Adult nasal chondromesenchymal hamartoma: a rare and benign tumour with aggressive malignant transformation

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SUMMARY

Nasal chondromesenchymal hamartoma (NCMH) is an extremely rare benign tumour of the nasal cavity predominantly described in infants. We report a case involving a 48-year-old woman who had been diagnosed with NCMH a year earlier and now re-presented with a short history of progressive nasal blockage, recurrent epistaxis and orbital apex syndrome. Histopathology was suggestive of malignant transformation into sinonasal sarcoma. However, following multidisciplinary team (MDT) discussions, including second and third opinions from external departments, the histological diagnosis was revised to 'NCMH with bizarre stromal cells'. Despite this, the lesion demonstrated malignant features of rapid, invasive growth and was treated with palliative radiotherapy. The patient later developed radiological evidence of lung and liver metastases with subsequent pulmonary emboli. Shortly after this, she passed away. This case is unique in its diagnostic challenge, with ambiguous histopathological findings, and highlights the importance of an MDT approach when managing complex sinonasal tumours.

BACKGROUND

Nasal chondromesenchymal hamartoma (NCMH) is a rare, benign sinonasal neoplasm of mesenchymal and cartilaginous tissue. It has been linked to DICER1 genetic mutations on chromosome 14 and predominantly occurs in young children and infants aged less than 1 year.[1] However, there have been eight reports of NCMH affecting adults and one previously reported case of malignant transformation involving a 40-year-old woman.[2]

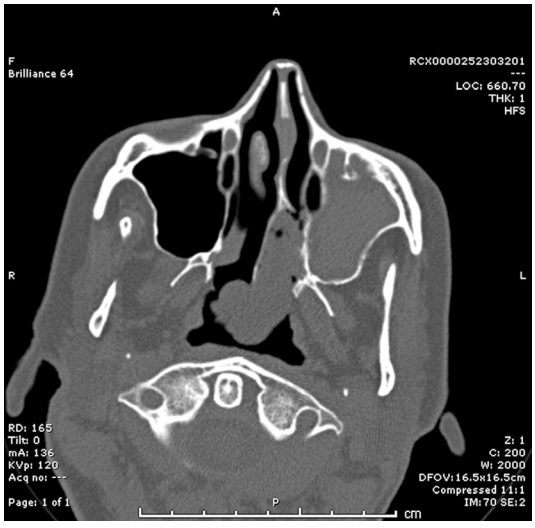

Figure 1 Non-contrast axial CT image demonstrating opacification of the left maxillary sinus and tumour extending posteriorly into the nasal cavity and towards the nasopharynx.

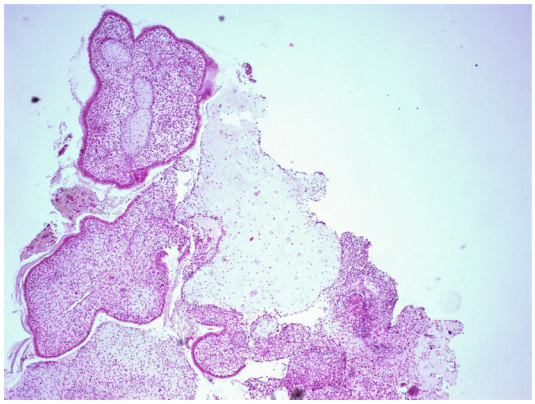

Figure 2 Photomicrograph demonstrating spindle cell proliferation and mature cartilage underlying nasal mucosa. Taken at 2× magnification and with H&E staining.

Presentation of NCMH is variable and dependent on which local structures are involved. The most common presentations include nasal congestion, nasal mass and eye signs.

Epistaxis and nasal blockage are common ear, nose and throat (ENT) presentations. The differential diagnoses for these presentations are wide and include benign pathology, such as inverted papilloma, inflammatory polyps, hamartoma, mucocele and cholesterol granuloma, and malignant pathology, including squamous cell carcinoma and sinonasal sarcoma.

