Prenatal Genetics, Reproductive Genetics

AB042. Prenatal genetic counseling in the Philippine setting: a case series

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Background: Prenatal genetic counseling service in the Philippines is limited and primarily serves to guide the obstetrician in the management of the case. Moreover, termination of pregnancy, especially if due to genetic conditions, is not practiced. Prenatal testing methods, however, has moved forward from simple ultrasonography to utilizing molecular methods of testing. This can lead to challenges for the geneticist/genetics counselor to provide effective, insightful, and helpful counseling.

Methods: Here, we discuss three cases of pregnancies with fetal malformations on congenital anomaly scans and subsequent amniocentesis for interphase FISH for fetal karyotyping limited to the three common autosomal trisomies.

Results: One pregnancy showed trisomy 18, the second was trisomy 21, while the last case was normal for chromosomes 21, 13, and 18. The lack of pre-test counseling, different prognosis of each trisomy, the prenatal options that can then be presented to the family, and importance of post-delivery counseling and support are important points learned from these cases.

Conclusions: Pre- and post-test counselling and option of pregnancy are essential for genetic counselling service.

Keywords: Prenatal diagnosis; amniocentesis; karyotyping; counseling

doi: 10.21037/atm.2017.s042

Cite this abstract as: Cavan BC, Mendoza A, Veloso G. Prenatal genetic counseling in the Philippine setting: a case series. Ann Transl Med 2017;5(Suppl 2):AB042. doi: 10.21037/atm.2017.s042