Documents

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Prevalence and clinical correlations of SF3B1 variants in lactotroph tumours

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Abstract
Objective: A somatic mutational hotspot in the SF3B1 gene was reported in lactotroph tumours. The aim of our study was to examine the prevalence of driver SF3B1 variants in a multicentre independent cohort of patients with lactotroph tumours and correlate with clinical data. Design and methods: This was a retrospective, multicentre study involving 282 patients with lactotroph tumours (including 6 metastatic lactotroph tumours) from 8 European centres. We screened SF3B1 exon 14 hotspot for somatic variants using Sanger sequencing and correlated with clinicopathological data. Results: We detected SF3B1 variants in seven patients with lactotroph tumours: c.1874G > A (p.Arg625His) (n = 4, 3 of which metastatic) and a previously undescribed in pituitary tumours variant c.1873C > T (p.Arg625Cys) (n = 3 aggressive pituitary tumours). In two metastatic lactotroph tumours with tissue available, the variant was detected in both primary tumour and metastasis. The overall prevalence of likely pathogenic SF3B1 variants in lactotroph tumours was 2.5%, but when we considered only metastatic cases, it reached the 50%. SF3B1 variants correlated with significantly larger tumour size; higher Ki67 proliferation index; multiple treatments, including radiotherapy and chemotherapy; increased disease-specific death; and shorter postoperative survival. Conclusions: SF3B1 variants are uncommon in lactotroph tumours but may be frequent in metastatic lactotroph tumours. When present, they associate with aggressive tumour behaviour and worse clinical outcome.

Author Keywords
aggressive; carcinoma; lactotroph tumour; prolactinoma; SF3B1

Index Keywords
Ki 67 antigen, nucleic acid binding protein, prolactin, splicing factor 3b1, unclassified drug, phosphoprotein, RNA splicing factor, SF3B1 protein, human, transcription factor; adult, Article, cancer chemotherapy, cancer radiotherapy, cohort analysis, disease specific survival, exon, female, genetic association, histopathology, human, hypophysis tumor, major clinical study, male, metastasis, prevalence, primary tumor, prolactin blood level, prolactinoma, proliferation index, retrospective study, Sanger sequencing, tumor volume, clinical trial, genetics, hypophysis tumor, multicenter study, prevalence, prolactin secreting cell; Humans, Lactotrophs, Phosphoproteins, Pituitary Neoplasms, Prevalence, Retrospective Studies, RNA Splicing Factors, Transcription Factors

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