49,XXYY,+18 in a liveborn male

We report a karyotype 49,XXYY,+18 involving aneuploidy of the sex chromosomes plus trisomy 18 which relates to a case reported in this journal.¹ This is the first reported case of triple aneuploidy in a male, that is, additional chromosomes X, Y, and 18. As in cases of 48,XXY,+18² and the one 49,XXXY,+18,¹ typical features of trisomy 18 were seen, without the phenotypic characteristics of sex chromosome aneuploidy which are not expressed in newborns.

The male proband was delivered by Caesarean section because of fetal distress after 36 weeks of a pregnancy complicated by hydramnios. The mother was 39 and her husband 44 years old. The baby was small for dates: weight 1410 g, crown-heel 39 cm, crown-rump 26.5 cm, head circumference 30.4 cm, and placental weight 312 g. Apgar scores were 2 at 1 minute, 4 at 5 minutes, and 4 at 10 minutes with survival for only 1.5 hours. Hypertelorism, an anti-mongoloid slant, prominent forehead, micrognathia, and small, low set ears were observed (figure a). Bilateral buphthalmos was probably present but was not confirmed at necropsy. The left hand showed a simian crease and a flexion deformity of the second finger. Rocker-bottom feet were present (figure b). The testes were undescended. The heart showed tetralogy of Fallot and persistent ductus arteriosus. The lungs were poorly expanded and showed general immaturity microscopically. The adrenal glands were congested with the cortex showing signs of disruption. The brain appeared mature but with degeneration of the subependymal vessels. The oesophagus terminated in a blind pouch at the upper end and the lower end joined the right main bronchus.

Standard cytogenetical procedures including GTL and CBG banding were used on lymphocytes and fibroblasts of the proband and his parents. The proband was 49,XXYY,+18 in both tissues. The mother showed a few 45,X and 47,XXX cells in both tissues at a level (4.7%) consistent with her age. Her husband was mosaic 46,XY/47,XXY. In his lymphocytes we found 2.6% 46,XY cells but 16.7% was recorded 7 years previously when he was also found to be azoospermic. In the fibroblasts there was only 1% of 46,XY cells. Mosaic 46,XY/47,XXY males can be fertile,³ but we remain uncertain of the paternity of the proband with no possibility of further testing. There is an 8 year old normal 46,XY son from the present marriage.

There is a greater than chance association of XXY with trisomy 18,² so it seems likely that the chance of association of any state of sex chromosome aneuploidy, including XXYY, with trisomy 18 will also be greater than random.

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FIGURE (a) Profile of proband. Note indication of buphthalmos, small, low set ears, and micrognathia. (b) The proband showing hypertelorism and rocker-bottom feet.

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