, possibly, autism.

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Record 6 of 48

Title: Mild Phenotype in a Patient With Mosaic del(8p)/inv dup del(8p)

Author(s): Hand, M (Hand, Matthew); Gray, C (Gray, Carolyn); Glew, G (Glew, Gwen); Tsuchiya, KD (Tsuchiya, Karen D.)

Source: AMERICAN JOURNAL OF MEDICAL GENETICS PART A **Volume:** 152A **Issue:** 11 **Pages:** 2827-2831 **DOI:** 1

Times Cited in Web of Science Core Collection: 7

Total Times Cited: 8

Abstract: We report on a female with a mild phenotype who is mosaic for two cell lines with different structural abnormalities imbalances. Molecular cytogenetic and G-banded chromosome analyses demonstrated that one cell line has a large terminal 8p other cell line contains a derivative chromosome 8, known as an inv dup del(8p) in the literature. This female has development are associated with either 8p abnormality in non-mosaic form. The attenuated phenotype in this individual may be due to comp other cell line. (C) 2010 Wiley-Liss, Inc.

Accession Number: WOS:000284005700025

PubMed ID: 20830805

Language: English

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Record 7 of 48

Title: Genomic profile of copy number variants on the short arm of human chromosome 8

Author(s): Yu, SH (Yu, Shihui); Fiedler, S (Fiedler, Stephanie); Stegner, A (Stegner, Andrew); Graf, WD (Graf, William D.)

Source: EUROPEAN JOURNAL OF HUMAN GENETICS **Volume:** 18 **Issue:** 10 **Pages:** 1114-1120 **DOI:** 10.1038/ejhg.2
Times Cited in Web of Science Core Collection: 18
Total Times Cited: 19

Abstract: We evaluated 966 consecutive pediatric patients with various developmental disorders by high-resolution microarra and found 10 individuals with pathogenic copy number variants (CNVs) on the short arm of chromosome 8 (8p), representing Two patients with 8p terminal deletion associated with interstitial inverted duplication (inv dup del(8p)) had different mechani intermediate during meiosis. Three probands carried an identical similar to 5.0Mb interstitial duplication of chromosome 8p23 observed at nucleotide coordinates of similar to 10.45, 24.32-24.82, 32.19-32.77, and 38.94-39.72 Mb involving the formation CNVs with deletion-or duplication-specific start or stop coordinates on the 8p provide useful information for exploring the bas rearrangements in the human genome. European Journal of Human Genetics (2010) 18, 1114-1120; doi: 10.1038/ejhg.2010.66

Accession Number: WOS:000282089600008
PubMed ID: 20461109
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Record 8 of 48

Title: A case with de novo inv dup del(8p) associated with dextrocardia and corpus callosum agenesis

Author(s): Ergun, MA (Ergun, Mehmet A.); Kula, S (Kula, Serdar); Karaer, K (Karaer, Kadri); Percin, EF (Percin, E. Ferda)

Source: PEDIATRICS INTERNATIONAL **Volume:** 52 **Issue:** 5 **Pages:** 845-846 **DOI:** 10.1111/j.1442-200X.2010.03181.x
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Accession Number: WOS:000282375500031
PubMed ID: 20880309
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Record 9 of 48

Title: Telomere Capture as a Frequent Mechanism for Stabilization of the Terminal Chromosomal Deletion Associated with In

Author(s): Yu, S (Yu, S.); Graf, WD (Graf, W. D.)

Source: CYTOGENETIC AND GENOME RESEARCH **Volume:** 129 **Issue:** 4 **Pages:** 265-274 **DOI:** 10.1159/000315887
Times Cited in Web of Science Core Collection: 21
Total Times Cited: 23

Abstract: We report 4 interstitial inverted duplications with associated terminal deletions (inv dup del) involving the short arm chromosome 13 by microarray-based comparative genomic hybridization (aCGH) combined with chromosome banding (GTG (FISH) analyses. Formation of the intermediate dicentric chromosomes in 3 of them occurred through breakage-fusion-bridge mechanism) and in the fourth one it occurred through the mediation of the inverted low-copy repeats on chromosome 8p23.1. a third one was suspected to be associated with telomere capture for the healing of the terminal deletions. These findings indic frequently used for stabilizing the broken chromosome ends in this type of genomic rearrangements. In addition, the inv dup d dup del on chromosome 13 in humans, the inv dup del(5) represents the first observation of inv dup del(5p) with an associated remaining two inv dup del(8p) were also discussed. Copyright (C) 2010 S. Karger AG, Basel

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Language: English

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Record 10 of 48

Title: U-type exchange is the most frequent mechanism for inverted duplication with terminal deletion rearrangements

Author(s): Rowe, LR (Rowe, L. R.); Lee, JY (Lee, J-Y); Rector, L (Rector, L.); Kaminsky, EB (Kaminsky, E. B.); Brothman, L.); South, ST (South, S. T.)

Source: JOURNAL OF MEDICAL GENETICS **Volume:** 46 **Issue:** 10 **Pages:** 694-702 **DOI:** 10.1136/jmg.2008.065052 **P**

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Total Times Cited: 40

Abstract: Background: Chromosomal rearrangements resulting in an interstitial inverted duplication with concomitant termina of chromosome 8 in 1976. Since then, this type of alteration has been identified and characterised for most chromosome arms. explain the origin of this type of rearrangement. All three mechanisms involve formation of a dicentric chromosome that then produce a monocentric duplicated and deleted chromosome. However, the events leading to the formation of the dicentric chro one mechanism, either parent carries a paracentric inversion. This results in formation of a loop during meiotic pairing with a the second mechanism, inverted low copy repeats in the same chromosome arm allow partial folding of one homologue onto it inverted repeats. The third mechanism involves a pre-meiotic double-strand break with subsequent fusion, or U-type exchange mechanisms require a single copy region to exist between the duplicated and deleted regions on the derivative chromosome, ar rearrangement can be used to distinguish between these mechanisms.
Methods and results: Using G-banded chromosome analysis, fluorescence in situ hybridisation (FISH) and array comparative g new cases of inverted duplication with terminal deletion of 2q, 4p, 5p, 6q, 8p, 9p, 10q, 13q, 15q, 18p, 18q, and 22q.
Conclusions: These new cases, combined with previously described cases, demonstrate that U-type exchange is the most frequ be observed on most, or perhaps all, chromosome arms.

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Record 11 of 48

Title: Chromosome 8p23.1 Deletions as a Cause of Complex Congenital Heart Defects and Diaphragmatic Hernia

Author(s): Wat, MJ (Wat, Margaret J.); Shchelochkov, OA (Shchelochkov, Oleg A.); Holder, AM (Holder, Ashley M.); Brema Aditi); Bacino, C (Bacino, Carlos); Scaglia, F (Scaglia, Fernando); Zori, RT (Zori, Roberto T.); Cheung, SW (Cheung, Sau Wa (Kang, Sung-Hae Lee)

Source: AMERICAN JOURNAL OF MEDICAL GENETICS PART A **Volume:** 149A **Issue:** 8 **Pages:** 1661-1677 **DOI:** 1(

Times Cited in Web of Science Core Collection: 73

Total Times Cited: 78

Abstract: Recurrent interstitial deletion of a region of 8p23.1 flanked by the low copy repeats 8p-OR-REPD and 8p-OR-REPI that can include congenital heart malformations and congenital diaphragmatic hernia (CDH). Haploinsufficiency of GATA4 is development of these birth defects. We describe two individuals and a monozygotic twin pair discordant for anterior CDH all c caused by this recurrent interstitial deletion as demonstrated by array comparative genomic hybridization. To better define the with alterations of genes on 8p23.1, we review the spectrum of congenital heart and diaphragmatic defects that have been repo mutations and interstitial, terminal, and complex chromosomal rearrangements involving the 8p23.1 region. Our findings allov region on chromosome 8p23.1 and suggest that haploinsufficiency of other genes, in addition to GATA4, may play a role in th associated with 8p23.1 deletions. These findings also underscore the importance of conducting a careful cytogenetic/molecular and postnatal cases involving congenital defects of the heart and/or diaphragm. (C) 2009 Wiley-Liss, Inc.

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PubMed ID: 19606479

Language: English

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Record 12 of 48

Title: Inverted duplications deletions: underdiagnosed rearrangements??

Author(s): Zuffardi, O (Zuffardi, O.); Bonaglia, M (Bonaglia, M.); Ciccone, R (Ciccone, R.); Giorda, R (Giorda, R.)

