skull or brain at that time. The risk of chromosomal abnormality was explained, and then she underwent amniocentesis at 26 weeks of pregnancy. The chromosome analysis of cultured amniocytes indicated a 47,XY+13 complement. Intensive ultrasonography was performed and a cleft lip, polydactyly and overlapping finger were visualized at 28 weeks of gestation. At 36 weeks, slight dilatation of the lateral ventricles of the brain and coarctation of the aorta were detected. At 37 weeks, PROM occurred and one hour later, a male infant weighing 2444 g was born transvaginally. The baby had a left cleft lip and palate and rigid hands with polydactyly. There were no major abnormalities in his head and eyes. A cardiac ultrasound by a pediatric cardiologist yielded an interruption of the aortic arch. After discussion with the pediatrician, the parents did not want to perform the operation and decided to observe a natural course. At 6 days of life, the baby died with cardiac failure.

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Uehara, S.¹, Sato, K.¹, Hashiyada, M.², Nata, M.², Yaegashi, N.¹ and Okamura, K.¹ ¹Department of Obstetrics and Gynecology, and ²Department of Forensic Medicine, Tohoku University School of Medicine, Sendai, Miyagi. *X chromosome inactivation patterns in 45,X/46,XX mosaics*

Although preferential X chromosome inactivation (XCI) has been observed in women with various kinds of structural abnormalities of the X chromosome, little is known about XCI patterns in X/XX mosaics. Therefore, to investigate XCI patterns in X/XX mosaics, genomic DNA was extracted from peripheral blood samples of 15 female subjects who showed different proportions of 45,X cell clones. Karyotyping with G-banding or high resolution banding revealed no structural aberrations of X chromosomes in the subjects. XCI patterns were analyzed using two assays. The first assay was the BsrXI restriction endonuclease detection of an X-linked phosphoglycerate kinase gene (PGK) polymorphism following digestion of the DNA with methylation-sensitive HpaII, or with methylation-insensitive AflAI as a control. The second assay was the detection of a CAG triplet repeat polymorphism in the X-linked androgen receptor gene (AR) after sodium bisulfite treatment. Of the 15 subjects, 11 were informative due to heterozygosity for at least one of the polymorphisms (6 were heterozygous for the PGK polymorphism and 9 were heterozygous for the AR polymorphism). Four of the 11 informative subjects (36%) showed preferential XCI for at least one of the polymorphisms, which was a much higher incidence than previously reported for normal females. Moreover, 3 of these 4 women had proportions of 45,X cell clones greater than 20%. Although our results may be due to several possible cytogenetic or molecular mechanisms, the most likely explanation is that cases of 45,X/46,XX that contain relatively high levels of 45,X cell clones probably arose due to structural aberrations of the X chromosome undetectable by conventional karyotyping.

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Sun, X.Z.¹, Takahashi, S.¹, Kubota, Y.¹, Sato, H.¹, Fukui, Y.² and Inouye, M.³ ¹Environmental and Toxicological Sciences Research Group, National Institute of Radiological Sciences, Chiba, ²Department of Anatomy, Tokushima University School of Medicine, Tokushima, and ³Shin Nippon Biomedical Laboratories, Ltd., Kagoshima. *Neocortical heterotopias and their three-dimensional distribution in the developing brain subjected to X-irradiation in utero*

We have previously reported the histological features of neocortical heterotopia (NHA) in mouse brains following prenatal exposure to X-irradiation. This study focused on determining the types of NHA and their three-dimensional distribution. Pregnant ICR mice were subjected to a single whole-body X-irradiation at a dose of 1.5 Gy on embryonic day 13. Brains from pups were obtained at different ages. Specimens were serially sectioned in the frontal plane at 15 μm with the aid of a freezing microtome, and every fourth section was retained and stained with crystal violet or HE. The image data of these sections were collected in a computer and reconstructed to three dimensional brain structures using the Magellan 3.6 program. Then sectional images were drawn on a computer display at 240 μm intervals, and the positions of the different types of NHA were marked using color coding. Three types of NHA were recognized in the irradiated brains: neocortical Layer I heterotopia (L1H), periventricular heterotopia (PHA), and hippocampal heterotopia (HHA). L1H were identified as small patches in the caudal occipital cortex and were located more laterally in the neocortex in caudal sections than in rostral sections. PHA were located more rostrally than L1H, found from the most caudal extent of the presumed motor cortex to the most caudal extent of the lateral ventricle. HHA appeared as continuous linear bands. They were frequently associated with the anterior parts of the PHA.

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Sumida, H.¹, Nitta, Y.² and Hoshi, M.³ ¹Department of Clinical Radiology, Faculty of Health Sciences, Hiroshima International University, Kurose, Hiroshima, and ²International Radiation Information Center, Research Institute for Radiation Biology and Medicine, Hiroshima University, Hiroshima. *Exencephaly and reduction of NT-3 expression by ²⁵²Cf exposure on mouse embryos*