presented short stature and had no dysmorphic features. He graduated high school with a professional diploma, with educational support but no learning difficulties. His psychological assessment showed no cognitive deficit.

Results: Microarray analysis identified a 1.19 Mb interstitial deletion in 6p25.3p25.2 involving three genes, including FOXC1, as well as a 1.39 Mb copy gain in chromosome 17q12 encompassing 14 OMIM genes, including LHX1 and HNF1B. FISH studies revealed that the deletion was de novo and the duplication was inherited in tandem from his father.

Discussion: To date, only a few cases of small 6p25 deletions overlapping our case have been described in the literature. This deletion explains the patient´s eye phenotype as FOXC1 gene is associated with anterior segment developmental anomalies of the eye.

17q12 duplication can explain the family's neurodevelopmental phenotype. The absence of DD/ID in our patient may be related to the incomplete penetrance of these features in both syndromes and a smaller number of genes involved in comparison to other cases. Targeted educational intervention has probably contributed to the successful academic outcome.

P78 - REPORT OF A RARE 3Q29 INTERSTITIAL MICRODELETION: PRENATAL DIAGNOSIS AND POSTNATAL FOLLOW-UP

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Distal interstitial deletions in the 3q29 region are rare. The characterization of new prenatal diagnosis (PND) cases and their follow-up may add knowledge about the affected region.

A 21-year-old woman was referred for PND at 20 weeks of gestation due to fetal increased nuchal translucency, cystic renal dysplasia and hyperechogenic focus on the left ventricle. A microarray (CMA) study was performed using the CytoScan 750K (Thermo Fischer®), after a normal result in the rapid aneuploidies diagnosis by QF-PCR (Devyser®).

CMA identified a ~594 Kb interstitial deletion at 3q29 - arr[GRCh37] 3q29 (196190768-196784544)x1, in a XX fetus; a similar deletion was identified in the mother. This is located into the described 3q29 recurrent deletion syndrome region that spans 1.6 Mb. The pregnancy continued after genetic counseling, and the term newborn presented an Apgar score of 10/10.

The identified microdeletion encompasses several genes, including 3 OMIM morbid genes, RNF168, NRROS, and CEP19, associated with autosomal recessive heredity. It is not possible to detect prenatal characteristics associated with them. The deletion may be considered of uncertain clinical significance, possibly pathogenic.

Regarding prenatal age, two reports described deletions partially overlapping with the present one: a case of a de novo deletion and another with maternal inheritance. Both presented intrauterine growth restriction and the first had ventricular septal defect (VSD).

The 3q29 recurrent microdeletions detected in PND are also rare, presenting reduced birth weight, with VSD occurring only in isolated cases. The pregnancy was uneventful in most cases.

In the present case, normal cardiac evaluation and psychomotor development were found in the neonatal and postnatal periods; there were no facial dysmorphisms or morphological changes. Only a small left kidney and the presence of cortical cystic formations were observed and are being monitored by pediatric nephrology. At the 12-month appointment, the child presented normal psychomotor development.

Amongst the cases reported with microdeletions in 3q29, the presence and severity of symptoms are variable, and the penetrance is incomplete. Even in inherited deletions phenotypic variability has been reported, and thus, inheritance may not be the most relevant factor to predict the phenotype.

The case presented brings an addition to the rare data in prenatal age and reinforces the knowledge about distal interstitial microdeletions in the 3q29 region in prenatal age and in the neonatal and postnatal periods.

P79 - FAMILIAL ADENOMATOUS POLYPOSIS (FAP) - PREVALENCE OF PATHOGENIC VARIANTS USING A NEW MULTIGENE PANEL: RETROSPECTIVE REVIEW IN A SYNLAB LABORATORY CENTER

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Introduction: Colorectal carcinoma (CRC) is one of the most common neoplasms worldwide, and there are several genetic syndromes that predispose to CRC. Some of these syndromes are also characterized by the development of many (tens to thousands) polyps in the rectum and colon. Genetic analysis by next generation sequencing has become increasingly available and cheaper, and we, therefore, designed a new multigene panel, which includes several genes associated with hereditary polyposis syndromes, allowing for a more precise diagnosis.

Methods: To determine the pathogenic/likely pathogenic (P/LP) variants prevalence in intestinal polyposis cases, we reviewed the results of 226 cases tested at our center. All samples were tested by a multigene panel, that included the following genes: APC, BMPR1A, MSH3, MUTYH, NTHL1, POLE, POLD1, PTEN, RNF43, SMAD4 and STK11. We divided the cases in two subgroups: patients with personal and family history of polyposis and patients with polyposis, but no family history of polyposis. NGS was performed on Ilumina Platform, using the Twist Human Comprehensive Exome (Twist Bioscience).

Results: Our study revealed 23 P/LP variants in all analysed genes (23/226=10%). However, we noticed that in the subgroup of patients with personal and family history of polyposis (n=9) the prevalence is much higher (22%) than in the other subgroup (n=216; prevalence of 9%).

Most of the P/LP variants (82%) detected in these patients were in the APC and MUTYH genes. The remaining variants (18%) were detected in the NTHL1, POLE, PTEN and SMAD4 genes.

Discussion: This retrospective study demonstrated that APC and MUTYH genes continue to be the main genes associated with polyposis. Since in almost a fifth of the cases the underlying genetic cause was due to P/LP variants in other genes, the use of a polyposis multigene panel should be the standard testing approach. This approach will result in a higher diagnosis rate and more tailored surveillance strategy.

P80 - MUTATION PATTERN IN PORTUGUESE PATIENTS WITH HEREDITARY AND SPORADIC BREAST AND OVARIAN CANCER. EXPERIENCE OF A SYNLAB LABORATORY CENTER

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Introduction: Next-generation sequencing (NGS) allows for a more cost-effective and quickly detection of pathogenic and likely pathogenic variants. Identification of these variants in breast and ovarian cancer allows for increased clinical surveillance, early detection and surgical decision and predicts the response to poly (ADP-ribose) polymerase (PARP) inhibitors.

Methods: To determine the pathogenic/likely pathogenic (P/LP) variants prevalence in HBOC and SBOC cases, we performed a retrospective review of 1038 samples received at the Synlab laboratory of Porto center between January 2022 and August 2023. All samples were tested by