APPENDIX O
RCOP GUIDELINES FOR AUTOPSY INVESTIGATION OF FETAL AND PERINATAL DEATH

All hospital post-mortem procedures are subject to parental consent that must not be exceeded. The following guidelines apply to an unrestricted post-mortem examination.

1. External examination

- body weight (to nearest gram, if less than 5kg)
- head circumference
- crown-heel and crown-rump lengths
- abdominal circumference
- foot length
- maceration (if baby is born dead)
- meconium staining
- full description (e.g. fontanelles, eyes, ears, nose, mouth and palate, digits, palmar creases, umbilicus and state of cord, genitalia, anus etc).
- dysmorphic features, congenital malformations and deformities
- other abnormalities

2. Internal examination

- comment on cranial, thoracic and abdominal cavities
- retention and fixation of the brain where practicable, subject to informed consent
- systematic description of major organs and tissues
- specific reference to ductus arteriosus and umbilical vessels
- weights of all major organs in digital balance (to 0.1g)
- comment on muscle and skeleton

3. Placenta

Placenta to be examined in all cases. A convenient method of ensuring the placenta is available in each case may be to send all placentas from babies admitted to the special care baby unit/neonatal intensive care unit to the pathology department. Whilst these need not be examined unless the baby dies, many departments would, in any case, consider it good practise to examine them.

- 3 dimensions
- trimmed weight
- umbilical cord (length, vessels, abnormalities)
- membranes (complete, incomplete, colour, abnormalities)
- fetal, maternal and cut surfaces

For further reference, please see: http://www.rcpa.edu.au/Library/Practising-Pathology/Macroscopic-Cut-Up/Specimen/Gynaecology-and-perinatal/Placenta

4. Histology

- at least one block of all major thoracic and abdominal organs (right and left lungs, heart, liver, kidney, thymus, adrenals and pancreas)
- costochondral junction (over 24 weeks' gestation)
- adequate sampling of brain (varies with case: minimum of one block from hind brain and one from cerebral hemispheres)
- adequate sampling of placenta (cord, membranes, focal lesions, grossly normal parenchyma to include amnion and decidua)

5. Chromosome analysis and genetic testing of the stillborn infant and placenta

If not previously performed antenatally via amniocentesis or other diagnostic fetal sample, a molecular karyotype (i.e. chromosomal microarray, CMA) should be performed for all stillborn infants[1-3]. CMA is preferred over routine conventional G-banded karyotype for two main reasons: (i) high success rate with CMA as cell culture is not required (87.4% successful analysis with CMA vs. 70.5% with karyotype)[2]; (ii) better diagnostic yield with CMA compared with conventional karyotype (8.8% vs. 6.5% detection rate for genetic abnormalities for antepartum stillbirths respectively)[2]. If additional DNA testing for single gene disorders (including metabolic conditions) is being considered, then a request for DNA storage can be made to the cytogenetic laboratory.

Suitable samples for CMA evaluation of the fetus include:

(i) **Fetal tissue** (e.g. cartilage from the patella or costchondral junction)

(ii) If consent for autopsy or fetal tissue collection has not been given, but cytogenetic testing is desired, then an **umbilical cord** sample (1cm segment taken from the placental end) or **placental biopsy** (1cm^3 block of tissue taken from the fetal side of the placenta) would be suitable.

6. Other special procedures and investigations

- X-ray ideally should be undertaken for suspected skeletal dysplasia and multiple malformations
- photography mandatory for dysmorphic fetuses and babies without ante-mortem diagnosis; advised for other gross abnormalities
- bacteriology (blood/spleen/lung/CSF), if clinically indicated
- virology, if clinically indicated
- storage of fibroblasts/frozen tissue/DNA, if clinically indicated
- biochemistry, if clinically indicated
- haematology, if clinically indicated
- neuropathology, if clinical or radiological evidence of CNS pathology or the brain appears abnormal on external examination

7. Autopsy reports

- demographic details
- date of autopsy
- details of consent and any restrictions

- availability of clinical records at time of post-mortem, including anomaly scans if relevant
- clinical history
- systematic description of external, internal and placental examination and results of X-rays and other ancillary investigations
- summary of major findings including sex and apparent gestation, estimated timing of death in babies born dead, adequacy of growth and nutrition, presence/absence of congenital abnormalities, major pathological lesions, evidence of chronic stress or disease prior to death, placental examination
- commentary addressing the clinical questions and significance of pathological findings
- mode/cause of death
- record of photographs and any samples retained
- record of disposal of any tissues or samples
- a provisional report on the macroscopic findings should be issued within 24-48 hours of the autopsy, with histology and further investigations including chromosome analysis incorporated into a final report when available
- timely dispatch to clinicians with particular reference to the timing of postnatal appointments

References

1. Reddy UM, Page GP, Saade GR. The role of DNA microarrays in the evaluation of fetal death. *Prenat Diagn* 2012; **32**(4): 371-5.

2. Reddy UM, Page GP, Saade GR, et al. Karyotype versus microarray testing for genetic abnormalities after stillbirth. *New England Journal of Medicine* 2012; **367**(23): 2185-93.

3. Rosenfeld JA, Tucker ME, Escobar LF, et al. Diagnostic utility of microarray testing in pregnancy loss. *Ultrasound in obstetrics & gynecology : the official journal of the International Society of Ultrasound in Obstetrics and Gynecology* 2015; **46**(4): 478-86.