

Canadian Association of Neuropathologists
L'Association Canadienne des Neuropathologistes

CASE No 5

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The mother of this 18.5 week gestation male foetus is 26 years old (G2P1). She has one daughter who is well. An ultrasound performed at 17 weeks gestation revealed a head size compatible with 15 weeks gestation compared to a femur length of 17.7 weeks, micrognathia, back slanted forehead, hypertelorism, extended knees, clenched hands, scoliosis and postaxial polydactyly of one foot. The pregnancy was electively terminated at 18.5 weeks gestation. The birth weight was 182 grams. Gross examination of the brain revealed a brain weight of 8.15 grams, cerebellar aplasia and agenesis of the corpus callosum.

Materials submitted: Haematoxylin and eosin stained section of the left cerebral hemisphere

Question: Diagnosis?