CASE PRESENTATION

A 47-year-old woman presented to her local ENT department with symptoms of chronic left nasal congestion, rhinorrhoea, unilateral nasal blockage and facial pain. She had no notable medical history and was otherwise in good health. On examination, a left nasal polyp was identified. CT and MRI of her sinuses demonstrated opacification of the left maxillary sinus and tumour extending posteriorly into the nasal cavity and towards the nasopharynx (figure 1). She subsequently underwent left functional endoscopic sinus surgery (FESS) and polypectomy of a large left pale polyp that was originating from the left sphenoid sinus. Histopathology

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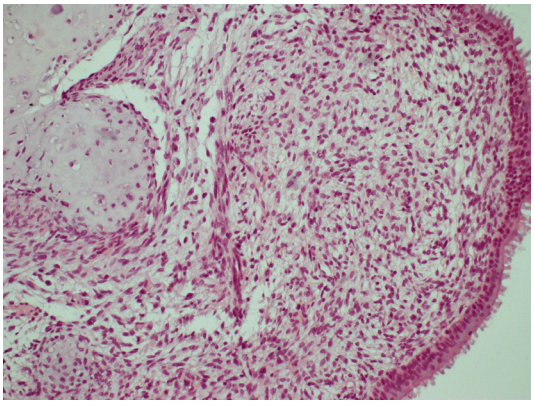

Figure 3 Photomicrograph demonstrating spindle cell proliferation and mature cartilage underlying nasal mucosa. Taken at 20× magnification and with H&E staining.

reported the nasal polyp as benign, and following surgery, she was symptom-free.

One year later, the patient re-presented with recurrence of symptoms. A CT scan showed recurrence of disease and she underwent a revision FESS and polypectomy. The histopathology report at this point suggested NCMH or chondro-osseous respiratory epithelial hamartoma. Unfortunately, following surgery, the patient's symptoms remitted only briefly. Within a few weeks, she developed worsening rhinorrhoea, complete left-sided nasal blockage and severe headaches.

While awaiting follow-up, the patient developed intractable left-sided epistaxis, requiring three inpatient admissions over the course of a few weeks, each requiring bilateral nasal packing and ultimately a sphenopalatine artery ligation during which repeat biopsies were taken from the left nasal cavity. Microscopic analysis of biopsied tissue was sought from three separate specialist centres within the UK. Photomicrographs of the lesion demonstrated spindle cell proliferation, mature cartilage underlying nasal mucosa and presence of bizarre stromal cells, initially thought to represent a malignant sinonasal sarcoma (figures 2 and 3). However, following multidisciplinary team (MDT) input and specialist advice, a diagnosis of a rapidly growing benign chondromesenchymal hamartoma was made.

While awaiting a definitive histopathology report, the patient underwent extensive imaging, including CT neck and chest, and MRI of the sinuses. At this time, there was no radiological evidence of metastasis.

MRI sinuses and head (figures 4 and 5) revealed extensive tumour in the left nasal cavity extending into the maxillary sinus, left orbital apex and encasing the internal carotid artery within the cavernous sinus. In addition, there was involvement of the left optic nerve, left superior orbital fissure, and sphenopalatine foramen and foramen rotundum bilaterally with the tumour extending through the cribriform plates displacing the olfactory bulbs.

Her case was discussed in a local MDT meeting and subsequently referred to a specialist tertiary centre in London. Following clinic review in London, it was decided that surgical excision was not a viable or safe option. She was subsequently scheduled for urgent palliative radiotherapy to provide symptomatic relief.

OUTCOME AND FOLLOW-UP

The patient underwent a 12-week course of local radiotherapy resulting in initial marked regression of symptoms.

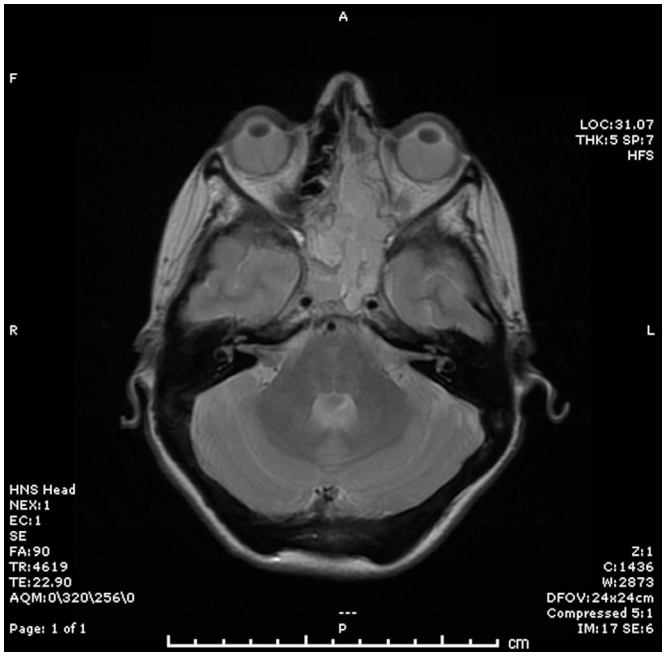

Figure 4 T2-weighted fluid-attenuated inversion recovery axial MRI demonstrating a large sinonasal tumour within the left nasal cavity. There is encasement and narrowing of the left internal carotid artery in the anterior aspect of the left cavernous sinus.

