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abstract

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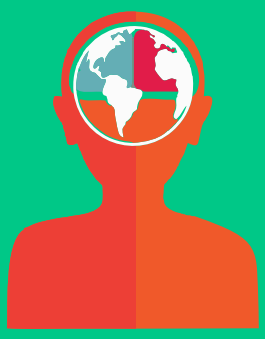

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abstract

Coexistence of GH-Secreting Pituitary Macroadenoma, Mayer–Rokitansky–Küster–Hauser Syndrome, and Neurofibromatosis Type 1 in an Adolescent Female: A Rare Clinical Triad

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Introduction: Growth hormone–secreting pituitary macroadenomas result in excessive IGF-1 levels and cause acromegaly or gigantism. Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a congenital Müllerian anomaly seen in 46XX females, characterized by agenesis of the uterus and upper vagina. Neurofibromatosis Type 1 (NF1) is a multisystem disorder marked by café-au-lait macules and neurofibromas due to NF1 gene mutations. Although rare associations between these disorders have been documented in pairs, their simultaneous occurrence has not been reported to date. To our knowledge, this is the first documented case of their coexistence in a single patient.

Methodology: Written informed consent was obtained from the patient and her guardian. No identifying information is disclosed. Genetic testing was recommended for further characterization.

Results: A 15-year-old female presented with progressive headaches, visual blurring, vertigo, and primary amenorrhea. She had previously undergone transsphenoidal resection of a GH-secreting pituitary macroadenoma. Clinical and radiological findings confirmed gigantism, NF1, and MRKH syndrome. She underwent further tumor debulking via right pterional craniotomy, with postoperative complications managed supportively. Follow-up imaging revealed residual tumor requiring adjuvant

radiotherapy.

Conclusion: This case represents a previously undocumented clinical triad involving GH-secreting pituitary macroadenoma, NF1, and MRKH syndrome. While their genetic loci are in proximity, no shared pathogenic pathway is known. Reporting this case expands the phenotypic spectrum of endocrine and developmental anomalies and highlights the importance of multidisciplinary evaluation and future genetic investigations.

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