Source: CLINICAL GENETICS **Volume:** 75 **Issue:** 6 **Pages:** 505-513 **DOI:** 10.1111/j.1399-0004.2009.01187.x **Published**

Times Cited in Web of Science Core Collection: 40

Total Times Cited: 42

Abstract: Zuffardi O, Bonaglia M, Ciccone R, Giorda R. Inverted duplications deletions: underdiagnosed rearrangements?? Molecular techniques led to the discovery that several chromosome rearrangements interpreted as terminal duplications were i terminal deletions. Inv dup del rearrangements originate through a symmetric dicentric chromosome that, after asymmetric bre chromosome. In recurrent inverted duplications the dicentric chromosome is formed at meiosis through non-allelic homologou cases, dicentric intermediates are formed by non-homologous end joining or intrastrand annealing. Some authors hypothesized formed directly in the zygote. Healing of the broken dicentric chromosomes can occur not only in a telomerase-dependent way circularization thus creating translocated or ring inv dup del chromosomes. In all the cases reported up to now, the duplicated r but we can safely assume that there is another group of rearrangements where the deleted region is longer than the duplicated p cytogeneticist will suspect the presence of a deletion and confirm it by FISH with a subtelomeric probe, but he/she will almost

the conventional analysis techniques used until now have led to a substantial underestimate of the frequency of inv dup del rea array-CGH in routine analysis will allow a more realistic estimate. Obviously, the concomitant presence of deletion and duplic genotype/phenotype correlations.

Accession Number: WOS:000266488100001

PubMed ID: 19508415

Language: English

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Record 13 of 48

Title: Chromosome 8p as a potential hub for developmental neuropsychiatric disorders: implications for schizophrenia, autism

Author(s): Tabares-Seisdedos, R (Tabares-Seisdedos, R.); Rubenstein, JLR (Rubenstein, J. L. R.)

Source: MOLECULAR PSYCHIATRY **Volume:** 14 **Issue:** 6 **Pages:** 563-589 **DOI:** 10.1038/mp.2009.2 **Published:** JUN 2

Times Cited in Web of Science Core Collection: 113

Total Times Cited: 119

Abstract: Defects in genetic and developmental processes are thought to contribute susceptibility to autism and schizophrenia identifying susceptibility genes and abnormalities in the development has been difficult. However, the importance of genes wit neuropsychiatric disorders and cancer is well established. There are 484 annotated genes located on 8p; many are most likely c Molecular genetics and developmental studies have identified 21 genes in this region (ADRA1A, ARHGEF10, CHRNA2, CH FGF17, FGF20, FGFR1, FZD3, LDL, NAT2, NEF3, NRG1, PCM1, PLAT, PPP3CC, SFRP1 and VMAT1/SLC18A1) that are disorders (schizophrenia, autism, bipolar disorder and depression), neurodegenerative disorders (Parkinson's and Alzheimer's c nonprotein-coding RNAs (microRNAs) are located at 8p. Structural variants on 8p, such as copy number variants, microdeleti to autism, schizophrenia and other human diseases including cancer. In this review, we consider the current state of evidence fi expression and endophenotyping studies for the role of these 8p genes in neuropsychiatric disease. We also describe how a mu with deficits in specific components of social behavior and a reduction in its dorsomedial prefrontal cortex. We finish by discu respect to neuropsychiatric disorders and cancer, despite the shortcomings of this evidence. Molecular Psychiatry (2009) 14, 5 online 10 February 2009

Accession Number: WOS:000266236300004

PubMed ID: 19204725

Language: English

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Record 14 of 48

Title: Transmitted duplication of 8p23.1-8p23.2 associated with speech delay, autism and learning difficulties

Author(s): Glancy, M (Glancy, Mary); Barnicoat, A (Barnicoat, Angela); Vijeratnam, R (Vijeratnam, Rajan); de Souza, S (de Huang, SW (Huang, Shuwen); Maloney, VK (Maloney, Viv K.); Thomas, NS (Thomas, N. Simon); Bunyan, DJ (Bunyan, Dav (Barber, John C. K.)

Source: EUROPEAN JOURNAL OF HUMAN GENETICS **Volume:** 17 **Issue:** 1 **Pages:** 37-43 **DOI:** 10.1038/ejhg.2008.1

Times Cited in Web of Science Core Collection: 34

Total Times Cited: 35

Abstract: Duplications of distal 8p with and without significant clinical phenotypes have been reported and are often associat complexity. Here, we present a duplication of 8p23.1-8p23.2 ascertained in a child with speech delay and a diagnosis of ICD-1 his mother who had epilepsy and learning problems. A combination of cytogenetic, FISH, microsatellite, MLPA and oaCGH a extended over a minimum of 6.8Mb between 3 539 893 and 10 323 426 bp. This interval contains 32 novel and 41 known gen plausible candidate gene for autism at present. The distal breakpoint of the duplicated region interrupts the CSMD1 gene in 8p MSRA and RP1L1 genes in 8p23.1.

An interchromosomal insertion between a normal and polymorphically inverted chromosome 8 is proposed to explain the orig imbalances of distal 8p are needed to determine whether the autistic component of the phenotype in this family results from th dosage imbalance of an individual susceptibility gene.

Accession Number: WOS:000261588800006

PubMed ID: 18716609

Language: English

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Welcome Trust	

We are grateful to the Sanger Centre for providing the Ensembl tiling path clones and to Dr Hiroaki Shizuya for kinc are supported as part of the National Genetics Reference Laboratory (Wessex) by the UK Department of Health Ge Conception Unit. The image enhancement equipment used for this work was provided by the Welcome Trust and T Trust

Record 15 of 48

Title: Unusual 8p inverted duplication deletion with telomere capture from 8q

Author(s): Buysse, K (Buysse, Karen); Antonacci, F (Antonacci, Francesca); Callewaert, B (Callewaert, Bert); Loeys, B (Loe (Siu, Victoria); Mortier, G (Mortier, Geert); Speleman, F (Speleman, Frank); Menten, B (Menten, Bjoern)

Source: EUROPEAN JOURNAL OF MEDICAL GENETICS **Volume:** 52 **Issue:** 1 **Pages:** 31-36 **DOI:** 10.1016/j.ejmg.200

Times Cited in Web of Science Core Collection: 18

Total Times Cited: 19

Abstract: Inverted 8p duplication deletions are recurrent chromosomal rearrangements that are mediated through non-allelic h olfactory receptor (OR) gene clusters at 8p23.1. These rearrangements result in a proximal inverted duplication of various exte clusters and a terminal 8p deletion. The terminal deletions are stabilized by direct addition of telomeric repeats, so called telom unusual inverted duplication deletion of 8p. Stabilization of the broken chromosome end was achieved by telomere capture ins additional duplication of 8q24.13 -> qter on the short arm of chromosome 8. Moreover, the inverted duplication was only 3.4 M cytogenetically undetectable. To the best of our knowledge this is the smallest inverted duplication reported hitherto. We descr array CGH of this unusual inv dup del (8p) and a previously reported patient with a similar 8q duplication and review the litera

Accession Number: WOS:000263088400007

PubMed ID: 19041960

Language: English

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Record 16 of 48

Title: FLUORESCENCE IN SITU HYBRIDIZATION AND SINGLE NUCLEOTIDE POLYMORPHISM OF A NEW CASE

Author(s): Caglayan, AO (Caglayan, A. O.); Engelen, JJM (Engelen, J. J. M.); Ghesquiere, S (Ghesquiere, S.); Alofs, M (Alo (Dunbar, M.)

Source: GENETIC COUNSELING **Volume:** 20 **Issue:** 4 **Pages:** 333-340 **Published:** 2009

Times Cited in Web of Science Core Collection: 2

Total Times Cited: 2

Abstract: Fluorescence h? Situ Hybridization and single, nucleotide polymorphism of a new case with inv dup del(8p): Invert is a complex chromosome rearrangement leading among others to deletion of the chromosome region distal to the duplication deletion of 8p and the results of SNP-array analysis and fluorescence in situ hybridization (FISH) are reported here. Our result dup del(8p) cases.

