Recent Incidence of Trisomy 21 in a Japanese Hospital

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To the Editor:

Trisomy 21, the chromosome abnormality responsible for over 95% of Down syndrome, is one of the most common conditions encountered in the genetic clinic. Recently, the increasing share of pregnancies with advanced maternal age and its impact on the increased incidence of trisomy 21 have been reported in the United States [1-3]. Our hospital is one of main perinatal centers in Tokyo, Japan (about 2,100 deliveries per year). In this study, we examined the recent incidence of trisomy 21 associated with maternal age at our hospital.

We reviewed the obstetric records of all Japanese singleton pregnancies at our hospital form January 2005 through January 2011. Demographic information and the characteristics of labor were extracted from patient charts. The conditions of the study patients were Japanese singleton pregnancies whose estimated date of delivery were from January 2006 through December 2011 and managed from 14 weeks or earlier at our hospital. In this study, thus, we have excluded the cases as follows (1) multifetal pregnancies, (2) non-Japanese pregnancies, (3) the time of first visit of our hospital at \geq 15 weeks' gestation, and/or (4) spontaneous fetal demise before 22 weeks' gestation. We examined the maternal age at age of the estimated date of delivery.

There were 28 cases of trisomy 21 in 10964 Japanese singleton pregnancies (1/392). In our hospital, there were 39 cases of trisomy 21 in total during this period; however, there were 8 cases of trisomy 21 refereed to our hospital at \geq 15 weeks' gestation due to fetal abnormality, 2 non-Japanese cases of trisomy 21 and 1 case of trisomy 21 in twin pregnancy. During this period, the average maternal age of all pregnancies was 32.3 ± 5.5 years old and 35% of the mothers were 35 years old or more at the estimated date of delivery; while the average maternal age in the cases with trisomy 21 was 38.0 ± 4.7 years old.

The incidence of trisomy 21 in cases of maternal age of < 35

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years (3 cases, average: 27.7 ± 3.5 years), 35 - 39 years (15 cases, average: 37.5 ± 1.1 years) and ≥ 40 years (10 cases, average: 41.7 ± 1.9 years) were 1/2364, 1/209 and 1/74, respectively. These incidences by maternal age were almost similar to those reported previously in the United States [4].

Of the 28 cases of trisomy 21, 11 cases (39%) were diagnosed as trisomy 21 prenatally using amniocentesis due to maternal request. Ten cases of them (91%) were artificial aborted; thus, the incidence of live births with trisomy 21 was 1/608 (18 cases). The average maternal age in the cases diagnosed prenatally was significant higher than that diagnosed postnatelly (40.1 \pm 3.3 vs. 36.6 \pm 5.0 years, p < 0.01 by unpaired *t*-test). The tendency is also similar to that reported in the United States [1-3].

In conclusion, in Japan the recent increased share of pregnancies with advanced maternal age seemed to contribute to the increased incidence of trisomy 21 as observed in other countries.

References

- Egan JF, Smith K, Timms D, Bolnick JM, Campbell WA, Benn PA. Demographic differences in Down syndrome livebirths in the US from 1989 to 2006. Prenat Diagn. 2011;31(4):389-394.
- 2. Hassold T, Sherman S. Down syndrome: genetic recombination and the origin of the extra chromosome 21. Clin Genet. 2000;57(2):95-100.
- Resta RG. Changing demographics of advanced maternal age (AMA) and the impact on the predicted incidence of Down syndrome in the United States: Implications for prenatal screening and genetic counseling. Am J Med Genet A. 2005;133A(1):31-36.
- Sheets KB, Crissman BG, Feist CD, Sell SL, Johnson LR, Donahue KC, Masser-Frye D, et al. Practice guidelines for communicating a prenatal or postnatal diagnosis of Down syndrome: recommendations of the national society of genetic counselors. J Genet Couns. 2011;20(5):432-441.

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