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Trisomy 21 Is Responsible For Disorder Down Syndrome

Patrizia Messana*

Department of Obstetrics & Gynaecological Nursing, Akdeniz University, Antalya, Turkey

***Corresponding author:** Patrizia Messana, Department of Obstetrics & Gynaecological Nursing, Akdeniz University, Antalya, Turkey, E-mail: messanapatrizia@gmail.com

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Description

Down Syndrome driven by an additional duplicate of chromosome 21, and delicate X disorder, driven by loss of the RNA-restricting protein FMRP, are two normal hereditary reasons for scholarly incapacity and mental imbalance. We hypothesize that DS and FXS may share underlying mechanisms based on the number of DS-implicated transcripts bound by FMRP. We find that DS and FXS human pluripotent stem cell models and glutamatergic neuron models both exhibit overlapping transcriptional perturbations and increased protein expression of specific targets. Based on these findings, specific molecular perturbations that are shared by DS and FXS can be used to prioritize targets; additionally, they demonstrate the functional relevance of previous associations between disease-implicated genes and FMRP targets. Trisomy 21 is responsible for the common congenital disorder Down Syndrome (DS).The prevalence and life expectancy of DS individuals have significantly increased as a result of advances in medical treatment for fatal complications in early childhood and an increase in maternal age with population aging.

Life Expectancy of Down Syndrome

Despite this rise in the number of adults with DS, little is known about their hematological condition. Leukopenia, macrocytosis, and thrombocytopenia are the three hematological abnormalities that we present here as adult DS-associated features. Notably, as one got older, these features became more noticeable. Multiple aging-related disorders in the general population would benefit from new research perspectives and a deeper comprehension if further analyses were conducted on adults with DS. This study aims to understand how mothers in Turkey construct their motherhood in the context of personal growth despite the emotional and social challenges they face and how a Down syndrome diagnosis affects their experiences. In this study, grounded theory was used. Mothers were interviewed in depth, and the data were gathered and analyzed using a constant comparative method that was systematic and hierarchical. Additionally, three primary categories emerged: re-creation of motherhood; factors that influence the procedure; and a response to a life that is changing. The mothers described personal development as stronger feelings, improved communication skills, and becoming closer to God. To learn more about the unique experiences and personal development processes of Down syndrome moms, more cultural research is needed. Brain abnormalities in Down Syndrome (DS) have been shown by neuroimaging studies, but the underlying causes of the dysfunction have not been identified. Seed-based Functional Connectivity (FC) with the significant DC clusters was used to compare the DS group's degree centrality abnormalities to those of the control group.

In addition, we identified differences between the brain networks of DS and controls using seed-based FC and significant DC clusters. Over the past few decades, the field of prenatal genetic testing has undergone significant change, and its social, legal, and ethical repercussions have been the subject of extensive debate. In the past, both Germany and Israel were known for having very different laws and how professionals and laypeople felt about genetic testing. This study examines the interaction between lived experience and cultural scripts and their impact on the formation of personal views regarding disability and prenatal testing. Based on qualitative analysis of 37 semi-structured interviews, it compares disability activists from Germany and Israel. Despite the emergence of new technologies, we have discovered that the differences between Germany and Israel persist, and that family members and disability activists reflect the norms of their sociocultural environments, highlighting the role society plays in shaping the perspectives of those who have direct experience of disability.

Alzheimer's Disease Neuropathology

We describe the dynamic changes in the cellular and humoral responses of 41 DS patients after receiving a booster vaccine dose following an initial study on a cohort of adults with DS one to three and six months after receiving a two-dose SARS-CoV-2 vaccination regimen. An unanticipated and startling phenomenon has emerged as a result of advancements in medicine and an increase in longevity. People with Down syndrome are more likely to develop Alzheimer's disease as they get older. Additionally, it tends to appear earlier in them than in typical cases. Infection with viral, bacterial, or parasitic microorganisms such as the Herpes Simplex Virus and Toxoplasma gondii, among others may also be the underlying cause of Alzheimer's disease, according to increasing evidence. Although Trisomy 21is widely accepted as the cause of Down syndrome, its underlying cause is unknown. I propose that an

Alzheimer's-capable microbe is the cause of Down syndrome itself, which in some susceptible individuals is followed by Alzheimer's disease, and that the two conditions progress simultaneously. Trisomy 21, a genetic mutation that also increases a person's likelihood of developing Alzheimer's disease neuropathology amyloid plaque and neurofibrillary tangle formation in later life, is the cause of Down's syndrome.

Using support vector regression, we wanted to see how well diffusion-weighted imaging and connectomic modeling could predict brain amyloid plaque burden, baseline cognition, and longitudinal cognitive change. Members with Down's disorder effectively finished a full Pittsburgh Compound B PET-MR convention and memory evaluation at two timepoints. Based on the structural connectome, we found that graph theory metrics of node degree and strength can accurately predict global amyloid deposition. Additionally, we demonstrate that the structural network's baseline connection density is a promising indicator of future cognitive performance. The main directional effects were significant decreases in white matter connectivity in relation to both higher rates of cognitive decline and PiB status. These results show that non-invasively evaluating the prognosis for Alzheimer's disease makes use of machine learning techniques and that white matter plays a crucial role in the progression of neuropathology.