Unfortunately, over the following 6 months, there was extensive tumour regrowth. CT imaging at this point demonstrated pulmonary and liver metastases with intrahepatic inferior vena cava obstruction and multiple pulmonary emboli. She unfortunately passed away shortly after this.

DISCUSSION

NCMH was first described in 1998 with the vast majority of reported cases in children, predominantly infants.[3] A total of 59

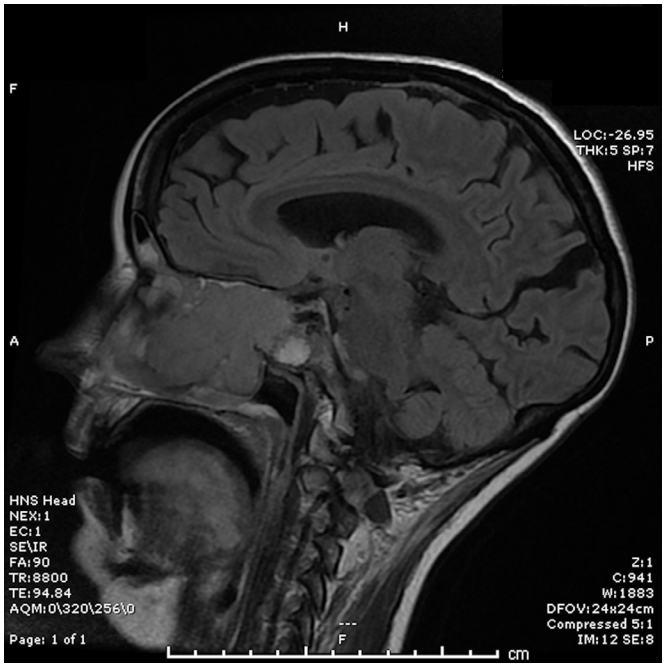

Figure 5 T2-weighted fluid-attenuated inversion recovery sagittal MRI demonstrating tumour bulk and opacification of the nasal cavity.

cases have been described, 49 of which were diagnosed in those aged less than 18 years of age.[4 5]

In 2013, Li *et al* described the first case of malignant transformation in NCMH. This involved a 40-year-old woman presenting with nasal obstruction and epistaxis. Histological and immunohistochemical analyses showed mixed morphology. A portion of the mass in this case was consistent with NCMH while some areas exhibited cytological atypia and marked mitoses. Atypical mesenchymal spindle cells were immunoreactive for vimentin, CD99 and smooth muscle actin diffusely. Cartilaginous cells were immunopositive for S-100 protein and Ki-67 index high in atypical areas. Unfortunately, a rapid recurrence of tumour was observed 3 months following radical resection.

Ambiguous histology and immunohistochemistry is also reflected in our case with bizzare stromal cells found focally positive for desmin, spindle cells positive for actin and showing prominent hyperchromatic nuclei. No increased mitotic activity was seen. Yet, despite a benign final histological diagnosis of 'NCMH with bizarre stromal cells', the tumour behaved malignantly with radiological evidence of advanced metastatic disease and aggressive local invasion.

Learning points

- Nasal chondromesenchymal hamartoma (NCMH) tumours can be a rare differential diagnosis of epistaxis and nasal congestion.
- NCMH has the potential for aggressive malignant transformation.
- NCMH tumours can demonstrate rapid regrowth and local invasion following resection.
- Advanced NCMH tumours may be treated with radiotherapy in cases with where a surgical approach is not feasible or safe.

Treatment of NCMH is typically surgical. This is usually via minimally invasive endonasal endoscopic resection. In our case, surgical resection was initially planned; however, due to rapid intracranial expansion of the tumour and involvement of highly important and delicate structures, including the internal carotid artery and optic nerve, a palliative approach with radiotherapy was adopted.

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