[Turcot syndrome--a rare extra-intestinal manifestation of familial adenomatous polyposis?]

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The case of a 15-year-old male with Turcot syndrome is presented. When the patient was aged 10 years a medulloblastoma was diagnosed. Five years later he developed multiple adenomatous polyps of the colon and multiple "congenital hypertrophy of the retina" (CHRPE), the most common extraintestinal manifestation of FAP, were described. Family history revealed familial adenomatous polyposis with 12 family members exhibiting a FAP. The mode of inheritance of Turcot syndrome is controverted. Our case strengthens the hypothesis that the syndrome is a further extraintestinal (rare) manifestation of the FAP gene.

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