Accession Number: WOS:000273888600005

PubMed ID: 20162868

Language: English

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Record 17 of 48

Title: Clinically abnormal case with paternally derived partial trisomy 8p23.3 to 8p12 including maternal isodisomy of 8p23.3

Author(s): Aktas, D (Aktas, Dilek); Weise, A (Weise, Anja); Utine, E (Utine, Eda); Alehan, D (Alehan, Dursun); Mrasek, K (Eggeling, Ferdinand); Thieme, H (Thieme, Heike); Tuncbilek, E (Tuncbilek, Ergul); Liehr, T (Liehr, Thomas)

Source: MOLECULAR CYTOGENETICS **Volume:** 2 **Article Number:** 14 **DOI:** 10.1186/1755-8166-2-14 **Published:** 20(

Times Cited in Web of Science Core Collection: 2

Total Times Cited: 2

Abstract: Background: Because of low copy repeats (LCRs) and common inversion polymorphisms, the human chromosome rearrangements. Each of these rearrangements is associated with several phenotypic features. We report on a patient with vario delay in connection with an inverted duplication event, involving chromosome 8p.
Methods: Chromosome analysis, multicolor banding analysis (MCB), extensive fluorescence in situ hybridization (FISH) anal performed.
Results: The karyotype was characterized in detail by multicolor banding (MCB), subtelomeric and centromere-near probes as p23.3->qter). Additionally, microsatellite analysis revealed the paternal origin of the duplication and gave hints for a mitotic re
Conclusion: A comprehensive analysis of the derivative chromosome 8 suggested a previously unreported mechanism of form aberration leading to maternal isodisomy, followed by an inverted duplication of the 8p12p23.3 region.

Accession Number: WOS:000208460900013

PubMed ID: 19566937

Language: English

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Record 18 of 48

Title: High precision size measurement of centromere 8 and the 8q24/c-myc gene region in metaphase and interphase human f

Author(s): Batram, C (Batram, C.); Baddeley, D (Baddeley, D.); Kreth, G (Kreth, G.); Cremer, C (Cremer, C.)

Source: JOURNAL OF STRUCTURAL BIOLOGY **Volume:** 164 **Issue:** 3 **Pages:** 293-303 **DOI:** 10.1016/j.jsb.2008.09.00

Times Cited in Web of Science Core Collection: 4

Total Times Cited: 5

Abstract: The hypothesis that distinct chromatin domains expand and are remodelled differently when they undergo transcript accepted. The condensation changes by which chromosomes are transformed at the metaphase-interphase transition are especia studied by light microscopy; however, quantitative information of the size on specific small chromatin domains during the cell problem is the determination of structural features close to the resolution limit. In this report we use a novel approach to quant domain and the centromeric region of chromosome 8 in doubly labelled normal human foreskin fibroblasts using confocal lase were analysed in both metaphase spreads and interphase nuclei. These high precision measurements revealed a somewhat sma centromere region in metaphase compared to interphase. Surprisingly, within the same cells the lateral extension of the 8q24/c interphase than in metaphase. For comparison the centromere size was more condensed in metaphase than in interphase. This i specific chromatin domains with opposite condensation behaviour. (C) 2008 Elsevier Inc. All rights reserved.

Accession Number: WOS:000264521200007

PubMed ID: 18835450

Language: English

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Record 19 of 48

Title: Defensins and the dynamic genome: What we can learn from structural variation at human chromosome band 8p23.1

Author(s): Hollox, EJ (Hollox, Edward J.); Barber, JCK (Barber, John C. K.); Brookes, AJ (Brookes, Anthony J.); Armour, JA

Source: GENOME RESEARCH **Volume:** 18 **Issue:** 11 **Pages:** 1686-1697 **DOI:** 10.1101/gr.080945.108 **Published:** NOV

Times Cited in Web of Science Core Collection: 60

Total Times Cited: 63

Abstract: Over the past four years, genome-wide studies have uncovered numerous examples of structural variation in the hur that changes copy number, such as deletion and duplication, and structural variation that does not change copy number, such as region that contains all these types of variation spans the chromosome band 8p23.1. This region has been studied in some dept our current understanding of the variation of this region. We also consider whether this region is a good model for other structu the implications of this variation are for clinical studies. Finally, we discuss the bioinformatics challenges raised, discuss the ev priorities for structural variation research.

Accession Number: WOS:000260536100002

PubMed ID: 18974263

Language: English

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Record 20 of 48

Title: A novel locus for generalized epilepsy with febrile seizures plus in French families

Author(s): Baulac, S (Baulac, Stephanie); Gourfinkel-An, I (Gourfinkel-An, Isabelle); Couarch, P (Couarch, Philippe); Depie
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Source: ARCHIVES OF NEUROLOGY **Volume:** 65 **Issue:** 7 **Pages:** 943-951 **DOI:** 10.1001/archneur.65.7.943 **Publishe**
Times Cited in Web of Science Core Collection: 23
Total Times Cited: 24

Abstract: Background: Generalized epilepsy with febrile seizures plus (GEFS(+)) is a familial autosomal dominant entity cha
afebrile seizures. Mutations in 3 genes - the sodium channel alpha 1 subunit gene (SCN1A), the sodium channel beta 1 subuni
receptor subunit gene (GABRG2) - and linkage to 2 other loci on 2p24 and 21q22 have been identified in families with GEFS
Objectives: To localize by means of linkage analysis a new gene for GEFS(+) in a large family with 11 affected members and
with GEFS(+).
Design: Family- based linkage analysis.
Setting: University hospital.
Patients: Five French families with GEFS(+) and at least 7 available affectedmemberswith autosomaldominanttransmission. A
afebrile generalized tonic-clonic seizures or absence epilepsy.
Main Outcome Measures: We analyzed 380 microsatellite markers and conducted linkage analysis.
Results: In the largest family, a 10-cM-density genomewide scan revealed linkage to a 13-Mb (megabase) interval on chromos
logarithm of odds (LOD) score of 3.00 (at Theta= 0) for markers D8S351 and D8S550 and a multipoint LOD score of 3.23. A
possibly linked to chromosome 8p23-p21 and the region was narrowed to a 7.3- Mb candidate interval, flanked by markers D8
identified mutations in the coding exons of 6 candidate genes (MTMR9, MTMR7, CTSB, SGCZ, SG223, and ATP6V1B2) lo
Conclusions: Wereport a sixth locus for GEFS(+) on chromosome 8p23- p21. Because no ion channel genes are located in this
will probably uncover a new mechanism of pathogenesis for GEFS(+).

Accession Number: WOS:000257596100012
PubMed ID: 18625863
Language: English

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Record 21 of 48

Title: Two patients with atypical interstitial deletions of 8p23.1: Mapping of phenotypical traits

Author(s): Paez, MT (Paez, Marco T.); Yamamoto, T (Yamamoto, Toshiyuki); Hayashi, KI (Hayashi, Ken-ichi); Yasuda, T (Y
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Source: AMERICAN JOURNAL OF MEDICAL GENETICS PART A **Volume:** 146A **Issue:** 9 **Pages:** 1158-1165 **DOI:** 10
2008
Times Cited in Web of Science Core Collection: 23
Total Times Cited: 24

Abstract: Chromosomal 8p23 deletion syndrome is recognized as a malformation syndrome with clinical symptoms of facial a and congenital heart defects. The responsible gene for the heart defects in this syndrome has been identified as GATA4 on 8p2 8p23.1 were investigated; one patient showed moderate developmental delay and Ebstein anomaly, and the other showed mild defect. The precise deletion sizes, 17 and 2.9 Mb, were determined by FISH analyses using BAC clones as probes. The latter d GATA4 in the previously reported patients, and the critical regions and genes for clinical manifestation of 8p23 deletion syndr behavioral abnormality, and developmental delay, were discussed. (C) 2008 Wiley-Liss, Inc.

Accession Number: WOS:000255422900007

PubMed ID: 18393291

Language: English

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ISSN: 1552-4825

Record 22 of 48

Title: Molecular cytogenetic characterization of a unique and complex de novo 8p rearrangement

Author(s): Cooke, SL (Cooke, Susanna L.); Northup, JK (Northup, Jill K.); Champaige, NL (Champaige, Neena L.); Zinser, V Paul A. W.); Lockhart, LH (Lockhart, Lillian H.); Velagaleti, GVN (Velagaleti, Gopatrao V. N.)

Source: AMERICAN JOURNAL OF MEDICAL GENETICS PART A **Volume:** 146A **Issue:** 9 **Pages:** 1166-1172 **DOI:** 10 2008

Times Cited in Web of Science Core Collection: 10

Total Times Cited: 10

Abstract: Human chromosome 8p is prone to recurrent rearrangements with inv dup del(8p) being most common. Each of the different clinical manifestations. Some of these recurrent rearrangements at 8p are mediated by an 8p submicroscopic paracent clusters present in one of the parents. However, recent reports have shown that some of the rearrangements are unique and con elements within Sp. Here, we report on a unique and complex 8p rearrangement with seizures as the major presenting feature i hybridization and microarray analyses with tiling path 8p array showed that the rearrangement is unique in that the 8p duplicat the more common inv dup del(8p), is not derived from parental submicroscopic inversion. Also unlike the inv dup del(8p), the nervous system malformations or cardiac defects. (C) 2008 Wiley-Liss, Inc.

