

Appendix 1

Cases of paternal origin of the trisomy excluded from study:

[1] Kohlhase J, Janssen B, Weidenauer K, Harms K, Bartels I. First confirmed case with paternal uniparental disomy of chromosome 16. *Am J Med Genet* 2000;91:190-191.

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Cases of partial trisomy excluded from study:

[1] Devi AS, Egan JFX, Campbell W, Ingardia C, Rosengren S, Tezcan K, Weiser J, Benn PA. Poor pregnancy outcome and the presence of trisomy 16 cells in amniotic fluid. *Am J Hum Genet* 1997;61:A151.

[2] Hsu WT, Shchepin DA, Mao R, Berry-Kravis E, Garber AP, Fischel-Ghodsian N, Falk RE, Carlson DE, Roeder ER, Leeth EA, Hajianpour MJ, Wang J-C C, Rosenblum-Vos LS, Bhatt SD, Karson EM, Hux CH, Trunca C, Bialer MG, Linn SK, Schreck RR. Mosaic trisomy 16 ascertained through amniocentesis: evaluation of 11 new cases. *Am J Med Genet* 1998;80:473-480.

[3] Schinzel A, Kotzot D, Brecevic L, Robinson WP, Dutly F, Dauwerse H, Binkert F, Baumer A, Ausserer B. Trisomy first, translocation second, uniparental disomy and partial trisomy third: a new mechanism for complex chromosomal aneuploidy. *Eur J Hum Genet* 1997;5:308-314.

Cases of concomitant aneuploidy excluded from study:

[1] Robinson WP, Barrett IJ, Bernard L, Telenius A, Bernasconi F, Wilson RD, Best RG, Howard-Peebles PN, Langlois S, Kalousek DK. Meiotic origin of trisomy in confined placental mosaicism is correlated with presence of fetal uniparental disomy, high levels of trisomy in trophoblast, and increased risk of fetal intrauterine growth restriction. *Am J Hum Genet* 1997;60:917-927.

Cases from UBC study with some data previously published:

[1] Kalousek DK, Howard-Peebles PN, Olson SB, Barrett IJ, Dorfmann A, Black SH, Schulman JD, Wilson RD. Confirmation of CVS mosaicism in term placentae and high frequency of intrauterine growth retardation association with confined placental mosaicism. *Prenat Diagn* 1991;11:743-450.

[2] Kalousek DK, Langlois S, Barrett IJ, Yam I, Wilson DR, Howard-Peebles PN, Johnson MP, Giorgiutti E. Uniparental disomy for chromosome 16 in humans. *Am J Hum Genet* 1993;52:8-16.

[3] Robinson WP, Barrett IJ, Bernard L, Telenius A, Bernasconi F, Wilson RD, Best RG, Howard-Peebles PN, Langlois S, Kalousek DK. Meiotic origin of trisomy in confined placental mosaicism is correlated with presence of fetal uniparental disomy, high levels of trisomy in trophoblast, and increased risk of fetal intrauterine growth restriction. *Am J Hum Genet* 1997;60:917-927.

[4] Peñaherrera MS, Barrett IJ, Brown CJ, Langlois S, Yong S-L, Lewis S, Bruyere H, Howard-Peebles PN, Kalousek DK, Robinson WP. An association between skewed X-chromosome inactivation and abnormal outcome in mosaic trisomy 16 confined predominantly to the placenta. *Clin Genet* 2000;58:436-446.

[5] Stavropoulos DJ, Bick D, Kalousek DK. Molecular cytogenetic detection of confined gonadal mosaicism in a conceptus with trisomy 16 placental mosaicism. *Am J Hum Genet* 1998;63:1912-1914.

Cases from UBC study overlapping with cases published by other research groups:

[1] Chan Y, Silverman N, Jackson L, Wapner R, Wallerstein R. Maternal uniparental disomy of chromosome 16 and body stalk anomaly. *Am J Med Genet* 2000;94:284-286.

[2] Dworniczak B, Koppers B, Kurlemann G, Holzgreve W, Horst J, Miny P. Uniparental disomy with normal phenotype. *Lancet* 1992;340:1285.

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- [5] Hsu WT, Shchepin DA, Mao R, Berry-Kravis E, Garber AP, Fischel-Ghodsian N, Falk RE, Carlson DE, Roeder ER, Leeth EA, Hajianpour MJ, Wang J-C C, Rosenblum-Vos LS, Bhatt SD, Karson EM, Hux CH, Trunca C, Bialer MG, Linn SK, Schreck RR. Mosaic trisomy 16 ascertained through amniocentesis: evaluation of 11 new cases. *Am J Med Genet* 1998;80:473-480.
- [6] Johnson MP, Childs MD, Robichaux III AG, Isada NB, Pryde PG, Koppitch III FC, Evans MI. Viable pregnancies after diagnosis of trisomy 16 by CVS: lethal aneuploidy compartmentalized to the trophoblast. *Fetal Diagn Ther* 1993;8:102-108.
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- [8] Schneider AS, Bischoff FZ, McCaskill C, Coady ML, Stopfer JE, Shaffer LG. Comprehensive 4-year follow-up on a case of maternal heterodisomy for chromosome 16. *Am J Med Genet* 1996;66:204-208.
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Cases from other published reports to date:

[1] Abu-Amero SN, Ali Z, Abu-Amero KK, Stanier P, Moore GE. An analysis of common isodisomic regions in five mUPD 16 probands. *J Med Genet* 1999;36:204-207.

[2] Association of Clinical Cytogenetics Working Party on Chorionic Villi in Prenatal Diagnosis. Cytogenetic analysis of chorionic villi for prenatal diagnosis: An ACC collaborative study of U.K. data. *Prenat Diagn* 1994;14:363-379.

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