

Letter to the Editor

Spinal segmental dysgenesis

To the Editor: I read with interest the article by Mahomed and Naidoo in the August 2009 issue of *SAJR*¹ of two exceptional cases of spinal segmental dysgenesis.

However, I believe that a normal corpus callosum has been misinterpreted as a developmental abnormality. Fig. 2(c) is furnished as proof of 'partial agenesis of the corpus callosum' in the first patient and that there is absence of the rostrum, genu and body whereas this image demonstrates a normal corpus callosum for a 2-week-old child. The terminology of the suggested condition is better termed 'hypogenesis of the corpus callosum'.² In hypogenesis the anterior portion (posterior genu and body) should be present (Fig. 1). In addition, whenever there is hypogenesis or agenesis, there is failure of inversion of the cingulate gyrus.

The normal cingulate gyrus follows a curved horizontal course similar to that of the corpus callosum. In absence of the corpus callosum the cingulate is not visualised on the sagittal image and gyri and sulci radiate at right angles to the CSF space ('sunburst appearance') (Fig. 1).³ This feature is used in neonates and in preterm children as an indication of the presence of the corpus callosum, because in early life the corpus callosum is very thin, isointense to cortex and therefore difficult to visualise. As children develop, more myelin is laid down, increasing the thickness of the corpus callosum and its signal on T1 (Fig. 2 a-c). Absence of the anterior portion of the corpus callosum with presence of the posterior portion only result from dysgenesis associated with holoprosencephaly and when there is a focal cortical infarct with fibres that cross through the anterior corpus callosum (local atrophy).

Other unanswered questions from the article are:

- Peripheral nerves exit the neural foramina below the reported termination of the cord on the sagittal image in case 1. Is this expected considering a criterion for diagnosis should be 'absent exiting nerve roots'?
- What are the unusual structures demonstrated on the axial T2 surrounded by fat? Is one of the structures a visible spinal cord? The cord is reported to terminate at T6 but the image is at the level of the isthmus of the horseshoe kidney which appears to be lower than T10 on the sagittal views.

Reports of rare abnormalities are necessary and the authors are commended for publishing these. However, expert opinion should be obtained prior to publication of complex abnormalities to avoid misdiagnosing normal anatomy as pathology. Normal paediatric brain developmental anatomy knowledge is critical before attempting to interpret complex abnormalities such as segmental spinal dysgenesis.

The legends should read:

- Fig. 1. There is hypogenesis of the corpus callosum. The components that are present are part of the genu and body. Note the partially inverted cingulate gyrus anteriorly while posteriorly the vertically oriented gyri and sulci radiate from the CSF space.
- Fig. 2 (a - c). Normal development of the corpus callosum in infancy. (a) A 2-day-old neonate demonstrates a very thin but present corpus callosum. This is confirmed by the everted cingulated gyrus lying horizontally separating the vertically oriented gyri from the CSF space. (b) At 2 months of age the corpus callosum itself becomes more distinct. (c) At 4 months of age the corpus callosum develops a thickness and high signal due to laying down of myelin.

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1. Mahomed N, Naidoo J. Spinal segmental dysgenesis. *South African Journal of Radiology* 2009; 6: 29-32.
2. Przybojewski S, Andronikou S. Corpus callosum hypogenesis vs. dysgenesis. *South African Journal of Radiology* 2006; 6: 24.
3. Barkovich AJ. *Pediatric Neuroimaging*. 3rd ed. Philadelphia: Lipincott Williams & Wilkins, 2000: 254-255, 323-327.

The authors of the SSD case report reply: We apologise for the incorrect terminology used, as the corpus callosum is in fact an immature corpus callosum. This observation was incidental and the article did not suggest that it was a feature of SSD. In response to Dr Andronikou's second question, the dysgenetic segment in Fig 2a is between T12 to L5, but we acknowledge that the words 'terminating at thoracic level 6', as in the article, might have caused this confusion.

It seems that our article has created much interest, and we would like to take this opportunity to elaborate further on this rare malformation, particularly to clarify its relationship with caudal regression syndrome (CRS).

SSD and CRS probably represent two faces of a single spectrum of segmental malformations of the spine and spinal cord. They differ from an embryological point of view, in the segmental location of the derangement along the longitudinal axis of the embryo. In SSD, the intermediate segment is involved as opposed to the caudal segment in CRS.

It is important to distinguish SSD from CRS and other spinal malformations as SSD is unlikely to benefit from untethering procedure, since the neurological disturbance is related to the congenital hypoplasia or absence of roots or segment of the spinal cord, rather than from cord tethering.¹

1. Tortori-Donati P, Fondelli MP, Rossi A, Raybaud CA, Cama A, Capra V. Segmental spinal dysgenesis: Neuro-radiologic findings with clinical and embryologic correlation. *Am J Neuroradiol* 1999; 20: 445-456.