

Cutaneous form of maculopapular mastocytosis in a foal

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Background – Cutaneous mastocytosis is a rare benign disease occurring in domestic animals and humans. In previous reports, dermal findings in foals were accompanied by systemic mast cell infiltrations, whereas lesions in human cutaneous mastocytosis, including urticaria pigmentosa and solitary mastocytoma, are usually restricted to the skin.

Objectives – To describe a new variant of equine cutaneous maculopapular mastocytosis lacking systemic involvement.

Animals – A 2.5-month-old warmblood foal with multiple skin nodules since birth.

Methods – Clinical examination (including haematology, fine needle biopsy and thoracic radiographs), postmortem examination, histopathology and immunohistochemistry.

Results – Clinical examination showed 41 skin nodules that contained numerous mast cells as detected by cytology. Macroscopic examination at postmortem examination revealed intradermal circumscribed lesions ranging from 2 to 5 cm in diameter. Histologically, they were composed of well differentiated mast cells with metachromatic granules stained with toluidine blue accompanied by many eosinophils. Immunohistochemically, mast cells had mast cell growth factor receptor c-KIT predominating at the cell surface and intracytoplasmic expression of tryptase. In other organs similar mast cell infiltrations were not detected.

Conclusions and clinical importance – The case presented here fulfils the criteria of equine cutaneous maculopapular mastocytosis (ECMM), representing a rare entity in foals that is reported to be associated with spontaneous regression, although the long-term prognosis is not known. Unlike in previous reports, lesions described here were restricted to the skin. This may imply that ECMM is primarily a dermal disease sharing similarities with urticaria pigmentosa in young children.

Introduction

Neoplastic mast cell disorders have been described in many domestic animal species and humans.^{1,2} They are most commonly diagnosed at advanced age and may have different clinical outcomes.^{3,4} In contrast, cutaneous mastocytosis (CM) is a rare disorder typically appearing very early on in life.^{5,6} In humans, CM is mostly restricted to the skin and shows a benign behaviour with recurrence until puberty. In comparison, mastocytosis can be more aggressive and associated with systemic forms when occurring in adults.⁶ The major forms of CM described in

humans include cutaneous mastocytoma, multifocal mastocytosis (also known as urticaria pigmentosa, UP) and diffuse CM.⁶ CM arising early in life sporadically occurs in dogs, cats and cattle, and can arise in a multifocal or diffuse pattern.^{7–10} In addition, a maculopapular form of CM is a rare disorder in young horses; only two case reports of CM in foals have been published.^{5,11} Both publications implicated a benign behaviour of equine cutaneous maculopapular mastocytosis (ECMM) as lesions disappeared within an interval of 30 days. However, unlike in humans, scientific evidence for a good long-term prognosis for ECMM in foals is limited as both foals were necropsied at approximately 1 year of age. Both foals with ECMM showed systemic mast cell infiltrations in addition to skin lesions, which is in contrast to the majority of human cases.^{5,11} In human CM, mutations in the stem cell growth factor receptor c-KIT inducing uncontrolled mast cell proliferation are suggested to be responsible for some cases in adults, whereas such mutations seems to be absent in younger patients.¹² Although the immunohistochemical c-KIT staining pattern is discussed to have a prognostic significance in mast cell tumours in dogs, it is not associated with clinical outcome in the equine counterpart.^{13,14}

Accepted 6 March 2016

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Sources of Funding: This study was self-funded.

Conflict of interest: The authors declare that there is no competing interest. Gundi Müller and Achim D. Gruber were involved in this case during their time at the Department of Pathology (University of Veterinary Medicine, Hannover, Germany). This study was not supported or influenced by Merck KGaA.

