(MGUS), they can evolve to Multiple Myeloma (MM), a malignant plasma cells neoplasm, that still remains with no cure. A good establishment of MM risk stage is mandatory and genetic analysis has been proposed as a good strategy for a more accurate prognosis. Interphase Fluorescence *in Situ* Hybridization (i-FISH) is the only technique applied in the clinical practice for MG patients' genetic analysis, even though it seems not to be enough. Therefore, the aims of this project were the genomic and molecular characterization of MG patients and the validation of Array Comparative Genomic Hybridization (aCGH) and Multiplex Ligation-dependent Probe Amplification (MLPA) technologies in the MG patients' prognosis. A total of 13 patients were studied (3 patients with MGUS, 5 patients with MM and 5 patients with relapsed MM). Plasma cells ($CD138^+$) from bone marrow samples were isolated through immunomagnetic separation, being then selected to perform aCGH and/or MLPA. From the 10 studied samples by aCGH, 7 presented 13q deletions, 5 presented 1q gain, trisomy 9 was detected in 4 samples and 14q deletions were detected in 4 samples. MLPA technology only partially confirmed the genetic alterations detected by aCGH. Furthermore, it allowed the study of other 3 samples which were not possible to be analysed by aCGH. This pilot study contributed to the unveiling of the genomic alterations behind MG in the studied cohort, thus allowing the first steps in the possible validation of aCGH and/or MLPA as complementary techniques to i-FISH in the clinical practice. The study of more cases is needed to confirm the usefulness of these approaches.

**P55| Patients' views: a new tool for quality assessment of genetic counselling**

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It is consensual that appropriate genetic counselling is essential when a genetic test is offered or where there is a risk for a genetic condition. Assessing quality of genetic counselling, however, is a challenge for genetic services worldwide, given the scarcity of effective tools available to this effect. Recent studies in Portugal reinforced precisely this lack of tools, as well as the need to define quality indicators that support them. A pioneer instrument for quality assessment of genetic counselling practice by professionals has been developed. Currently, we started the construction of an analogous tool designed for quality assessment from the consultand's perspective. Here, we present the methodological design and preliminary results of the development and the validation process of this new tool. The proposed scale was submitted to pre-test validation with 7 consultands at CGPP, between June and August of the current year. Cognitive interviews with 5 of these participants were performed to explore in-depth the adequacy of the items, instructions and scale options. All main national genetic services were invited to collaborate in the recruitment of a minimum sample of 120 patients, for the validation process. This study was approved by the FPCEUP Ethics Committee. Based on the above mentioned procedures, the present version of the scale has 52 items, organized in 5 dimensions focusing on: (1) relevance of the genetic information; (2) the way consultand's emotional issues and personal characteristics were addressed; (3) the relationship and communication issues; (4) genetic counselling outcomes; and (5) services provision. The process of psychometric validation started last September and should be completed next May. In the short-term, it is expected that this tool will allow an evaluation of genetic consultations and services from the users standpoint, as well as a comprehensive understanding of the genetic counselling process, where both the professionals' and the consultands' views can be compared.

**_Clinical Case reports**

**P56| *TBL1XR1* single gene CNV: copy loss in mother and daughter with ID and ADHD**

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The genetic diagnosis of intellectual disability (ID) has witnessed a major breakthrough in the past decade with the routine use of microarray technology. While many ID related genes are now uncovered, others are still in need of more data to support a stronger clinical delineation. We report a six year old girl referred to our genetics department for ID, behavioral problems and epilepsy. She also presented attention deficit hyperactivity disorder (ADHD), and clinical observation revealed unspecific facial dysmorphic features. Array-CGH analysis detected a 1.22 Mb deletion, on 3q26.32, encompassing *TBL1XR1* gene (PerkinElmer GGX-HD 180k, Genoglyphix v3.1). Subsequent FISH studies established that the variant was inherited from the affected mother and was absent in her maternal grandmother and two maternal uncles, who have no phenotype. *TBL1XR1* gene encodes a protein that is part of both nuclear receptor corepressor (N-CoR) and histone deacetylase 3 (HDAC3) complexes, and plays an essential role in transcriptional activation mediated by nuclear receptors. Point mutations in this gene have been previously related to autism and Pierpont Syndrome while microdeletions were shown to cause syndromic ID. However, there are only a few reported patients. In our case, this single gene CNV segregating with the ID phenotype in the family, provides further evidence of *TBL1XR1* haploinsufficiency and adds to the characterization of the associated phenotype.

**P57| Interstitial 6q22.1-q22.31 microdeletion: narrowing the critical region for ID/DD and movement disorders**

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