the cases had a normal NIPT thus avoiding an invasive procedure. With the highest detection and lowest false-positive rates, cell-free DNA screening test proved to be a reliable prenatal test for chromosomal abnormalities with a positive outcome if applied to the general obstetric population.

P35 – MULTI-OMICS INTEGRATIVE PATHWAY ANALYSIS IN HEAD AND NECK SQUAMOUS CELL CARCINOMA

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Context: Head and Neck Squamous Cell Carcinoma (HNSCC) is an aggressive disease that arises by molecular deregulation events including the accumulation of copy number alterations and changes in methylation profiles that result in modifications in gene expression levels and downstream signalling pathways. The study of these alterations is essential to understand the development and progression of HNSCC.

Methods: Copy number alteration (CNA), RNA-Seq and methylation data from tumor tissue from 416 HNSCC patients was downloaded from The Cancer Genome Atlas (TCGA). mRNA expression data for normal tumor- adjacent tissue was also retrieved. The genes contained in the regions altered in more than 40% of patients were extracted, for RNA-Seq data only the genes differentially expressed between tumour and normal tumor-adjacent tissue were used. A threshold of 0.3 was set for selecting methylated genes and those methylated in under 40% of patients were filtered out. The overrepresented pathways in each dataset were determined and those that were statistically significant ($p < 0.01$) and the respective genes altered in the cohort were analysed and compared between omics. R programming language was used for analysis.

Results: The top 3 most overrepresented signalling pathways were Pathways in Cancer, PI3K-Akt signaling pathway and Metabolic pathways when considering CNA data; Neuroactive ligand-receptor interaction, Olfactory transduction and Staphylococcus aureus infection in methylation data and Metabolic pathways, Axon Guidance and Cytokine-citokine receptor interaction when considering the RNA-Seq data. Two genes (*ADCY8* and *AGTR1*) were common to both CNA and methylation data pathways and *EGFR* was common to the latter and RNA-Seq data. For CNA and RNA-Seq data, a set of 8 genes was found to be common to the overrepresented signalling pathways.

Conclusion: This is a preliminary study that corroborates the complexity of molecular interactions that disrupt essential signalling pathways and give origin to HNSCC. The integration of multiple omics in the study of cancer is a hot topic that must be further explored in order to positively impact the clinical course of HNSCC patients.

P36 – THE PSYCHOSOCIAL EXPERIENCE OF MEMBERS OF FAMILIES WITH HEREDITARY AMYLOID TRANSTHYRETIN AMYLOIDOSIS WITH POLYNEUROPATHY: PRELIMINARY RESULTS OF A MIXED-METHODS SYSTEMATIC REVIEW

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Introduction: Hereditary amyloid transthyretin amyloidosis with polyneuropathy, besides its chronicity and devastating progression, provokes a strong psychological impact on the life of these patients and their relatives. Thereby, genetic counsellors, psychologists and other health professionals are challenged to work together for the best possible care of those families. Aiming at promoting the development and delivery of clinically supportive services, we conducted a mixed-methods systematic review about the psychosocial experience of members of families with this condition.

Methodology: Manuscripts published between January 1992 and December 2019 were searched using 16 databases. The work includes a methodological quality assessment of selected studies, a postsynthesis sensitivity analysis, and an overall assessment of the thematic synthesis.

Results: Of 7,394 manuscripts identified, 220 were reviewed in full text and 70 met the eligibility criteria. Preliminary findings of the reviewed studies suggest that the disorder and its life implications may pose a significant psychosocial burden for the patients and their relatives. During their lifetime, members of these families generally became caregivers, implying changes in family roles, and parent's disease and death are frequent early in their life.

Discussion: Psychosocial experience of members of families with this disorder is not enough studied. Although scientific literature has described the life paths of these persons, further research on other key disease variables (e.g., including the psychosocial experience based on timing of clinical onset in the life cycle and effects of the most recent treatment interventions) can help fill research gaps and optimize health care services that support the families with the condition.

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