Case report

A 2.5-month-old brown warmblood filly was presented with 41 cutaneous nodules measuring 2 to 5 cm in diameter and being most prominent on the back, rump and limbs (Figure 1). The masses were either hairless, grey and ulcerated, or sparsely covered with dark brown to black hair. The nodules were moveable on the fascia and moderately painful during palpation. No pruritus was evident. Clinical evaluation included haematology, fine needle aspiration from the nodules and radiographs. The only abnormality on a serum chemistry panel and complete blood count was a moderate leucocytosis ($16.5 \times 10^9/L$; reference range $5\text{--}12 \times 10^9/L$) with a left shift. Radiography of the thorax was unremarkable and there were no features of mast cell mediator related disease such as pruritus, oedema, other circulatory disturbances, respiratory distress or abdominal pain. Clinical signs, in addition to high numbers of mast cells harvested from skin nodules during aspiration, suggested cutaneous mastocytosis, although cutaneous habronemiasis, onchocerciasis or eosinophilic granuloma represented possible differential diagnoses.

The owner declined further diagnostics and clinical care, and requested euthanasia due to financial reasons. Subsequently, full post-mortem examination was performed and revealed the lesions to be restricted to the skin. Several tissues were collected for histopathology and included the following organs: skin (nine nodules), lymph nodes, spleen, bone marrow, thymus, liver, small and large intestine, kidney, heart, skeletal muscle, epiphysis of long bone, cerebrum, cerebellum, adrenal gland and thyroid gland. Light microscopy showed cell-rich circumscribed nodular masses extending from the superficial dermis into the subcutis that were associated with varying amounts of infiltrating eosinophils (Figure 2). All examined nodules revealed a similar appearance. They were predominately composed of round cells arranged in closely packed, perivascular to interstitial, multifocal to coalescing sheets (Figure 3). Cells were well differentiated with unremarkable nuclei



Figure 1. Clinical presentation of neonatal equine mastocytosis: the skin shows multiple nodular masses, some of which were ulcerated.

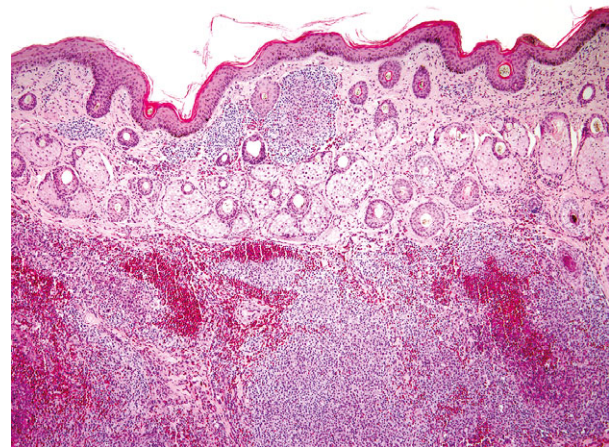


Figure 2. Neonatal equine mastocytosis, histopathology: dermal nodules are highly cellular, extend from the superficial into the deep dermis and subcutis, and contain varying numbers of eosinophils. Haematoxylin and eosin stain, 40× magnification.

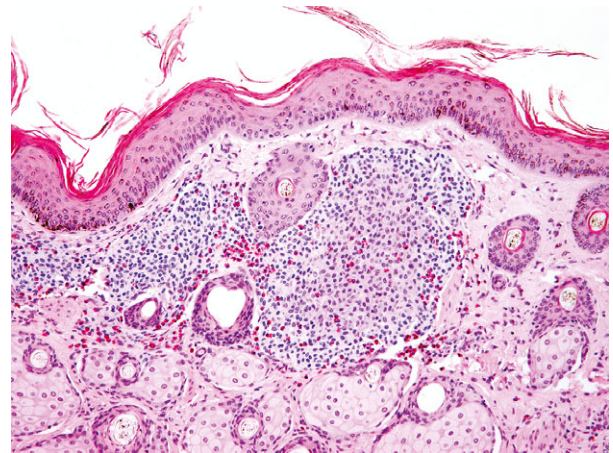


Figure 3. Neonatal equine mastocytosis, histopathology: mast cells are arranged in multinodular to diffuse, perivascular to interstitial, closely packed sheets. Haematoxylin and eosin stain, 100× magnification.

showing less than one mitotic figure per high power field. They contained moderate amounts of light, basophilically stippled cytoplasm with metachromatic granules demonstrated by toluidine blue stain and identifying them as mast cells (Figure 4). This was further accompanied by single, fragmented, shrunken, hypereosinophilic to hyalinized collagen fibres surrounded by partly degenerated eosinophils, as well as multifocal necrosis associated with few occasionally multinucleated macrophages and neutrophils. Skin nodules were not associated with increased dermal or epidermal pigmentation. Multifocally, there was mild epidermal hyperplasia.

