INCREDIBLE GENOME

DMD THE GREAT

have a nice graph

5′

3′



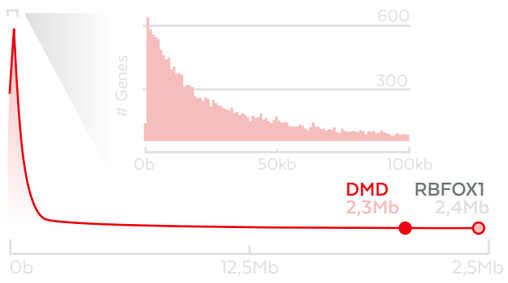
# *DMD, the great*

EXON

0b 1kb 2kb 3kb

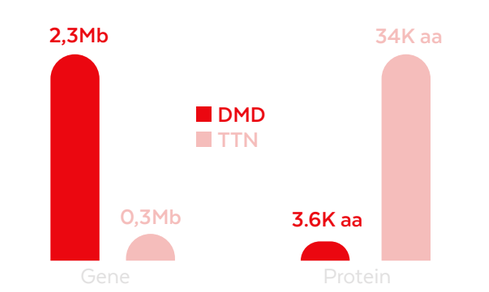
One of the biggest human genes, DMD gene encodes dystrophin, a cytoskeletal protein which bridges the inner cytoskeleton and the extracellular matrix. Of its remarkably 2.3 Mb only 11.3 kb (0.05%) corresponds to the coding region distributed into 79 exons. Due to the length and huge intron content, the DMD gene have been a import model for understanding transcription and splicing mechanisms. Localized in the chromosome X, deleterious germline mutations in DMD gene are responsible for a series of diseases including Duchenne muscular dystrophy (DMD), a X-linked syndrome with still poor outcome. Genetic therapies to restore dystrophin function (e.g. genome editing) are still in development.

## BIG BROTHER




In Gencode version 38, only 3 genes are bigger than DMD: (1) a RNA binding protein RBFOX1 with 2,47Mb, (2) a neurexin protein CNTNAP2 with 2,3 Mb and (3) a tyrosine phosphatase protein PTPRD with 2,29 Mb.

## NOT THE LARGEST PROTEIN




Due to the huge amount of intronic region, the DMD gene doesn't encode a to as big protein (3,685 amino acids). In the other hand, the TTN gene have the largest amount of exons (363!), the longest human exon (17kb!!) and encode the biggest human protein (34,350 amino acids!!!)

## WHY ARE YOU TAKING SO LONG?


Transcribing such a enormous amount of bases takes time. The first estimate time to RNA polymerase II do the task was 16 hours, but more recent studies about the polymerase processivity estimate a 10 hours journey from beginning to the end of DMD gene.



Rodrigo Barreiro 2021 / version 1.0 / Sources: Most gene metrics were retrieved from Gencode v38 annotation gtf file. DMD and TTN GeneCards (genecards.org) were used for protein metrics and structure. Articles: Duchenne muscular dystrophy. Nat Rev Dis Primers 7, 14 (2021). https://doi.org/10.1038/s41572-021-00255-4; Tuffery-Giraud, S., Miro, J., Koenig, M. et al. Normal and altered pre-mRNA processing in the DMD gene. Hum Genet 136, 1155–1172 (2017). https://doi.org/10.1007/s00439-017-1820-9; Plots were made in R, ggplot package and refined in Affinity Designer.