Necropsy findings in a child with FG syndrome

In 1983, Burn and Martin reported from this unit two male cousins with mental retardation, congenital hypotonia, intractable constipation, failure to thrive, and dysmorphic facies as possible examples of the X linked FG syndrome. Few pathological studies have been reported and we felt that it would be interesting to report the necropsy findings in the proband of Burn and Martin.

The brain. The brain weighed 800 g (normal for age 1100 g) and was brachycephalic. The convolutional pattern was simple with broader and fewer gyri than normal. A very large 'cystic' cavum septi pellucidi was present which communicated with the lateral and third ventricles through large fenestrations (figure). The corpus callosum was present throughout, but thin. Histologically, only acute anoxic changes and one isolated neuronal heterotopia in the cerebellar white matter were observed; no other evidence of migration defect was observed.

The septum pellucidum, which develops as a result of caudal growth of the corpus callosum, often shows a midline cleft-like space, called the cavum septi pellucidi, especially in fetuses and young children. Friede has suggested that a large cavum septi pellucidi may be a 'forme fruste' of agenesis of the corpus callosum and may be associated with a thin corpus callosum. This finding is therefore of interest, since case 2 reported by Opitz and

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Kaveggia\(^3\) was found at necropsy to have an absent posterior one-third of the corpus callosum, and case 4 reported by Thompson et al\(^4\) had complete absence of the corpus callosum on computerised brain scan.

The appearance of the brain in the necropsy photograph of the original case 1 who died accidentally\(^5\) is similar to the present case, having fewer and broader gyri than normal. Unlike our case, that patient was reported as having other evidence of a neuronal migration defect and megalencephaly. The brain was normal in necropsies of two other FG patients\(^6\) and showed non-specific subependymal infiltrates in one other.\(^5\)

Gastrointestinal. Histopathological examination of the colon, ileum, stomach, and oesophagus showed normal smooth muscle. Ganglion cells were present which reconfirmed that Hirschsprung’s disease was not the cause of the severe constipation.

Skeletal muscle (frozen 20 hours from death). The fibres of the skeletal muscle were generally small (9 to 13 \(\mu\)m in diameter; normal 18 to 26 \(\mu\)m). There was glycogen and lipid accumulation in muscle fibres, the former possibly relating to intravenous dextrose feeding. The muscle was otherwise normal. Histological examination of muscle has not been reported before in FG syndrome, but is important because of the hypotonia which may be severe. We reviewed the muscle specimens of case 1 reported by Thompson et al\(^4\). These were normal apart from general smallness of fibres. We conclude that it is most likely that the hypotonia in FG syndrome is central in origin.

Other organs were unremarkable, apart from the lung which showed changes of pneumonic consolidation.

The authors wish to thank Dr M H Webster and Dr J Burn for allowing us to report this patient. Dr D G F Harriman, Department of Pathology, University of Leeds, kindly supplied the original histological slides of the muscle of case 1 of Thompson et al\(^4\).\(^*\)

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References

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doi: 10.1136/jmg.23.4.372

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