Chromosome 21

Chromosome 21 in neurosurgery

- Polyamine biosynthesis dysregulation in Alzheimer's disease and Down syndrome cellular models
- Down Syndrome Biobank Consortium: A perspective
- Bladder and bowel dysfunction in Down syndrome with neural tube defect: case report and review of the literature
- Type-I-interferon signaling drives microglial dysfunction and senescence in human iPSC models of Down syndrome and Alzheimer's disease
- Aging with Down Syndrome-Where Are We Now and Where Are We Going?
- Astrocytes in Down Syndrome Across the Lifespan
- Human iPSC-derived Down syndrome astrocytes display genome-wide perturbations in gene expression, an altered adhesion profile, and increased cellular dynamics
- Charcot-Marie-Tooth type 4B2 demyelinating neuropathy in miniature Schnauzer dogs caused by a novel splicing SBF2 (MTMR13) genetic variant: a new spontaneous clinical model

---

Chromosome 21 is one of the 23 pairs of chromosomes in humans. Chromosome 21 is both the smallest human autosome and chromosome, with 48 million base pairs (the building material of DNA) representing about 1.5 percent of the total DNA in cells. Most people have two copies of chromosome 21, while those with three copies of chromosome 21 have Down syndrome, also called “trisomy 21”.

In Down syndrome children, functional constipation and lower urinary tract infections have been described, together with higher risk for incontinence and delayed sphincter control. At present, no clear association between Down syndrome, Bladder Bowel Dysfunction and neural tube defects has been previously described.

Onesimo et al. describe two female patients with Down syndrome presenting Bladder Bowel Dysfunction in association with neural tube defects, who both underwent personalized multidisciplinary intervention and pelvic floor rehabilitation, with good clinical outcomes.

At present, no screening program has been established in order to rule out neural tube defects or neurogenic urinary anomalies in Down syndrome patients presenting bowel and/or bladder dysfunction. In our opinion, presence of spinal abnormalities, despite rare, may be contribute to urinary symptoms and should be ruled out in patients presenting progressive or persistent Bladder Bowel Dysfunction. Early diagnosis and management of spinal cord defects associated with neurogenic urinary dysfunction may allow to prevent possible complications [1)]

1)

Onesimo R, Agazzi C, Massimi L, Giorgio V, Leoni C, Zampino G, Rendeli C. Bladder and bowel dysfunction in Down syndrome with neural tube defect: case report and review of the literature. Ital J Pediatr. 2023 Jul 20;49(1):89. doi: 10.1186/s13052-023-01412-z. PMID: 37475046.