Accession Number: WOS:000255422900008

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Language: English

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Record 23 of 48

Title: Two classes of low-copy repeats comediate a new recurrent rearrangement consisting of duplication at 8p23.1 and triplic

Author(s): Giorda, R (Giorda, Roberto); Ciccone, R (Ciccone, Roberto); Gimelli, G (Gimelli, Giorgio); Pramparo, T (Prampa MC (Bonaglia, Maria Clara); Giglio, S (Giglio, Sabrina); Genuardi, M (Genuardi, Maurizio); Argente, J (Argente, Jesus); Roc (Zuffardi, Orsetta)

Source: HUMAN MUTATION **Volume:** 28 **Issue:** 5 **Pages:** 459-468 **DOI:** 10.1002/humu.20465 **Published:** MAY 2007

Times Cited in Web of Science Core Collection: 33

Total Times Cited: 34

Abstract: We describe a new type of rearrangement consisting of the duplication of 8p23.1 and the triplication of 8p23.2 [dup retardation and minor facial dysmorphisms. Array-comparative genomic hybridization (CGH), fluorescence in situ hybridizati allowed us to demonstrate that this rearrangement is mediated by the combined effects of two unrelated low-copy repeats (LCI clusters of olfactory receptor genes (OR-REPs) lying at 8p23.1. The second type of LCRs consists of a 15-kb segmental duplic and enclosing a nonrepeated sequence of approximately 130kb, named MYOM2-REP because of its proximity to the MYOM2 third case with a dicentric chromosome 8 demonstrated that the rearrangement had been generated by nonallelic homologous r Based on our findings, we propose a model showing that a second recombination event at the level of the OR-REPs leads to th This rearrangement can only arise during meiosis in heterozygous carriers of the polymorphic 8p23.1 inversion, whereas in sul homozygous for the inversion only the dicentric chromosome can be formed. Our study demonstrates that nonallelic homologo generate more complex rearrangements and cause a greater variety of genomic diseases.

Accession Number: WOS:000245788700006

PubMed ID: 17262805

Language: English

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ISSN: 1059-7794

Funding:

Funding Agency	Grant Number
Telethon	GGP05177

Record 24 of 48

Title: DNA sequence and analysis of human chromosome 8

Author(s): Nusbaum, C (Nusbaum, C); Mikkelsen, TS (Mikkelsen, TS); Zody, MC (Zody, MC); Asakawa, S (Asakawa, S); T Kodira, CD (Kodira, CD); Schueler, MG (Schueler, MG); Shimizu, A (Shimizu, A); Whittaker, CA (Whittaker, CA); Chang, J Dewar, K (Dewar, K); FitzGerald, MG (FitzGerald, MG); Yang, XP (Yang, XP); Allen, NR (Allen, NR); Anderson, S (Anders Blechschmidt, K (Blechschmidt, K); Bloom, T (Bloom, T); Borowsky, ML (Borowsky, ML); Butler, J (Butler, J); Cook, A (Co

(DeArellano, K); DeCaprio, D (DeCaprio, D); Dooley, KT (Dooley, KT); Dorris, L (Dorris, L); Engels, R (Engels, R); Glockn Hagopian, DS (Hagopian, DS); Hall, JL (Hall, JL); Ishikawa, SK (Ishikawa, SK); Jaffe, DB (Jaffe, DB); Kamat, A (Kamat, A) R); Lokitsang, T (Lokitsang, T); Macdonald, P (Macdonald, P); Major, JE (Major, JE); Matthews, CD (Matthews, CD); Mauce Mihalev, AH (Mihalev, AH); Minoshima, S (Minoshima, S); Murayama, Y (Murayama, Y); Naylor, JW (Naylor, JW); Nicol, J SB (O'Leary, SB); O'Neill, K (O'Neill, K); Parker, SCJ (Parker, SCJ); Polley, A (Polley, A); Raymond, CK (Raymond, CK); R (Rodriguez, J); Sasaki, T (Sasaki, T); Schilhabel, M (Schilhabel, M); Siddiqui, R (Siddiqui, R); Smith, CL (Smith, CL); Snedd JA); Tenzin, P (Tenzin, P); Topham, K (Topham, K); Venkataraman, V (Venkataraman, V); Wen, GP (Wen, GP); Yamazaki, S QD (Zeng, QD); Zimmer, AR (Zimmer, AR); Rosenthal, A (Rosenthal, A); Birren, BW (Birren, BW); Platzer, M (Platzer, M); ES)

Source: NATURE **Volume:** 439 **Issue:** 7074 **Pages:** 331-335 **DOI:** 10.1038/nature04406 **Published:** JAN 19 2006

Times Cited in Web of Science Core Collection: 64

Total Times Cited: 460

Abstract: The International Human Genome Sequencing Consortium (IHGSC) recently completed a sequence of the human g focused on chromosome 8. Although some chromosomes exhibit extreme characteristics in terms of length, gene content, repe chromosome 8 is distinctly typical in character, being very close to the genome median in each of these aspects. This work des for the chromosome, which represents just over 5% of the euchromatic human genome. A unique feature of the chromosome is distal 8p that appears to have a strikingly high mutation rate, which has accelerated in the hominids relative to other sequenced a number of genes related to innate immunity and the nervous system, including loci that appear to be under positive selection(gene cluster(3,4) and MCPH1(5,6), a gene that may have contributed to the evolution of expanded brain size in the great apes. better understanding of both normal and disease biology and genome evolution.

Accession Number: WOS:000234682100044

PubMed ID: 16421571

Language: English

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ISSN: 0028-0836

Funding:

Funding Agency	Grant Number
Medical Research Council	G0000107

Record 25 of 48

Title: Molecular characterization of del(8)(p23.1p23.1) in a case of congenital diaphragmatic hernia

Author(s): Shimokawa, O (Shimokawa, O); Miyake, N (Miyake, N); Yoshimura, T (Yoshimura, T); Sosonkina, N (Sosonkina (Mizuguchi, T); Kondoh, S (Kondoh, S); Kishino, T (Kishino, T); Ohta, T (Ohta, T); Remco, V (Remco, V); Takashima, T (Ta Yoshiura, K (Yoshiura, K); Niikawa, N (Niikawa, N); Matsumoto, N (Matsumoto, N)

Source: AMERICAN JOURNAL OF MEDICAL GENETICS PART A **Volume:** 136A **Issue:** 1 **Pages:** 49-51 **DOI:** 10.100

Times Cited in Web of Science Core Collection: 47

Total Times Cited: 49

Abstract: A 36-week-old fetus was referred to the medical center because of his cystic mass and fluid in left thoracic cavity, a manage neonatal problems at 37 weeks of gestation. Emergent surgical repair of the left diaphragmatic hernia was performed, the following day. Chromosome analysis of cultured amniotic fluid cells indicated 46,XY,del(8)(p23.1p23.1). This is the fourth diaphragmatic hernia. Microarray comparative genomic hybridization analysis using DNA of cultured amniotic fluid cells sho mapped to the region between two low copy repeats (LCRs) at 8p23.1 previously described. Microsatellite analysis revealed th parents did not carry 8p23.1 polymorphic inversion. These data strongly suggested that the 8p23.1 interstitial deletion should h that of inv dup del(8p) whose structural abnormality is always of maternal origin and accompanies heterozygous 8p23.1 polym Liss, Inc.

Accession Number: WOS:000230213200009

PubMed ID: 15937941

Language: English

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Record 26 of 48

Title: Array based CGH and FISH fail to confirm duplication of 8p22-p23.1 in association with Kabuki syndrome

Author(s): Hoffman, JD (Hoffman, JD); Zhang, Y (Zhang, Y); Greshock, J (Greshock, J); Ciprero, KL (Ciprero, KL); Emanu EH); Weber, BL (Weber, BL); Ming, JE (Ming, JE)

Source: JOURNAL OF MEDICAL GENETICS **Volume:** 42 **Issue:** 1 **Pages:** 49-53 **DOI:** 10.1136/jmg.2004.024372 **Publi**

Times Cited in Web of Science Core Collection: 21

Total Times Cited: 21

Abstract: Background: Kabuki (Niikawa - Kuroki) syndrome comprises a characteristic facial appearance, cleft palate, conge Various cytogenetically visible chromosomal rearrangements have been reported in single cases, but the molecular genetic bas recent report described a duplication of 8p22 - p23.1 in 13/ 13 patients.
Objective: To determine the frequency of an 8p duplication in a cohort of patients with Kabuki syndrome.
Methods: An 8p duplication was sought using two independent methods - array based comparative genomic hybridisation (aC FISH) in 15 patients with a definitive clinical diagnosis of Kabuki syndrome.
Results: No evidence for a duplication of 8p was obtained by FISH or aCGH in any of the 15 patients.
Conclusions: 8p22 - p23.1 duplication may not be a common mechanism for Kabuki syndrome. Another genetic abnormality r patients.

