established lymphoblastoid cells was similar to that in the peripheral blood lymphocytes, while in cultured skin fibroblasts the cell line with 45,X predominated (90%). High-resolution banding patterns of the abnormal chromosome were consistent with dic(Y)(qter-p11.2::p11.2qter).

Review of the literature showed 5 previously reported cases with dic(Yq) and a complete male phenotype, 4 of whom was ascertained because of azoospermia. The biopsy finding in the present case along with the existence of a unique case with a nonmosaic dic(Yq) and azoospermia argues that impaired meiotic pairing between dic(Yq) and X chromosomes leads to futile spermatogenesis. Moreover, the cytogenetic finding in our case suggests that the gene encoding testis determining factor may be assigned to Yp11.2.

OHSHIGE, T., K. NAGAI, T. IKEDA, N. MORI and S. MARUTA, Department of Obstetrics and Gynecology, Miyazaki Medical College, Kiyotake, Miyazaki, and Maruta Hospital, Miyakonojo, Miyazaki. A case of Down's syndrome with translocation diagnosed in the first trimester by ultrasonography. Several congenital diseases show fetal cranial and craniocervical masses. In 1985, Benacerraf et al. described increased skin thickness at the back of the fetal neck as being highly specific for Down's syndrome. But there is no report of translocation type of 21-trisomy with this sign detected in the first trimester. We report a case of Down's syndrome with translocation type of 21-trisomy, in which nuchal fold thickness was shown at 13 weeks' gestation.

Case: 25-year-old woman, primigravida. Ultrasound performed at 13 weeks' gestation revealed echo lucent area in the fetal occipital region and right side of the neck. From this ultrasonic sign, the fetus was suspected of Down's syndrome. The fetus, terminated at 17 weeks' gestation, showed cranial edema (5 mm thick) and redundant skin (3 x 3 cm) on the right side of the neck. Chromosomal analysis of umbilical cord blood revealed 21-trisomy, translocation type (46, XY,-14, +t(14q21q)). The karyotypes of parents were normal.

Conclusion: Nuchal fold thickness in the first trimester was detected in translocation type of 21-trisomy. Our observation suggests that this anomaly could also be found in translocation type.

KANEKO, M., T. IKEDA, Y. HARUYAMA and N. MORI, Department of Obstetrics and Gynecology, Miyazaki Medical College, Kiyotake, Miyazaki. Prenatal diagnosis of osteogenesis imperfecta. Osteogenesis imperfecta (O.I.) is one of the lethal short-limbed dwarfs. Most prenatally diagnosed cases die of intracranial hemorrhage and respiratory insufficiency either in utero or in early infancy. Prenatal diagnosis of O.I. is essential for perinatal management. We report a case diagnosed as O.I. prenatally and discuss the problems in diagnosis and management. [Case report] A 35-year-old woman, gravida 1, para 0, first visited our hospital at 35 weeks gestation. There was no family history of malformation in either the patient or her partner. An ultrasound examination revealed a single fetus in the breech presentation and oligohydramnios. At 38 weeks gestation, she was admitted for elderly primipara, breech presentation, oligohydramnios and intrauterine growth retardation. The bicipital diameter was 87 mm (mean for 38 wk gestation). The abdominal circumference was 296 mm (mean for 33 wk). All four limbs were short and bowed making it difficult to obtain a satisfactory plane for measurement. The left femur was broken. A KUB revealed a poorly calcified skull. From these findings, O.I. was suspected. At 39 weeks, cesarean section was performed and she delivered a 2,135 g male infant with Apgar score 8 at 1 and 9 at 5 minutes. Examination of the infant revealed thinning of the skull bone, blue sclerae, short limbs, deformity and multiple fracture. The spine and chest wall were normal. [Conclusion] When O.I. is suspected prenatally, it is important to evaluate the fetal chest and lungs in order to ascertain whether it is a lethal type or not.

TSUTSUI, Y., A. KASHIWAI, N. KAWANURA, M. NAGAHAMA and I. NARUSE, Institute for Developmental Research, Aichi Prefectural Colony, Kasugai, Aichi. Postnatal brain damage of mice induced by intraventricular injection of murine cytomegalovirus in the late stage of gestation. Mouse embryos were infected with murine cytomegalovirus (MCMV) by injecting the virus into the cerebral ventricles in the late stage of gestation. The brains of the offspring were then analyzed by immunohistochemical methods. Brains of offspring, which were injected with relatively high titers of MCMV (1 x 10^6 p.f.u./embryo) on day 13 of gestation ex utero or on day 15 of gestation in utero, showed massive necrosis of the cerebral cortex with gliomesodermal proliferation around 10 days after birth. Viral antigen positive cells in the brains showed zonal arrangement in the cortex and hippocampus. Immunohistochemical double staining showed that some viral antigen positive cells had also reacted with antibody to neuron-specific enolase (NSE) but had hardly reacted with brain-type
creatinine kinase (CK-BB) or glial fibrillary acidic protein (GFAP). Brains of offspring, which were injected with relatively low titers of virus (1 × 10^3 p.f.u./embryo) on day 15 of gestation, showed zonal arrangement of viral antigen positive cells mainly in the cortex and hippocampus and some of these offspring showed atrophy of the cortex and hippocampus. (We thank Dr. K. Kato, Department of Biochemistry of our institute for providing the antibodies to NSE and CK-BB.)

