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Application of aCGH in stillbirth investigation

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Objective

To evaluate the advantage of aCGH to karyotype as stillbirth investigation.

Methods

Array comparative genomic hybridization (aCGH) was offered to patients in addition to conventional cytogenetics for any stillbirth cases from the year 2013 in prenatal diagnostic laboratory of Tsan Yuk Hospital in Hong Kong. Those who chose to have aCGH would have placental tissue, fetal skin biopsy and parental bloods sent to the laboratory. If aCGH was not requested, QF-PCR for common aneuploidies were performed for those samples not suitable for conventional cytogenetic study. The data from October 2013 to March 2018 was reviewed.

Results

There were 41 out of 146 patients who had stillbirth after 24 weeks of gestation and requested for aCGH in additional to karyotype. 39 had normal aCGH and 2 cases had 16p13. 11 duplication detected on aCGH. The yield was 100% for obtaining a result. Among the 146 cases of karyotyping, 13 (8. 9%) cases did not have report due to culture failure of the placental tissue or contamination. 4 out of these 13 (30. 8%) failed samples had aCGH showing normal result while 8 (61. 5%) out of these had QF-PCR performed for common aneuploidy and one mosaic Trisomy 21 was detected. Only one sample didn't have any result generated which means that, 92. 3% (12/13) of those samples with failed cytogenetic studies could have aCGH results.

Conclusion

ACGH is superior to karyotyping as an investigation for stillbirth because it can give a higher diagnostic yield. It could detect more abnormalities when compared to conventional cytogenetics. However it might also generate results with uncertain implication on stillbirth. Cost might limit its application. It might be an alternative to conventional cytogenetics.