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A rare and misleading presentation of a primary glial tumour. The usefulness of molecular profiling, even in "obvious" cases

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We report the case of a 57 years old, male patient. The patient was previously diagnosed with an small cell neuro-endocrine carcinoma of the lung for which he received treatment. The patient encountered sudden collapse with a short epileptic episode. During routine scanning (CT/MRI brain) a circumscript lesion was found in the right frontal cerebral lobe. Biopsy revealed a small cell neuro-endocrine tumour. Immunohistochemistry revealed positivity for Synaptofysin, CD56 and TTF-1. Chromogranin was negative. Ki67 was highly expressed. Pankeratin was negative. The immune profiling was similar to the previously diagnosed lung process. Surprisingly GFAP was strongly positive as well. Due to the GFAP expression, molecular profiling was performed. This, leading undoubtedly to the conclusion that this was not a metastatic process from the lung but a strongly neuro-endocrine differentiated glioblastoma mimicking metastasis of a small cell lung carcinoma in a patient known with such a tumour. This case is illustrative for the use of molecular profiling in brain tumours, even when it appears to be an "obvious" diagnosis. MRI-scan revealed a small mass in the right frontal lobe, Due to its round structure a metastasis was suggested, but a primary brain tumour was not excluded due to the partly cystic appearance of the lesion. Diagnostic biopsy was advised. H&E histology revealed a small cell pattern with massive necrosis, moulding and many mitosis. The pattern was similar to the previous process in the lung. However, not similar to the lung was the appearance of mild vascular proliferation.

Conclusion and literature: Although this tumour had the initial appearance of metastasis of a pulmonary small cell neuroendocrine carcinoma, additional molecular profiling prevented this unusual pitfall. Eventually the lesion appeared to be rare differentiation of a glioblastoma multiforme.

Biography

Frank van de Goot is a registered Anatomical Pathologist and an expert witness in the field of forensic pathology. He studied Medicine and Anatomical Pathology at the VU University Medical Center in Amsterdam, The Netherlands, and Rechtsmedizin at the Zentrum für Rechtsmedizin in Frankfurt am Main, Germany. The first seven years of his career he worked both as a Forensic Pathologist at the Netherlands Forensic Institute (NFI) in The Hague, The Netherlands and as an Anatomical Pathologist at the VU University Medical Center. He now works at Symbiant Pathology Expert Centre, where he is on a mission to improve forensic and autopsy education for both medical and non-medical students.

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