Immunohistochemistry was performed on formalin-fixed and paraffin-embedded samples for detecting tryptase (monoclonal antibody to human tryptase, diluted 1:50, clone AA1, Dako; Hamburg, Germany) and stem cell growth factor receptor c-KIT (polyclonal antibody to human c-KIT, diluted 1:100; Dako) using the avidin–biotin complex method. Sections of equine and canine mast cell tumours were used for positive controls. Primary antibody-

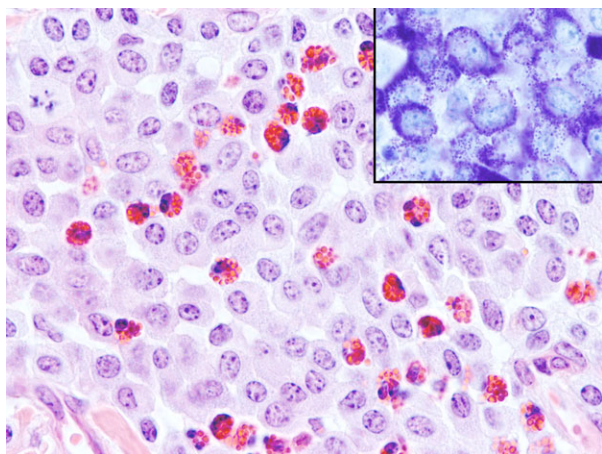


Figure 4. Neonatal equine mastocytosis, histopathology: mast cells are well differentiated with moderate amounts of light, basophilically stippled cytoplasm containing metachromatic granules (insert), unremarkable nuclei and only few mitoses. Haematoxylin and eosin stain, insert toluidine blue stain, both 600× magnification.

ies were replaced by ascites fluid from nonimmunized BALB/cJ mice (Cedarlane; Ontario, Canada) or normal rabbit serum (Sigma Aldrich; Taufkirchen, Germany) for negative controls. The percentage of positive cells was evaluated by counting a total of at least 800 cells at 400× magnification in randomly selected areas. Tryptase was found in the cytoplasm of 95% of mast cells, whereas c-KIT was predominantly expressed at the cell surface (mean 73%).

Increased mast cell infiltrations were not detected in other organs during histopathological evaluation of slides stained with haematoxylin and eosin as well as toluidine blue. The absence of intralesional parasites, secondary bacterial infections or fungi was confirmed with periodic acid Schiff reaction, Gram staining and Grocott silver impregnation, respectively.

Discussion

Pathological findings of the present case indicate a proliferative mast cell disease and fulfil the criteria of ECMM.¹⁵ This is a rare entity in foals and previous descriptions have been limited to a male standard bred and a male quarter horse foal, both of which showed an onset of the disease in the neonatal period.^{5,11} Similar to the present case, lesions in those animals were dominated by well differentiated mast cells accompanied by eosinophils.^{5,11} Although, in some respects, the lesions studied herein partly resemble those present in cutaneous habronemiasis, onchocerciasis or eosinophilic granuloma, none of these is characterized by sheets of mast cells.¹⁶ This implicates histopathology of skin biopsies as the most suitable method for establishing the diagnosis of ECMM in living foals. In both published cases, mast cell infiltrations were also detected in the spleen, lymph nodes and bone marrow, and the authors suggested ECMM to be a systemic disease with dermal manifestation.⁵ In the present case, however, lesions were exclusively restricted to the skin and this suggests ECMM to be primarily a dermal disease that may show systemic involvement. This is

in accordance with CM in children, which is commonly restricted to the skin, whereas systemic involvement is rarely reported at advanced age and commonly associated with a more aggressive disease.⁶ ECMM seems to be of benign behaviour and spontaneous regression associated with increasing intralesional fibroblast proliferation was reported in previous cases during a 30-day period.^{5,11,14} Because mast cells are well differentiated and lesions can regress spontaneously, it is still under debate whether ECMM represents hyperplastic lesions or true neoplasms.^{5,6,15,17} A self limiting clinical course is also reported for CM in dogs and children.^{6,18} However, scientific evidence for a good long-term prognosis in ECMM is limited.^{5,11}