Accession Number: WOS:000226510000008

PubMed ID: 15635075

Language: English

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Funding Agency	Grant Number
NICHD NIH HHS	HD26979

Record 27 of 48

Title: Polymorphic segmental duplications at 8p23.1 challenge the determination of individual defensin gene repertoires and tl sequence

Author(s): Taudien, S (Taudien, S); Galgoczy, P (Galgoczy, P); Huse, K (Huse, K); Reichwald, K (Reichwald, K); Schilhabel, K); Shimizu, A (Shimizu, A); Asakawa, S (Asakawa, S); Frankish, A (Frankish, A); Loncarevic, IF (Loncarevic, IF); Shimizu, Platzer, M (Platzer, M)

Source: BMC GENOMICS **Volume:** 5 **Article Number:** 92 **DOI:** 10.1186/1471-2164-5-92 **Published:** DEC 10 2004

Times Cited in Web of Science Core Collection: 41

Total Times Cited: 42

Abstract: Background: Defensins are important components of innate immunity to combat bacterial and viral infections, and (defensin (DEF) genes are located in a 2 Mb range of the human chromosome 8p23.1. This DEF locus, however, represents one final human genome sequence which contains segmental duplications, and recalcitrant gaps indicating high structural dynamic Results: We find that inter- and intraindividual genetic variations within this locus prevent a correct automatic assembly of the which currently even contains misassemblies. Manual clone-by-clone alignment and gene annotation as well as repeat and SNI alignment significantly improving the DEF locus representation. Our assembly better reflects the experimentally verified varia numbers. It contains an additional DEF cluster which we propose to reside between two already known clusters. Furthermore, and several pseudogenes expanding the hitherto known DEF repertoire. Analyses of BAC and working draft sequences of the (complex as in humans and DEF genes and a cluster are multiplied. Comparative analysis of human and chimpanzee DEF gene structure. Whether this might contribute to differences in disease susceptibility between man and ape remains to be solved. For repertoires we provide a molecular approach based on DEF haplotypes.
Conclusions: Complexity and variability seem to be essential genomic features of the human DEF locus at 8p23.1 and provide representation in the human reference sequence. Dissection of paralogous sequence variations, duplicon SNPs ans multisite va based methods is the way for future studies of interindividual DEF locus variability and its disease association.

Accession Number: WOS:000226607400001

PubMed ID: 15588320

Language: English

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ISSN: 1471-2164

Record 28 of 48

Title: Molecular characterization of inv dup del(8p): Analysis of five cases

Author(s): Shimokawa, O (Shimokawa, O); Kurosawa, K (Kurosawa, K); Ida, T (Ida, T); Harada, N (Harada, N); Kondoh, T Yoshiura, K (Yoshiura, K); Kishino, T (Kishino, T); Ohta, T (Ohta, T); Niikawa, N (Niikawa, N); Matsumoto, N (Matsumoto,

Source: AMERICAN JOURNAL OF MEDICAL GENETICS PART A **Volume:** 128A **Issue:** 2 **Pages:** 133-137 **DOI:** 10.1

Times Cited in Web of Science Core Collection: 46

Total Times Cited: 48

Abstract: We analyzed five patients with inverted duplication deletion of 8p [inv dup del(8p)] using fluorescence in situ hybri polymorphism (STRP) analysis. In all patients, inv dup del(8p) consisted of a deleted distal segment, an intact in-between segr of them, the proximal breakpoint of the deletion and one of the breakpoints of the duplication were identical, each located at or 8p23. FISH analysis showed all their mothers to be heterozygous carriers of an 8p23 inversion [inv(8)(p23)]. STRP analysis in maternally derived chromosomes. The duplicated segments had two copies of maternal, either heterozygous or homozygous al those in 16 patients with inv dup del(8p) and their parents by Floridia et al. [1996: Am J Hum Genet 58:785-796] and subsequ al. [2001: Am J Hum Genet 68:874-883]. Based on these findings, we propose a model for the inv dup del(8p) formation. The

inv(8)(p23) heterozygous carrier mothers form a loop at the pachytene period of meiosis I. Inv dup del(8p) with heterozygous meiotic recombination within the loop. Inv dup del(8p) with the homozygous duplication arises through two meiotic recombin within the loop and the other between the loop and centromere). Subsequent rescue by eliminating a part of the duplicated segı viable inv dup del(8p). The frequency of the inv(8)(p23) allele is 39% in a normal Japanese population, comparable to 26% in Genet 68:874-883]. The proposed mechanism of formation of inv dup del(8p) requires two independent events (a recombinatic which may explain its rarity. (C) 2004 Wiley-Liss, Inc.

Accession Number: WOS:000222567900007

PubMed ID: 15214003

Language: English

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ISSN: 0148-7299

Record 29 of 48

Title: Mosaicism del(8p)/inv dup(8p) in a dysmorphic female infant: a mosaic formed by a meiotic error at the 8p OR gene an

Author(s): Vermeesch, JR (Vermeesch, JR); Thoelen, R (Thoelen, R); Salden, I (Salden, I); Raes, M (Raes, M); Matthijs, G (M

Source: JOURNAL OF MEDICAL GENETICS **Volume:** 40 **Issue:** 8 **Article Number:** e93 **DOI:** 10.1136/jmg.40.8.e93 **P**

Times Cited in Web of Science Core Collection: 35

Total Times Cited: 35

Accession Number: WOS:000184761400019

PubMed ID: 12920085

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ISSN: 0022-2593

Record 30 of 48

Title: Fetoplacental discrepancy involving structural abnormalities of chromosome 8 detected by prenatal diagnosis

Author(s): Soler, A (Soler, A); Sanchez, A (Sanchez, A); Carrio, A (Carrio, A); Badenas, C (Badenas, C); Mila, M (Mila, M);

Source: PRENATAL DIAGNOSIS **Volume:** 23 **Issue:** 4 **Pages:** 319-322 **DOI:** 10.1002/pd.590 **Published:** APR 2003

Times Cited in Web of Science Core Collection: 13

Total Times Cited: 13

Abstract: We describe the finding of three cell lines involving different structural abnormalities of chromosome 8 detected in (CVS) was performed on a pregnant woman because of advanced maternal age. Semidirect cytogenetic analysis showed a mos confirmed by fluorescence in situ hybridization (FISH). Amniocentesis was subsequently performed, and the karyotype obtain pregnancy was terminated; pathologic findings included clubfeet, clenched left hand, subcutaneous edema and bilateral hydroc 8 microsatellites performed on parents' blood and fetal tissues revealed a maternal meiotic origin of the inv dup(8p) with deleti the remaining 8p. We propose a model to explain the cytogenetic findings, which includes a first maternal meiotic error giving present in the ovum, a second error in one of the first zygote divisions with misdivision of the dicentric 8 giving rise to a cell li another cell line with inv dup(8p) confined to the fetal tissue and a third error in the trophoblast giving rise to a further cell line John Wiley Sons, Ltd.