KAMIYA, Y., M. OHNO, T. YAMATO and M. SHIMADA, Department of Pediatrics, Shiga University of Medical Science, Otsu, Shiga. Postnatal development of experimentally-induced microcephalic mice: Pathological study of visceral development.

We studied postnatal development in the internal organs of experimentally-induced microcephalic mice. These mice were produced by injecting cytosine arabinoside (Ara-C) to pregnant ICR-JCR mice. We made macrographic and microscopic observations on the 20, 30, 50 and 85 days of the organs: lung, heart, liver, kidney, small intestine and large intestine.

On macrography, each of the organs in the treated mice showed a smaller size with no other anomaly. We obtained the following results in microscopic investigation: In the large intestine, the submucosal and muscular layers were thin and Auerbach's myenteric plexuses disclosed severe immaturity with a small number of ganglion cells. The myocardial walls also were thin with little evidence of fibrosis. No microscopic change was proved in the other organs.

Growth was stunted and catch-up never occurred in the organs of the microcephalic mice. That was partially because of undernutrition. Besides, the hypoplasia of some organ systems was caused directly by Ara-C the same as brain injury inducing microcephalus. An example was given in the hypoplasia of Auerbach's plexus, in a mouse formed on the 13-14 days of gestation; coincident with the injecting of Ara-C in this experiment.

BEPPU, K., M. ENOMOTO, S. ADACHI, K. NABUCHI, M. AKASAKI, Y. TSUJI, I. MORIYAMA and M. ICHIJO, Department of Obstetrics and Gynecology, Nara Medical University, Kashihara, Nara. Three cases of holoprosencephaly.

Three cases of holoprosencephaly were reported. In case 1, a female newborn baby of semilobar type holoprosencephaly with facial anomalies including hypotelorism, nasal deformities, and cleft lip and palate, and with other anomalies such as tetralogy of Fallot was shown. Because of bad vital signs, she was introduced and admitted to our N.I.C.U., but died 60 hrs after birth. In the karyotype analysis, 13 trisomy was proved.

In case 2, a female newborn baby, who was delivered by Cesarian section due to dystocia caused by cephalopelvic-dysproportion, was introduced and admitted to our N.I.C.U. for the purpose of V-P shunt operation. This case was diagnosed as semilobar type holoprosencephaly with no other anomalies by CT, MRI, and USG examinations.

In case 3, a fetus was diagnosed of hydrocephalus by USG examination at the pregnant state of 29-weeks. This case proved to be a lobar type holoprosencephaly by MRI and USG examinations. In the pathological anatomy, no major anomalies were recognized except brain defects. Both in case 2 and 3, the causes were unknown.

Even today, a baby of holoprosencephaly can not be expected to survive, so correct and prudent intrauterine diagnosis in the early pregnant state is required.

ARIZAWA, M., Y. WAKAHAMA and M. NAKAYAMA, Department of Pathology, Osaka Medical Center and Research Institute for Maternal and Child Health, Izumi, Osaka. Clinicopathological study of holoprosencephaly.

The incidence of holoprosencephaly has been estimated at 14 in 144,670 births (0.01%) in Japan. Several etiological factors have been implicated in holoprosencephaly, 13 trisomy, 18 trisomy, maternal syphilis, and maternal diabetes mellitus. We summarized clinical signs and histological studies. The incidence of our study was 9 in about 12,000 births. 2 cases were 13 trisomy, 2 were 18 trisomy, 2 were maternal diabetes mellitus and 3 had no other complications. Cases of holoprosencephaly with normal chromosomal karyotype had no extracranial abnormalities. Extracranial abnormalities were often detected in trisomy cases, such as cardiac defect, overlapping fingers, polydactyly, or rocker-bottom feet. Hypoplasia of adrenal glands and hyperplasia of thymus possibly induced by hypoplasia of pituitary glands were observed in 6 cases. Two of them were complicated with diabetes in mother and one case had peculiar neurologic abnormalities such as defects of I to VI cranial nerves. A recent study reported there were cardiac or caudal defects with holoprosencephaly of diabetic mother, but our two cases had neither.