Activating mutations of c-KIT inducing extensive mast cell proliferation and survival due to proto-oncogene activation are assumed to be responsible for the different clinical behaviour in the minor population of persisting UP in human adults.¹² In contrast to dogs, c-KIT staining pattern is not associated with clinical outcome of mast cell tumours in horses.^{13,14} Therefore, the significance of the membranous c-KIT staining pattern in the present case, which may imply that c-KIT mutations do not play a role in the pathogenesis of this condition in foals, should be interpreted carefully because the long-term clinical course of ECMM is still unknown. In addition to proto-oncogene activation, an imbalance between mast cell proliferation and apoptosis is involved in the development of human mastocytosis.¹⁹

ECMM differs from the canine and feline counterparts as lesions in foals are sharply demarcated and elevated, whereas they show a poorly demarcated, papular pattern in dogs and cats.^{5,7,8,11,20} However, mast cells show perivascular to diffuse arrangements in sheets in both canine and equine cases. UP represents the most common form of CM in humans and the typical brown discolouration of the disseminated maculopapular lesions is responsible for the term “pigmentosa”.² Lesions in UP are most commonly heterogeneous with variable size (polymorphic) and may range from nodules to plaques and macules, can be elevated or flat, and may show sharp or indistinct margins.²¹ Their hyperpigmentation is related to increased melanin production as a result of melanocyte stimulation by mast cell growth factor.²² Hypermelanosis is also reported in feline CM but seems to be absent in canine patients.^{7,23} Although ECMM shares some similarities with human CM (especially UP), skin nodules were not associated with elevated melanocytes or melanomacrophages in the present case, which is consistent with previous reports.^{5,11}

To the best of the authors' knowledge this is the first description of ECMM in which the lesions were restricted exclusively to the skin without systemic involvement. Although ECMM is suggested to be a benign condition, further studies are needed to elaborate on the long term clinical course of this disease because scientific evidence for a good long-term prognosis is limited so far.

Acknowledgements

We are grateful to Bettina Buck, Petra Grünig and Klaus-Peter Kuhlmann for their excellent technical support.

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Résumé

Contexte – La mastocytose cutanée est une maladie bénigne rare décrite chez les animaux domestiques et l'homme. Dans des publications antérieures, les lésions dermiques des poulains étaient accompagnées par des infiltrations mastocytaires systémiques tandis que les lésions de mastocytose cutanée de l'homme, comprenant l'urticaire pigmentaire et les mastocytomes solitaires sont généralement limitées à la peau.

Objectifs – Décrire un nouveau variant de mastocytose maculo-papuleuse cutanée équine sans atteinte systémique.

Sujets – Un poulain de 2.5 mois pur-sang présentant depuis la naissance des nodules cutanés multiples.

Méthodes – Un examen clinique a révélé 41 nodules cutanés contenant de nombreux mastocytes mis en évidence par cytologie. Un examen macroscopique au cours de l'autopsie a révélé des lésions circonscrites intra-dermiques de 2 à 5 cm de diamètre. A l'examen histopathologique, il y avait des mastocytes bien différenciés avec des granules métachromatiques colorés au bleu de toluidine, accompagnés de nombreux éosinophiles. A l'examen immunohistochimique, les mastocytes avaient des récepteurs c-KIT prédominant à la surface cellulaire et une expression intracytoplasmique de tryptase. Un infiltrat mastocytaire semblable n'a été détecté dans aucun autre organe.