Accession Number: WOS:000182331500011

PubMed ID: 12673638

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ISSN: 0197-3851

Record 31 of 48
Title: The application of region-specific probes for the resolution of duplication 8p: a case report and a review of the literature
Author(s): Pabst, B (Pabst, B); Arslan-Kirchner, M (Arslan-Kirchner, M); Schmidtke, J (Schmidtke, J); Miller, K (Miller, K)
Source: CYTOGENETIC AND GENOME RESEARCH **Volume:** 103 **Issue:** 1-2 **Pages:** 3-7 **DOI:** 10.1159/000076280 **Pu**
Times Cited in Web of Science Core Collection: 5
Total Times Cited: 7
Abstract: The structural rearrangement in the short arm of a chromosome 8 in a clinically affected patient has been reinvestig painting and region specific YAC probes. An inverted duplication of the segment p22 --> p11.2 and a deletion of the subtelom approach, a more detailed resolution of the duplication/deletion 8p was possible. With the application of molecular cytogenetic segments within the clinical entity of duplication/deficiency 8p can be shown. Copyright (C) 2003 S. Karger AG, Basel.
Accession Number: WOS:000220538600002
PubMed ID: 15004455
Language: English
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Record 32 of 48
Title: 8p23 duplication reconsidered: is it a true euchromatic variant with no clinical manifestation?
Author(s): Tsai, CH (Tsai, CH); Graw, SL (Graw, SL); McGavran, L (McGavran, L)
Source: JOURNAL OF MEDICAL GENETICS **Volume:** 39 **Issue:** 10 **Pages:** 769-774 **DOI:** 10.1136/jmg.39.10.769 **Publ**
Times Cited in Web of Science Core Collection: 19
Total Times Cited: 21
Accession Number: WOS:000178538700013
PubMed ID: 12362038
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Record 33 of 48
Title: Direct duplication of 8p21.3 -> p23.1: A cytogenetic anomaly associated with developmental delay without consistent c
Author(s): Fan, YS (Fan, YS); Siu, VM (Siu, VM); Jung, JH (Jung, JH); Farrell, SA (Farrell, SA); Cote, GB (Cote, GB)
Source: AMERICAN JOURNAL OF MEDICAL GENETICS **Volume:** 103 **Issue:** 3 **Pages:** 231-234 **DOI:** 10.1002/ajmg.1
Times Cited in Web of Science Core Collection: 11
Total Times Cited: 12

Abstract: We report six cases in two families and a sporadic case with a direct duplication of region Sp21.3 --> 23.1. In one fa
was transmitted to one son and one daughter. In the second family, the father was mosaic for the anomaly that was transmitted
was initially described as an 8p+ with banding analysis and then delineated with fluorescence in situ hybridization (FISH) usir
painting, and 8p or 8p/8q subtelomeric probes. Deletion was not detected in the subtelomeric region of the abnormal chromosc
sporadic case. The phenotypic picture varies from normal to moderate mental retardation in the affected individuals. No consis
were observed among these cases. After comparing the chromosome region involved in our cases with those in others having c
thought that the segment 8p21.1 --> 21.3 might be the critical region for an 8p duplication syndrome. The parental origin of th
clinical significance. (C) 2001 Wiley-Liss, Inc.

Accession Number: WOS:000171277000009

PubMed ID: 11745996

Language: English

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ISSN: 0148-7299

Record 34 of 48

Title: Molecular cytogenetic characterization of a derivative chromosome 8 with an inverted duplication of 8p21.3 -> p23.3 ar

Author(s): Fan, YS (Fan, YS); Siu, VM (Siu, VM)

Source: AMERICAN JOURNAL OF MEDICAL GENETICS **Volume:** 102 **Issue:** 3 **Pages:** 266-271 **DOI:** 10.1002/ajmg.1

Times Cited in Web of Science Core Collection: 20

Total Times Cited: 21

Abstract: A derivative chromosome 8 was observed in a newborn boy who presented with low birth weight, multiple congeni
was further characterized at age 18 months by a high resolution G-banding analysis, spectral karyotyping, and fluorescence in
probes. The karyotype was described as 46,XY,der (8)(qter-->q24.13::p21.3 --> p23.3::p23.3 --> qter), representing an inverte
duplication of region 8q24.13-->qter, which attaches to the duplicated short arm segment at 8p2l.3. Different from previously r
(8p), no deletion was detected in the distal region of 8p in this case. This young child had manifested a broad nasal bridge, mic
agenesis of the corpus callosum, Dandy-Walker malformation, congenital heart defects, dysplastic kidneys, hydronephrosis, m
retardation. These features are compared with those commonly seen in cases with an inverted duplication of 8p and cases with
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PubMed ID: 11484205

Language: English

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ISSN: 0148-7299

Record 35 of 48

Title: Mosaic inv dup(8p) marker chromosome with stable neocentromere suggests neocentromerization is a post-zygotic ever

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Source: AMERICAN JOURNAL OF MEDICAL GENETICS **Volume:** 102 **Issue:** 1 **Pages:** 86-94 **DOI:** 10.1002/1096-862
AJMG1390>3.0.CO;2-T **Published:** JUL 22 2001

Times Cited in Web of Science Core Collection: 19

Total Times Cited: 21

Abstract: Marker human neocentromeres have been described in individuals where the chromosomes are non-mosaic, suggest
individuals where there is mosaicism, raising the possibility of neocentromere instability. We report two independently ascerta
supernumerary marker chromosome, shown by reverse chromosome painting to have an 8p origin, resulting in mosaicism for t
markers have a primary constriction but show no detectable centromeric or-satellite DNA, The marker in Patient 1 demonstrat
associated with nine different functionally critical centromere proteins. Investigation of peripheral blood lymphocytes from thi
year period showed 23-46% mosaicism for the marker chromosome with no decrease in incidence. In vitro investigation of pri

lymphoblast cell line derived from the patient demonstrated 100% stability of the marker chromosome indicating that neocentr for the mosaicism in the patient. This and other available data support a general model of neocentromerization as a post-zygoti supernumerary chromosome fragment has arisen during meiosis or post-fertilization at mitosis. (C) 2001 Wiley-Liss, Inc.

Accession Number: WOS:000169706400016

PubMed ID: 11471179

Language: English

Addresses: Royal Childrens Hosp, Murdoch Childrens Res Inst, Parkville, Vic 3052, Australia.
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ISSN: 0148-7299

Record 36 of 48

Title: Olfactory receptor-gene clusters, genomic-inversion polymorphisms, and common chromosome rearrangements

Author(s): Giglio, S (Giglio, S); Broman, KW (Broman, KW); Matsumoto, N (Matsumoto, N); Calvari, V (Calvari, V); Gime Ohashi, H (Ohashi, H); Voullaire, L (Voullaire, L); Larizza, D (Larizza, D); Giorda, R (Giorda, R); Weber, JL (Weber, JL); Lec (Zuffardi, O)

Source: AMERICAN JOURNAL OF HUMAN GENETICS **Volume:** 68 **Issue:** 4 **Pages:** 874-883 **DOI:** 10.1086/319506 **I**

Times Cited in Web of Science Core Collection: 259

Total Times Cited: 266

Abstract: The olfactory receptor (OR)-gene superfamily is the largest in the mammalian genome. Several of the human OR g equal to 10 members located on almost all human chromosomes, and some chromosomes contain more than one cluster. We de that unequal crossovers between two OR gene clusters in 8p are responsible for the formation of three recurrent chromosome r inversion polymorphism. The first two macrorearrangements are the inverted duplication of 8p, inv dup(8p), which is associat supernumerary marker chromosome, +der(8)(8p23.1pter), which is also a recurrent rearrangement and is associated with mino reciprocal of the inv dup(8p). The third macrorearrangment is a recurrent 8p23 interstitial deletion associated with heart defect maternal meiosis, we investigated the maternal chromosomes 8 in eight mothers of subjects with inv dup(8p) and in the mothe probes included between the two 8p-OR gene clusters. All the mothers were heterozygous for an 8p submicroscopic inversion and was present, in heterozygous state, in 26% of a population of European descent. Thus, inversion heterozygosity may cause leading to the formation of the inv dup(8p) or to its reciprocal product, the +der(8p). After the Yp inversion polymorphism, wh PRKX/PRKY translocation in XX males and XY females, the OR-8p inversion is the second genomic polymorphism that con chromosome rearrangements. Accordingly, it may be possible to develop a profile of the individual risk of having progeny wit

Accession Number: WOS:000167666000009

PubMed ID: 11231899

Language: English

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Giorda, Roberto	J-1052-2014	0000-0001-8175-9606
zuffardi, orsetta		0000-0002-1466-4559

ISSN: 0002-9297

Record 37 of 48

Title: Deletion of a 5-cM region at chromosome 8p23 is associated with a spectrum of congenital heart defects

Author(s): Giglio, S (Giglio, S); Graw, SL (Graw, SL); Gimelli, G (Gimelli, G); Pirola, B (Pirola, B); Varone, P (Varone, P); V Rossi, E (Rossi, E); Dellavecchia, C (Dellavecchia, C); Bonaglia, MC (Bonaglia, MC); Digilio, MC (Digilio, MC); Giannotti, Carrozzo, R (Carrozzo, R); Korenberg, JR (Korenberg, JR); Danesino, C (Danesino, C); Sujansky, E (Sujansky, E); Dallapicco O)

Source: CIRCULATION **Volume:** 102 **Issue:** 4 **Pages:** 432-437 **Published:** JUL 25 2000

Times Cited in Web of Science Core Collection: 56

Total Times Cited: 62

Abstract: Background-Cytogenetic evidence suggests that the haploinsufficiency of greater than or equal to 1 gene located in impairing heart differentiation and leading to a wide spectrum of congenital heart defects (CHDs), including conotruncal lesio defects, and pulmonary valve stenosis. An 8p heart-defect-critical region was delineated, and the zinc finger transcription facto these defects. We narrowed this region and excluded a major role of GATA4 in these CHDs.

