22q11.2 DELETION AND MENINGOMYELOCELE

The Spina Bifida Sequencing Consortium examined 715 parent-offspring trios. Their paper, which appeared in Science (1), reports six patients with 22q11.2 deletion and meningomyelocele (MM). They calculated that the deletion confers a 23-fold increased risk of MM compared with the general population. In a separate 22q11.2 deletion cohort the increased risk was of 12-15 fold. In mice, the knock-out of the *Crkl* gene, mapping in the minimal deletion interval, was sufficient to replicate neural tube defects, where both penetrance and expressivity were exacerbated by maternal folate deficiency.

In this context it is worth reading the article by Zamariolli et al. (2), titled "The impact of 22q11.2 copy-number variants on human traits in the general population", in which the authors report the results of their study on 405,324 unrelated participants of the UK Biobank.

1. https://www.science.org/doi/10.1126/science.adl1624
2. https://www.cell.com/ajhg/fulltext/S0002-9297(23)00005-8?_returnURL=https%3A%2F%2Flinkinghub.elsevier.com%2Fretrieve%2Fpii%2FS0002929723000058%3Fshowall%3Dtrue