

**Title: Absent abdominal musculature in a girl**

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A 6-months old girl was referred for further management of her congenital malformations. These were identified antenatally and, at birth, she was noted to have wrinkled abdominal skin (Figure 1), consistent with the syndrome of absent abdominal musculature (Eagle Barrett syndrome). The typical appearance of the abdomen in these patients previously led to the descriptive term of “prune-belly syndrome”. Additional malformations included an omphalocele and urological abnormalities, including megacystis and a left megaureter. She received a vesicostomy.

Absent abdominal musculature occurs almost exclusively in boys, associated with cryptorchidism. The aetiology is unclear and has been speculated to be either a primary developmental defect affecting both urinary tract and abdominal wall or a secondary defect in abdominal wall development due to distension from massive urinary tract dilatation. The latter is consistent with the male predominance, as posterior urethral valves are the most common congenital form of severe urinary tract dilatation. Investigations into Mendelian disorders often help to elucidate aetiology. So far, genetic investigations in a single family have associated this syndrome with a homozygous nonsense variant in *CHRM3*, encoding the muscarinic acetylcholine receptor 3. *CHRM3* was found expressed in bladder muscle, but not the abdominal wall, consistent with the absent abdominal musculature being a secondary consequence of urinary tract dilatation. Genetic investigations in this girl revealed a *de novo* heterozygous variant in *KCNMA1*, previously reported as causative for Liang-Wang syndrome and consistent with her phenotype. *KCNMA1* is expressed in smooth musculature and its dysfunction may explain the megacystis and megaureter. This also argues for a secondary defect in abdominal wall development.

There is no specific treatment for absent abdominal musculature, although reconstructive surgery (abdominoplasty) can be performed to improve the cosmetic appearance. The consequently enhanced muscle tone may also support bladder emptying via Valsalva manoeuvre.

## Figures

### Figure 1: Image of the abdomen

Note the wrinkled skin, reflecting the absence of abdominal musculature