Conclusions et importance clinique – Le cas décrit ici remplit les critères de mastocytose cutanée équine maculo-papuleuse (ECMM), entité rare chez les poulains qui est associée à une régression spontanée, bien que le pronostic à long terme ne soit pas connu. A la différence des cas précédemment décrits, les lésions étaient restreintes ici à la peau. Ceci peut impliquer que l'ECMM soit principalement une maladie dermique partageant des similitudes avec l'urticaire pigmentaire du jeune enfant.

Resumen

Introducción – la mastocitosis cutánea es una enfermedad benigna rara que ocurre en animales domésticos y humanos. En casos previos publicados, los hallazgos clínicos en la dermis de potros estaban acompañados por infiltración sistémica de mastocitos, mientras que las lesiones en mastocitosis cutánea humana, incluyendo la urticaria pigmentosa y el mastocitoma solitario, están normalmente restringidas a la piel.

Objetivos – describir una variante novedosa de mastocitosis cutánea maculopapular equina sin diseminación sistémica

Animales – un potro de 2,5 meses de edad de raza de sangre tibia presentaba numerosos nódulos en la piel desde su nacimiento.

Métodos – examen clínico (incluyendo hematología, biopsia de aguja fina, y radiografías torácicas), examen postmortem, histopatología e inmunohistoquímica.

Resultados – el examen clínico demostró 41 nódulos en la piel que contenían numerosos mastocitos detectados mediante citología. El examen microscópico postmortem reveló lesiones intradérmicas circunscritas que variaban de dos a 5 cm de diámetro. Histológicamente estaban compuestas de mastocitos bien diferenciados con gránulos metacromáticos teñidos por azul de toluidina acompañados por numerosos eosinófilos. Mediante inmunohistoquímica se demostró que los mastocitos poseían el receptor c-KIT predominantemente en la superficie celular así como expresión intracitoplásmica de triptasa. No se detectó infiltrado similar de mastocitos en otros órganos.

Conclusión e importancia clínica – el caso presentado aquí completa los criterios de la mastocitosis maculopapular cutánea e (ECMM), que representa una entidad rara en potros asociada con regresión espontánea, aunque se desconoce el pronóstico a largo plazo. A diferencia de informes previos, las lesiones descritas en este caso estaban restringidas a la piel. Esto puede implicar que ECMM es primariamente una enfermedad dermal y comparte similitudes con la urticaria pigmentosa infantil.

Zusammenfassung

Hintergrund – Die kutane Mastozytose ist eine seltene gutartige Erkrankung, die bei Haustieren und beim Menschen vorkommt. In früheren Berichten wurden bei Hautbefunden von Fohlen begleitende systemische Mastzellinfiltrationen beschrieben, während Veränderungen der kutanen Mastozytose, wie der Urticaria pigmentosa und solitäre Mastozytome des Menschen sich üblicherweise auf die Haut beschränken.

Ziele – Die Beschreibung einer neuen Variante der equinen kutanen makulopapulären Mastozytose ohne systemische Beteiligung.

Tiere – Ein 2,5 Monate altes Warmblutfohlen mit multiplen Hautknoten, die es seit der Geburt hatte, wurde beschrieben.

Methoden – Eine klinische Untersuchung (inklusive Hämatologie, Feinnadelbiopsie und Thoraxröntgenaufnahmen), eine Post mortem Untersuchung, Histopathologie und Immunhistochemie werden beschrieben.

Ergebnisse – Bei der klinischen Untersuchung bestanden 41 Hautknoten, die zahlreiche Mastzellen enthielten, die mittels Zytologie gefunden wurden. Die makroskopische Untersuchung bei der postmortem Untersuchung zeigte intradermale umschriebene Veränderungen, die zwischen 2 und 5 cm im Durchmesser waren. Histologisch bestanden sie aus gut differenzierten Mastzellen mit metachromatischen Granula, die sich mit Toluidinblau anfärbten und von zahlreichen Eosinophilen begleitet waren. Immunhistochemisch wiesen die Mastzellen den Mastzellwachstumsfaktor Rezeptor c-KIT vorwiegend an der Zelloberfläche auf und exprimierten intrazytoplasmatisch Tryptase. In anderen Organen konnten keine derartigen Mastzellinfiltrationen gefunden werden.

