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Case report

A Thirty-Seven-Year Follow-Up of Peutz–Jeghers Syndrome across Three Generations

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SUMMARY

Peutz-Jeghers syndrome (PJS) is a rare genetic disorder with autosomal-dominant pleiotropic inheritance, variable penetrance and characteristic signs of the disease that predisposes persons to increased risk of developing cancer, particularly in the gastrointestinal tract and the breast. Due to genetic nature of disease, in the familial Peutz–Jeghers syndrome, a multiplication of symptoms in the three-generation family members was established.

This paper represents an insight into the anamnesis of PJS in one family over thirty-seven years of follow-up, and is part of the broader study of this disorder. Article presents family history, clinical and histological findings and multiplication of symptoms of PJS across three generations.

Over thirty-seven years, PJS has been present in this family in the form of only mucocutaneous pigmentation but without clinically manifested signs (father), or with both melanine hyperpigmentation and gastrointestinal hamartomatous polyposis (his daughter and her son).

The symptoms rose suspicion of the existence of PJS complication, i.e. carcinoid-like syndrome with watery diarrheas accompanied by constipations in the affected mother and son who were surgically treated. Diagnosis of PJS was histopathologically confirmed in both cases: the presence of the polyps with hamartomatous pattern and conspicuous hyperplasia of chromogranin-positive (EC and L cells) and serotonin-positive (EC) cells. Malignant transformation of PJ- removed polyps was not found. Besides hamartoma, polyps as well as a tubular adenoma were found, with a low degree dysplasia without malignant transformation (son).

The authors discuss the findings in relation to the important role of the gastrointestinal endocrine cell hyperplasia, not only for better understanding of the growth and clinical symptoms of the PJ polyposis, but also for new approach and the possible application of anti-hormonal therapy in the treatment of these patients in the future, that is not currently in use.

Key words: **Peutz- Jeghers polyposis, gastrointestinal endocrine cell hyperplasia, immunohistochemistry**

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