using Zytovision dual-probe according to the manufacturer's protocol. The technique applied to manage the tissue was the fixation with formalin embedded in paraffin. The statistical analysis was performed through the Microsoft Office Excell 2016 and IBM SPSS Statistics version 23. It was analysed a sample of 108 patients, with median age of 60 ± 12 years (minimum and maximum ages was 30 years and 85 years, respectively), 95.3% of whom were female (103 patients) and 4.6% were male (5 patients). HER2 was positive in 20.4% (22 patients), equivocal for 10.2% and negative for 69.4% (75 patients). The medium time since the sample was received in the laboratory till the results were 6 ± 4days. The rate of HER2 positive in this study is accordingly to the literature. It may be too early to evaluate the impact of the HER2 status determination in the district population of the hospital. Accurate testing is extremely important since anti-HER2 target therapy has shown a great impact in the overall and disease free survival. Therefore the population in need, regardless where they are from, should have access to Genetic Laboratories.

_ Clinical Research

P28| A Portuguese Tool for Quality Assessment of Genetic Counselling by Genetics Healthcare Professionals

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Recent studies on patients' and professionals' views in Portugal highlighted a need for instruments and quality indicators of genetic counselling practice. In response, a novel tool was developed using the Reciprocal-Engagement Model (REM) as a theoretical-practical foundation as well as evidence-based insights from national research. After pre-test validation, the scale was submitted to psychometric validation, we used a sample of 30 participants that were mainly medical geneticists, who evaluated 81 counseling sessions, carried out at main national services between January and April 2017. Based on empirical and statistical criteria the best items were selected. The final 50 items-version comprises five dimensions: education, the counselees' characteristics as part of the process, relationship between counselor and counselee, potential effects of the process on the counselee, and services provision. Results also showed consistent psychometric properties of the scale, which was supported on theoretical and practice concepts of genetic counselling. The professionals involved in the validation process, highlighted as very relevant for assessment of their practice the association of each genetic counselling principle with specific goals, strategies and behaviors, in the REM model and, accordingly, underlying the structure of the new instrument. The constructed scale is a pioneer tool in Portugal and perhaps the first practical application of the REM in the context of genetic services in Europe. Research on quality assessment of genetic counseling practice, using the Portuguese new scale as the measure instrument, will in turn inform the applicability of the REM to our national context and others. With this study, we would like to raise the discussion on how relevant this new tool can be for further investigation on genetic counselling field and its potential impact in the improvement of genetic services in our country.

P29| An algorithm for the detection of common copy number alterations in cancer

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Copy number alterations (CNAs) are critical for cancer origination and progression. Recurrent CNAs - regions that have a high enough probability to be altered in at least some subjects of the cohort - are determinant to the detection of driver alterations, since those will be present in a considerable amount of the analyzed genomes and are disease-specific as opposed to random subject-specific alterations – passenger alterations. The disease-specific genetic signature, derived from the common CNAs, can then be defined as a function of the probability of alteration for a given region. A method for the determination of recurrent CNAs and the underlying probability distribution of the cancer in study, was developed. The algorithm partitions a dataset of array CGH or SNP array segmented profiles, by chromosome, recovering the overlapping regions, their break-points and probability of alteration. In order to test this algorithm, simulated datasets with known properties were generated. CNA data from three cancer types, obtained by array CGH, was downloaded from The Cancer Genome Atlas (TCGA) and subjected to the algorithm. All analyses were performed using R and Matlab. The algorithm performed well for 1000 tested simulated datasets, retrieving correctly both the regions' breakpoints and their probability of alteration. The error for that probability was found to decrease as the number of subjects in a cohort increased. The algorithm performed well in real datasets, retrieving correctly the most frequently altered regions and was successfully used to compare between groups established within the cohorts. The study of copy number alterations is crucial to understanding the development and progression of several conditions, the most prominent of those being cancer. Reducing considerably the number of regions to analyze as well as generating more structured data is essential to reduce noise on the complex datasets generated by genome-wide technologies.

P30| Prevalence of X-aneuploidies and X-structural abnormalities in a Portuguese population with primary amenorrhea or premature ovarian insufficiency

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Amenorrhea affects 1–3%of women of the reproductive age. Primary amenorrhea(PA) is defined as the absence of menarche in