Schlussfolgerungen und klinische Bedeutung – Der Fall, der hier vorgestellt wird, repräsentiert die Kriterien einer equinen kutanen makulopapulären Mastozytose (ECMM), eine seltene Erkrankung, die wie beschrieben, spontan zurückgeht, obwohl eine Langzeitprognose nicht bekannt ist. Anders wie in früheren Berichten, blieben die Veränderungen in diesem Fall auf die Haut beschränkt. Das könnte darauf hinweisen, dass ECMM primär eine Hauterkrankung darstellt, die Ähnlichkeiten mit Urticaria pigmentosa bei kleinen Kindern teilt.

要約

背景 – 皮膚肥満細胞症は家畜およびヒトに生じる稀な良性疾患である。過去の報告では、子馬における皮膚所見は全身性の肥満細胞浸潤と関連していた一方、ヒトの皮膚肥満細胞症における病変は色素性蕁麻疹および孤立性肥満細胞腫を含み、通常皮膚に局限している。

目的 – 全身性の関与を欠くウマの皮膚斑丘疹状肥満細胞症の新しい異型を解説すること。

供与動物 – 出生時から複数の皮膚結節の存在する2.5ヶ月齢のwarmblood種の子馬。方法—臨床検査(血液学、針生検および胸部レントゲン写真を含む)、死後剖検検査、病理組織学、免疫組織化学。

結果 – 臨床検査により細胞診で検出された多数の肥満細胞を含む41個の皮膚結節を認めた。死後剖検検査での顕微鏡学的な検査により皮内に直径2〜5cm大の境界明瞭な病変が明らかとなった。組織学的にそれらは好酸球を伴い、トリジンブルーで染色された異染色性顆粒を持つ分化した肥満細胞から構成されていた。免疫組織学的に、肥満細胞は肥満細胞増殖因子受容体のc-KITが細胞表面に目立ち、細胞質内にはトリプターゼが発現していた。他の臓器では類似した肥満細胞浸潤は検出されなかった。

結論および臨床的な重要性 – ここに示した症例は長期的な予後は不明だが、自然退縮と関連すると報告された子馬における稀な存在で、ウマの皮膚斑丘疹状肥満細胞症(ECMM)の診断基準を満たしていた。以前の報告と異なり、ここで解説された病変は皮膚に局限していた。これはECMMが小児における色素性蕁麻疹と類似性をしめす主に皮膚疾患であることを意味している可能性がある。

摘要

背景 — 皮肤肥大细胞增多症是一种出现在家养动物和人身上的罕见良性疾病。在之前的一篇研究中,幼年马真皮发现该病,并伴随全身性肥大细胞浸润,然而,人皮肤肥大细胞增多症病变包括色素性荨麻疹和单个肥大细胞瘤,通常局限于皮肤。

目的 — 描述一种无全身症状的马斑丘疹肥大细胞增多症的新变种。

动物 — 只2.5月龄温血马,自出生起就有多处皮肤结节。

方法 — 临床检查(包括血液学检查,细针活检和胸部X线检查)、尸检、组织病理学和免疫组化。

结果 — 临床检查显示,41个皮肤结节细胞学检查结果为大量肥大细胞。尸检眼观皮内局限性病变直径为2-5厘米。组织学显示他们由分化良好的肥大细胞组成,其具有甲苯胺蓝染色异染颗粒,同时伴随许多嗜酸性粒细胞。免疫组化显示,肥大细胞表面以肥大细胞生长因子受体c-KIT为主,胞浆内有类胰蛋白酶表达。未在其他器官检测到相似的肥大细胞浸润。

总结与临床意义 — 这个病例的出现完善了马皮肤斑丘疹肥大细胞增多症(ECMM)的标准,为幼年马的一种罕见疾病,曾报道过可自愈,但时间未知。与之前的研究不同,这次病变只局限于皮肤。这意味着ECMM主要发病于皮肤,与幼儿色素性荨麻疹有共同点。