Megacystis-microcolon intestinal hypoperistalsis syndrome: a rare type of functional intestinal obstruction in the newborn with characteristic alterations of smooth muscle proteins

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Introduction
Megacystis-microcolon intestinal hypoperistalsis syndrome (MMIHS) is a rare and severe type of functional intestinal obstruction in the newborn. This congenital disease is usually lethal. The etiology of MMIHS is not clear, but, although a genetic locus has not yet been identified, an autosomal recessive inheritance has been suggested. Previous studies report that reduction of contractile proteins may be responsible for the main abnormalities: functional intestinal obstruction and bladder distention.

Materials and methods
A prematurely born child that by antenatal ultrasonography showed distention of the urinary bladder presented one day after birth with intestinal obstruction. Laparotomy revealed microcolon and dilation of the small bowel. A subtotal resection of the small bowel with ileostomy was performed, sparing 35 cm of functional small intestine. Two months after birth, the ileostomy was removed and analyzed light microscopically and immunohistochemically with antibodies to protein gene product 9.5 (PGP 9.5), calponin, desmin, pan-actin and α-smooth muscle actin (α-SMA). Unfortunately, the child died one week after removal of the ileostomy.

Results
Conventional microscopy and immunohistochemistry of the perinatally resected small bowel specimen didn’t show any abnormalities. Conventional microscopy of the resected two months old ileostomy specimen showed normal mucosa (Fig. 1), muscularis mucosae (MM), submucosa and muscularis propria (MP). Immunohistochemistry for PGP 9.5 (Fig. 2) showed normal ganglionosis. Desmin was strongly expressed in the MM as well as the two layers of the MP (Fig. 3); a similar but weaker reaction was observed with Calponin (Fig. 4). Pan-actin showed a regular expression in the MM and in the longitudinal layer of the MP, but a reduced, dot-like reaction in the circular layer (Fig. 5). This odd pattern was even more evident with antibodies to α-SMA, showing absent expression in the circular layer of the MP (Fig. 6). Based on these findings, the diagnosis of MMIHS was made.

Discussion and conclusion
In this report of a child with MMIHS, we confirm previous findings of progressively reduced expression of contractile proteins in the wall of the small bowel in this fatal disease. Absence of a functional α3 subunit of the neuronal nicotinic acetylcholine receptor has been demonstrated in MMIHS. This has been related to the deletion of the proximal long arm of chromosome 15 (15q11). However, the exact cause of the loss of contractile proteins in the bowel and urinary bladder remain to be elucidated.

References