Methods and Results-We studied 12 patients (7 had CHD and 5 did not) with distal 8p deletions from 9 families by defining th molecular level by fluorescent in situ hybridization and short-tandem repeat analysis. Subjects with 8p deletions distal to D8S1 telomere, did not have CHD, whereas subjects with a deletion that included the more proximal region suffered from the spectr distal deletions. The 5-cM critical region is flanked distally by D8S1706 and WI-8327, both at approximate to 10 cM, and pro nor angiopoietin-2 (ANGPT2; a gene in 8p23 involved in blood vessel formation) were found to be deleted in some of the criti related to the parental origin of deletion.

Conclusions-Haploinsufficiency for a gene between WI-8327 and D8S1825 is critical for heart development. A causal relation and ANGPT2 haploinsufficiency and CHDs.

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PubMed ID: 10908216

Language: English

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zuffardi, orsetta		0000-0002-1466-4559

ISSN: 0009-7322

Record 38 of 48

Title: Prenatal diagnosis of inverted duplicated 8p

Author(s): MacMillin, MD (MacMillin, MD); Suri, V (Suri, V); Lytle, C (Lytle, C); Krauss, CM (Krauss, CM)

Source: AMERICAN JOURNAL OF MEDICAL GENETICS **Volume:** 93 **Issue:** 2 **Pages:** 94-98 **DOI:** 10.1002/1096-862 AJMG3>3.0.CO;2-3 **Published:** JUL 17 2000

Times Cited in Web of Science Core Collection: 9

Total Times Cited: 9

Abstract: The phenotype of inverted duplicated 8p, region 8p11,2-p23, reported in children and adults, includes: severe menta of corpus callosum, and other malformations including those of heart and kidneys. We report on the prenatal diagnosis of 2 cas ascertained by abnormal level 2 ultrasound findings. Case I presented at 16.5 weeks of gestation with massive distention of the

abnormality of the lower lumbar spine, absence of the sacral spine and a Dandy-Walker variant (interhemispheric cyst and enla weeks of gestation with agenesis of corpus callosum, slightly enlarged lateral ventricles, interhemispheric cyst and enlarged th aorta. The intracranial and cardiac anomalies were confirmed and further defined after delivery. Cytogenetic analysis in both c cases, fluorescence in situ hybridization (FISH) defined the abnormal chromosome, as a pseudodicentric chromosome with du p23 and deletion from p23 to pter. Our findings support those of prior reports of the inverted duplicated 8p chromosome with r our knowledge. (C) 2000 Wiley-Liss, Inc.

Accession Number: WOS:000087674100003

PubMed ID: 10869109

Language: English

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ISSN: 0148-7299

Record 39 of 48

Title: Parental origin and mechanisms of formation of cytogenetically recognisable de novo direct and inverted duplications

Author(s): Kotzot, D (Kotzot, D); Martinez, MJ (Martinez, MJ); Bagci, G (Bagci, G); Basaran, S (Basaran, S); Baumer, A (Ba (Brecevic, L); Castellan, C (Castellan, C); Chrzanowska, K (Chrzanowska, K); Dutly, F (Dutly, F); Gutkowska, A (Gutkowska Krajewska-Walasek, M (Krajewska-Walasek, M); Luleci, G (Luleci, G); Miny, P (Miny, P); Riegel, M (Riegel, M); Schuffenha H); Schinzel, A (Schinzel, A)

Source: JOURNAL OF MEDICAL GENETICS **Volume:** 37 **Issue:** 4 **Pages:** 281-286 **DOI:** 10.1136/jmg.37.4.281 **Publisl**

Times Cited in Web of Science Core Collection: 40

Total Times Cited: 40

Abstract: Cytogenetic, FISH, and molecular results of 20 cases with de novo tandem duplications of 18 different autosomal cl 12 cases with direct duplications, three cases with inverted duplications, and five in whom determination of direction was not p between non-sister chromatids (N-SCR) was found, whereas in the remaining 13 cases sister chromatids (SCR) were involved. almost equally in cases with SCR (3:4) and N-SCR (4:3). In the cases with proven inversion, there was maternal and paternal c cytogenetically determined breakpoints correlated with common or rare fragile sites. In five cases, including all those with pro corresponded to common or rare fragile sites. In at least two cases, one with an interstitial duplication (dup(19)(q11q13)) and c (p10p23)), concomitant deletions (del(8) (p23p23.3) and del(19)(q13q13)) were found.

Accession Number: WOS:000086453000007

PubMed ID: 10745046

Language: English

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ISSN: 0022-2593

Record 40 of 48

Title: Cloning, sequencing, and analysis of Inv8 chromosome breakpoints associated with recombinant 8 syndrome

Author(s): Graw, SL (Graw, SL); Sample, T (Sample, T); Bleskan, J (Bleskan, J); Sujansky, E (Sujansky, E); Patterson, D (Pa

Source: AMERICAN JOURNAL OF HUMAN GENETICS **Volume:** 66 **Issue:** 3 **Pages:** 1138-1144 **DOI:** 10.1086/302821

Times Cited in Web of Science Core Collection: 21

Total Times Cited: 22

Abstract: Rec8 syndrome (also known as "recombinant 8 syndrome" and "San Luis Valley syndrome") is a chromosomal diso with ancestry from the San Luis Valley of southern Colorado and northern New Mexico. Affected individuals typically have m seizures, a characteristic facial appearance, and other manifestations. The recombinant chromosome is rec(8)dup(8q) inv(8)(p2 pericentric inversion, inv(8)(p23.1q22.1). Here we report on the cloning, sequencing, and characterization of the 8p23.1 and 8 chromosome associated with Rec8 syndrome. Analysis of the breakpoint regions indicates that they are highly repetitive. Of 6 consists of repetitive gene family members-including Alu, LINE, and LTR elements -and the inversion took place in a small si elements. Analysis of 3.7 kb surrounding the 8q22 breakpoint region reveals that it is 99% repetitive and contains multiple LT within one of the LTR elements.

Accession Number: WOS:000088373200032

PubMed ID: 10712224

Language: English

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ISSN: 0002-9297

Record 41 of 48

Title: A case of inv dup(8p) with early onset breast cancer

Author(s): Seltmann, M (Seltmann, M); Harrington, P (Harrington, P); Ponder, BAJ (Ponder, BAJ); Willatt, LR (Willatt, LR); Anton-Culver, H (Anton-Culver, H)

Source: JOURNAL OF MEDICAL GENETICS **Volume:** 37 **Issue:** 1 **Pages:** 70-71 **DOI:** 10.1136/jmg.37.1.70 **Published:**

Times Cited in Web of Science Core Collection: 4

Total Times Cited: 5

Accession Number: WOS:000084894400017

PubMed ID: 10691415

Language: English

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ISSN: 0022-2593

Record 42 of 48

Title: Duplication of 8p with minimal phenotypic effect transmitted from a mother to her two daughters

Author(s): Gibbons, B (Gibbons, B); Tan, SY (Tan, SY); Barber, JCK (Barber, JCK); Ng, CF (Ng, CF); Knight, LA (Knight,

Source: JOURNAL OF MEDICAL GENETICS **Volume:** 36 **Issue:** 5 **Pages:** 419-422 **Published:** MAY 1999

Times Cited in Web of Science Core Collection: 15

Total Times Cited: 15

Accession Number: WOS:000080290700015

PubMed ID: 10353792

Language: English

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ISSN: 0022-2593

Record 43 of 48

Title: Prenatal diagnosis of an 8p23.1 deletion in a fetus with a diaphragmatic hernia and review of the literature

Author(s): Faivre, L (Faivre, L); Morichon-Delvallez, N (Morichon-Delvallez, N); Viot, G (Viot, G); Narcy, F (Narcy, F); Loi L); Aubry, MC (Aubry, MC); Raclin, V (Raclin, V); Edery, P (Edery, P); Munnich, A (Munnich, A); Vekemans, M (Vekemans

Source: PRENATAL DIAGNOSIS **Volume:** 18 **Issue:** 10 **Pages:** 1055-1060 **DOI:** 10.1002/(SICI)1097-0223(1998100)18:
OCT 1998

Times Cited in Web of Science Core Collection: 34

Total Times Cited: 34

Abstract: The prenatal diagnosis of an 8p23.1 deletion is reported. The diagnosis was ascertained at 22 weeks of gestation bec
hernia at ultrasound. Following cytogenetic studies and counselling, the pregnancy was terminated. An autopsy confirmed the
revealed also the existence of an atrio-ventricular canal (AVC) and an atrial septal defect (ASD). The clinical features of this a
those observed in 16 previously reported cases with an identical deletion of the short arm of chromosome 8. This suggests that
whenever a diaphragmatic hernia and/or an AVC is detected on ultrasound. (C) 1998 John Wiley & Sons, Ltd.

