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DNA-Analysis is Mandatory in Case of an Uncommon Malignancy

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In a child a diagnosis of sweat gland carcinoma was made on basis of a surgical specimen presumed to be taken from an occipital

lymphnode. DNA analysis confirmed mixing of specimens in the referring hospital. *Med. Pediatr. Oncol.* 29:65–66, 1997.

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Key words: diagnostic error; sweat gland carcinoma; DNA-analysis

INTRODUCTION

Recently Trinkle et al. reported in your journal on errors in chemotherapy. Recently we could confirm an error in the diagnostic work-up of a child suspected of suffering from a poorly differentiated metastatic adenocarcinoma. It was shown that the surgical specimen originated from an adult patient with breast-carcinoma.

CASE REPORT

In an 11-year-old girl a small bleeding lesion preceded a lymphnode swelling. Lesion and lymphnode swelling were both localized in the occipital area. In a 3 month-period the node had gradually increased in size to a diameter of 1.5 cm. There was some fatigue. In the referring hospital the lymphnode was excised and a diagnosis of a metastasis of a poorly differentiated adenocarcinoma was made. Review of the biopsy material, as was sent by the referring hospital, showed a lymphnode metastasis of an adenocarcinoma, immunohistochemically characterized by a positive staining for low and high molecular keratins, lactoferrin, EMA, vimentin, and SMA, and electronmicroscopically by the presence of squamous-, adeno-, and myelo-epithelial differentiations. This pattern suggested mammary, salivary, or sweat gland origin. Based on the site of involvement a metastasis of a sweat gland carcinoma was suggested. MRI-scanning of the head and neck revealed no abnormalities. Ultrasound examination of abdomen, thyroid, and breasts, as well as serum tumour markers (CEA, cancer antigen 125, alpha-fetoprotein) were unremarkable. The head of the girl was shaved for thorough examination of the skin. No primary tumour could be found. We presumed that the primary tumour, the red, pruritic lesion had either been eliminated at the time of the excision of the lump or spontaneously had regressed. We contacted the pediatrician and the pathologists of the referring hospital to check whether the specimen was indeed from the child. They stated on several occasions that this was the case. A posterior neck dissection was performed, which also re-

sulted in the removal of some of the neck musculature and part of the deltoid muscle. The 31 lymphnodes found were free of tumour. DNA from the posterior lymphnode dissection was compared with DNA of normal lymphoid tissue present in the initial lymphnode excision. The findings didn't match. They were discussed with the pathologist of the referring hospital, who reviewed the specimens received at the day of the initial lymphnode excision. Subsequently a tumour-negative lymphnode from a patient with mammary carcinoma was sent. The DNA-profile of this "axillary" lymphnode was DNA-identical to the material from the posterior neck-dissection. The mixing up of specimens the two patients was obvious.

In the first months after surgery the patient developed a dystorsion of the neck that could be redressed by physical therapy.

DISCUSSION

The suspected disease, i.e. sweat gland carcinoma, is extremely rare in children [2–4]. The tumour is seen at all ages, and there seems to be a predominance for girls [3,4]. The localisation in this girl was in line with descriptions in literature [4]. As we were assured (on several occasions) that the specimen was indeed from this child we treated the child accordingly by excision of regional lymphnodes [5,6].

By preoperative DNA-analysis, like comparison of surgery specimens with a peripheral blood sample, it

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would have been possible to detect the error prior to surgery. To our knowledge in pediatric oncology no reports existed on DNA-analysis to prove the origin of surgical specimens. We conclude that in case of a seldomly occurring malignancy; the origin of the specimen on which the diagnosis is based should be checked at initial presentation.

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