10q22.2q22.3. Karyotyping subsequently revealed a supernumerary ring marker chromosome. To date only eighty-eight sSMC derived from chromosome 1 have been reported. Although the phenotype is very variable, ranging from normality to severe intellectual disability (ID), some correlations have been drawn: the region 1p12 to 1q12 appears to be non-dosage dependant, and the size of the sSMC seems to correlate with severity. This case reinforces the finding that near-centromere partial trisomy 1 results frequently in dysmorphisms, ID and hypotonia. The resemblance to KS was interestingly reported 20 years ago in a patient presenting an interstitial duplication of the short arm of chromosome 1, with significant overlap to ours. More patients are needed to allow possible candidate genes to be suggested. No clinical significance could be attributed to dup 10q22.2q22.3. In conclusion, we illustrate a distinctive phenotype of a rare chromosome abnormality, while emphasizing the importance of resorting to complementary classical cytogenetic studies.

P63| Renpenning syndrome: a rare syndrome in two Portuguese patients

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Renpenning syndrome (MIM #309500) is a rare X-linked recessive disorder caused by *PQBP1* mutations. This gene encodes a protein predominantly expressed in the central nervous system during development, playing an important role in neurodevelopment and neuronal functions. The syndrome is characterized by intellectual disability, leanness, microcephaly, short stature (relative to familial target measurements) and dysmorphisms. We report two boys, who are the first and only children of two non-consanguineous couples and single cases in the families. Both have developmental delay, prenatal microcephaly, leanness, congenital heart defect and non-specific dysmorphisms. A next-generation sequencing panel of 6110 genes was performed and the pathogenic *PQBP1* (NM_005710.2) c.459_462del (p.Arg153Serfs*41) variant was identified in both patients in hemizygosity and was proven to be inherited in both. This variant is one of three recurrent variants that occur in an AG hexamer in *PQBP1* exon 4. It has been proposed a gain-of-function mechanism for this 4 bp deletion, namely that the mutant protein binds preferentially to non-phosphorylated FMRP through a new C-terminal epitope and promotes its ubiquitin-mediated degradation, causing synaptic dysfunction. The phenotypes of both patients were in concordance with the literature, even though one of the cases did not have short stature relative to familial target measurements. However, the dysmorphic features were not considered to be recognizable. Furthermore, both boys were single cases in the families, which made the diagnosis of an X-linked intellectual disability more challenging. The availability of new technologies in genetics allowed an accurate genetic counseling of the families, namely the identification of healthy female carriers, who have a 25% risk of having an affected child.

P64| Psychosocial experiences of young adults at risk for transthyretin familial amyloid polyneuropathy: early versus late-onset Portuguese case report

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Transthyretin familial amyloid polyneuropathy (FAP) has been characterized by its early-onset (before 40 years) in Portugal and a few studies have discussed the psychosocial impact of the disease in our population. Recently, more late-onset (after 50 years) Portuguese cases have been described. Patients with this particular later onset are frequently probands of their families, pointing out an unexpected family attribute and making more complex its management. Family members may experience greater difficulties in adapting to this new and severe condition and its genetic risk. Research on individual and family psychosocial experience of this particular form of FAP is not yet available. We sought to identify psychosocial effects of life paths related to the disease pattern. The present case report explored psychosocial experiences according to the familial pattern of disease onset. We describe two clinical cases of Portuguese young adults with genetic risk for early- and late-onset FAP, respectively. After written consent, semi-structured interviews were conducted, recorded and analysed using thematic analysis. The first case was a 23 years old female who had an extended period and a close experience with the disease in her relatives. The participant was aware of the consequences of FAP for her future life plan, if she was a carrier. The second case was a 23 years old male who did not know much about the disease. After learning about two months ago that his mother has late-onset FAP (without history of symptoms), he had no expectations about the consequences of the disease for his life. This case report is the commencement of a large research project on this topic. First insights suggest that some specific issues related with the familial pattern of disease onset may have a role in the psychological experience of at-risk subjects that perform presymptomatic testing. Personal experience with FAP seems to influence the psychosocial impact of presymptomatic testing, making the management of the disease more challenging by the families with late-onset FAP. This preliminary report may have some implications for practice in the context of genetic counselling.

P65| Recombinant chromosome derived from two independent translocations of the same maternal homologues: Incidental finding in a fetus

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Complex chromosome rearrangements (CCR) account for a very small number of cases described in the literature. It is very rare that both homologues of the same chromosome pair are