Accession Number: WOS:000076977800009

PubMed ID: 9826897

Language: English

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ISSN: 0197-3851

Record 44 of 48

Title: Normal adaptive function with learning disability in duplication 8p including band p22

Author(s): Brooks, SS (Brooks, SS); Genovese, M (Genovese, M); Gu, H (Gu, H); Duncan, CJ (Duncan, CJ); Shanske, A (Sh

Source: AMERICAN JOURNAL OF MEDICAL GENETICS **Volume:** 78 **Issue:** 2 **Pages:** 114-117 **DOI:** 10.1002/(SICI)1(
AJMG3>3.3.CO;2-6 **Published:** JUN 30 1998

Times Cited in Web of Science Core Collection: 16

Total Times Cited: 16

Abstract: Duplication 8p usually results in a syndrome characterized by profound mental retardation, mild facial anomalies, a
report on a large kindred segregating a Y;8 translocation in whom several individuals have duplication 8p22-->8pter. These inc
their unbalanced karyotype, The family was studied with G-banding and fluorescent in situ hybridization (FISH) using probes
family with other reported cases defines a mild clinical outcome for trisomy 8p22-->8pter in contrast to the severe findings wh
proximal segment. Am. J, Med, Genet, 78:114-117, 1998, (C) 1998 Wiley-Liss, Inc.

Accession Number: WOS:000074511200003

Language: English

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ISSN: 0148-7299

eISSN: 1096-8628

Record 45 of 48

Title: Duplication of 8p23.1: a cytogenetic anomaly with no established clinical significance

Author(s): Barber, JCK (Barber, JCK); Joyce, CA (Joyce, CA); Collinson, MN (Collinson, MN); Nicholson, JC (Nicholson, J
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Source: JOURNAL OF MEDICAL GENETICS **Volume:** 35 **Issue:** 6 **Pages:** 491-496 **DOI:** 10.1136/jmg.35.6.491 **Publisl**

Times Cited in Web of Science Core Collection: 57

Total Times Cited: 59

Abstract: We present seven families with a cytogenetic duplication of the short arm of chromosome 8 at band 8p23.1. The du
offspring in four of the seven families.
In three families, the source of the extra material and its euchromatic origin were established using FISH with a YAC which w
paint for chromosome 8. FISH signals from this YAC were significantly larger on the duplicated chromosome compared with t
members tested. Comparative genomic hybridisation (CGH) on a representative subject was consistent with these results.
The families were ascertained for a variety of mostly incidental reasons including prenatal diagnosis for advanced maternal ag
multiple phenotypically normal family members with no history of reproductive loss suggests the existence of a novel class of
euchromatic variants or duplications with no phenotypic effect.

Accession Number: WOS:000074038400011

PubMed ID: 9643291

Language: English

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Record 46 of 48

Title: Ataxic gait and mental retardation with absence of the paternal chromosome 8 and an idic(8)(p23.3): Imprinting effect o

Author(s): Piantanida, M (Piantanida, M); Dellavecchia, C (Dellavecchia, C); Floridia, G (Floridia, G); Giglio, S (Giglio, S); Danesino, C (Danesino, C); Schinzel, A (Schinzel, A); Zuffardi, O (Zuffardi, O)

Source: HUMAN GENETICS **Volume:** 99 **Issue:** 6 **Pages:** 766-771 **DOI:** 10.1007/s004390050445 **Published:** JUN 1997

Times Cited in Web of Science Core Collection: 19

Total Times Cited: 19

Abstract: A female child with mild dysmorphisms, motor and mental retardation had a 45,XX,-8,-8,+psu dic(8)(p23.3) karyot in a lymphoblastoid cell line. DNA analysis showed that the proposita was nullisomic for the 8pter region distal to D8S264, at DNA polymorphisms of 38 loci spread along the entire chromosome 8 revealed that only maternal alleles were present, distrib homozygous regions. This finding indicated that the rearrangement occurred during maternal meiosis in a chromosome recoml our knowledge this is the first case of uniparental maternal disomy for chromosome 8 and of nullisomy for the distal 1-cM por favour of the assumption that no imprinted genes are present on chromosome 8. Thus, dysmorphisms, motor and mental retard the nullisomy for the region distal to D8S264, a region in which a recessive gene for epilepsy with progressive mental retardat

Accession Number: WOS:A1997XB30500011

PubMed ID: 9187670

Language: English

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ISSN: 0340-6717

Record 47 of 48

Title: The same molecular mechanism at the maternal meiosis I produces mono- and dicentric 8p duplications

Author(s): Floridia, G (Floridia, G); Piantanida, M (Piantanida, M); Minelli, A (Minelli, A); Dellavecchia, C (Dellavecchia, C E); Gimelli, G (Gimelli, G); Croci, G (Croci, G); Franchi, F (Franchi, F); Gilgenkrantz, S (Gilgenkrantz, S); Grammatico, P (C (Wood, S); Danesino, C (Danesino, C); Zuffardi, O (Zuffardi, O)

Source: AMERICAN JOURNAL OF HUMAN GENETICS **Volume:** 58 **Issue:** 4 **Pages:** 785-796 **Published:** APR 1996

Times Cited in Web of Science Core Collection: 96

Total Times Cited: 100

Abstract: We studied 16 cases of 8p duplications, with a karyotype 46,XX or XY,dup(8p), associated with mental retardation, demonstrate that these 8p rearrangements can be either dicentric (6 cases) with the second centromere at the tip of the short arı region, from D8S349 to the telomere, including the defensin 1 locus, is deleted in all the cases. The region spanning from D8S present in single copy, and the remaining part of the abnormal 8 short arm is duplicated in the dicentric cases and partially dup of the duplication always spans up to D8S552 (8p23.1), while its proximal edge includes the centromere in the dicentric cases ones. The analysis of DNA polymorphisms indicates that the rearrangement is consistently of maternal origin. In the deleted re patient. In the duplicated region, besides one paternal allele, some loci showed two different maternal alleles, while others, wh only one maternal allele. We hypothesize that, at maternal meiosis I, there was abnormal pairing of chromosomes 8 followed b by D8S552 and D8S35 and by D8S252 and D8S349, which presumably contain inverted repeated sequences. The resulting dic 8p23.1(D8S552)::8p23.1-(D8S35)-8qter, due to the presence of two centromeres, breaks at anaphase I, generating an inverted

at the centromere or monocentric if it occurs between centromeres.

Accession Number: WOS:A1996UA38000017

PubMed ID: 8644743

Language: English

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ISSN: 0002-9297

Record 48 of 48

Title: D8S7 IS CONSISTENTLY DELETED IN INVERTED DUPLICATIONS OF THE SHORT ARM OF CHROMOSOMI

Author(s): MINELLI, A (MINELLI, A); FLORIDIA, G (FLORIDIA, G); ROSSI, E (ROSSI, E); CLEMENTI, M (CLEMEN CAMURRI, L (CAMURRI, L); BERNARDI, F (BERNARDI, F); HOELLER, H (HOELLER, H); RE, CP (RE, CP); MARAS (WOOD, S); ZUFFARDI, O (ZUFFARDI, O); DANESINO, C (DANESINO, C)

Source: HUMAN GENETICS **Volume:** 92 **Issue:** 4 **Pages:** 391-396 **DOI:** 10.1007/BF01247342 **Published:** OCT 1993

Times Cited in Web of Science Core Collection: 29

Total Times Cited: 29

Abstract: Ten patients with inverted duplication of 8p (inv dup 8p) were studied with cytogenetic, biochemical and molecular 8p12-p22 was always associated with a deletion of the locus D8S7 (mapped in 8p23.1) as demonstrated with the probe pSW5(blot. Restriction fragment length polymorphisms detected by probes pSW50 (1 case) and by pG2LPL35 (locus LPL) (two case the anomaly. The activity of glutathione reductase, whose gene maps in the duplicated region at 8p21.1, was increased in all pa 8p includes neonatal hypotonia, prominent forehead, large mouth with everted lower lip, abnormally shaped large ears, brain n Our findings indicate that the chromosome rearrangement is homogeneous at least for the presence of the deletion and support origin.

Accession Number: WOS:A1993MF05000016

PubMed ID: 7901142

Language: English

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