

CASE REPORT

PRIMARY CNS MELANOMA IN AN ALBINO: A RARE CASE REPORT

Kishore Sanjeev¹, Bhardwaj Aparna², Gupta Mohit³, Seema Acharya⁴, Kudesia Sandip⁵

HOW TO CITE THIS ARTICLE:

Kishore Sanjeev, Bhardwaj Aparna, Gupta Mohit, Seema Acharya, Kudesia Sandip. "Primary CNS Melanoma in an Albino: A Rare Case Report". Journal of Evolution of Medical and Dental Sciences 2014; Vol. 3, Issue 27, July 07; Page: 7609-7613, DOI: 10.14260/jemds/2014/2944

ABSTRACT: Primary intracranial melanoma is a rare and uncommon lesion. Association of primary CNS melanoma in an albino has not been reported in literature searched till now. We are presenting a rare case of primary CNS melanoma in a 52years old male with oculocutaneous albinism. The patient presented with repeated episodes of generalized headache, vomiting and ataxia for duration of 5months. MRI examination showed a tumor in the posterior fossa that was diagnosed as Ependymoma radiologically. Surgical treatment with total removal of tumor was done. Intra-operative squash cytology and frozen section followed by histopathology confirmed the diagnosis of Melanoma. A thorough investigation of the patient was performed including chest radiography, ocular examination, ultrasonography of abdomen and barium enema to rule out any other site of primary melanoma in the body. Thus a final diagnosis of primary CNS melanoma was given.

KEYWORDS: CNS, Melanoma, Oculocutaneous albinism, Ependymoma.

INTRODUCTION: Primary melanocytic tumours of CNS are rare and comprise 1% of all the tumours.^[1] An estimated incidence of 0.9 per 10 million of these tumours has been reported from the literature.^[2] Their spectrum varies from benign to malignant and includes leptomeningeal melanocytosis, melanocytoma to its overtly malignant counterpart melanoma.^[1] The diagnosis of intracranial melanomas is often complicated and a differential diagnosis of other pigmented lesions like pigmented Meningioma, Schwannoma, Medulloblastoma, Choroid plexus papilloma, Astrocytoma and pituitary tumours should be considered. A diagnosis of primary CNS melanoma should be made after extensive exclusion of cutaneous or mucosal / retinal melanomas by clinical and radiological examination.^[3] Herein, we present a rare case of Primary CNS melanoma in an albino patient who presented to Neurosurgery OPD with generalized headache.

CASE REPORT: A 52years old albino male patient presented to Neurosurgery department with repeated episodes of generalized headaches, vomiting and ataxia for duration of 3-4 months. MRI of the patient was performed and a mass lesion with T2 hypointense hemosiderin rim involving the cerebellum off midline that involved mainly left cerebellar hemisphere and vermis was detected. The mass measured 5.0x4.4x4.5cms. approximately on radiological examination. Based on these findings a diagnosis of Ependymoma was given on radiology. (Fig. 1a)

Intraoperative squash cytology and frozen section were performed. The intraoperative smears were highly cellular, composed of discohesive sheets of spindle to medium sized epithelioid like cells. These cells had prominent macronucleoli, intranuclear inclusions of cytoplasm and dense abundant eosinophilic cytoplasm. (Fig. 1b) Intracytoplasmic melanin could be identified easily. On Frozen section, sheets of malignant cells with eccentrically placed nuclei and punched out prominent nucleoli were seen. These cells displayed abundant eosinophilic cytoplasm and presence of fine to coarsely abundant brownish black pigment which bleached with potassium permanganate. (Fig. 1c &

CASE REPORT

d) Many mitotic figures were noted. Thus, an intraoperative diagnosis of malignant melanoma of CNS was made.

The tumour was resected in Toto and sent for histopathological examination. Grossly, excised tissue was a firm mass, dark brown to black in color measuring 5.5x5x4cms. (Fig. 2a) Microscopy revealed cellular malignant neoplasm showing sheets and loose nests of epithelioid like cells, ill defined fascicles of anaplastic spindle-shaped cells. These cells exhibited a moderate grade of cellular and nuclear pleomorphism, prominent macronucleoli and mitoses (>4/10 HPF). The cytoplasm showed abundant melanin pigment in the cytoplasm, which stained black with Masson Fontana.(Fig. 2b & c) Furthermore, Immunohistochemistry was performed employing S-100 and HMB-45 which exhibited strong and diffuse positive reaction.(Fig. 2d & e)

Thus, a confirmed diagnosis of Malignant Melanoma was made. Extensive search of primary comprising dermatological evaluation, ophthalmologic evaluation, X-ray chest and USG abdomen showed no primary lesion elsewhere. Based on these findings a confirmed diagnosis of Primary CNS Melanoma was given.

DISCUSSION: Albinism is an inborn error of metabolism that is genetically inherited and has an autosomal recessive (A-R) inheritance. It is due to deficiency of enzyme tyrosinase which is responsible for the hydroxylation of L-tyrosine to 3, 4, dihydroxyphenylalanine (DOPA) and its subsequent oxidation to dopaquinone. Subsequent polymerization of dopaquinone derivatives and its reaction with a specialized protein results in formation of melanoprotein in melanocytes.^[4] Albinos although have melanocytes but they do not contain melanin. Melanin acts as a sunscreen protecting the skin from the damaging effects of ultraviolet radiations hence warrants protection against various skin cancers such as squamous cell carcinoma (SCC), basal cell carcinoma (BCC), and malignant melanoma (MM). There have been sporadic occurrences of MM at different sites in albinos in literature such as metastatic malignant melanoma of the nasal cavity, choroidal malignant melanoma and primary malignant melanoma in lungs.^[5, 6] However, occurrence of Primary CNS melanoma in an albino patient has not yet been reported in literature searched.

Virchow first described the case of primary CNS melanoma in 1859, and labeled it diffuse Melanosarcoma.^[7] Primary intracranial melanomas are exceptionally rare. Most of the cases of CNS melanin bearing tumours are metastatic melanomas constituting 3-16% of secondary neoplastic foci in CNS, melanotic neurocutaneous syndromes and primary leptomeningeal melanoma.^[8, 9]

Primary melanocytic neoplasms arise from normally occurring leptomeningeal melanocytes which are derived from the neural crest during early embryonic development.^[10] Normal rests of pigmented cells within the CNS include the medulla, pons, substantia nigra, locus cerulus and the leptomeninges over the convexities, basal region and upper cervical spinal cord.^[1, 11] They may arise either de novo or in a pre-existing neurocutaneous melanosis.

These neoplasms are divided into three main types including diffuse melanocytosis, melanocytoma and malignant melanoma.^[1] From the histopathological point of view the principle differential is between melanocytoma and malignant melanoma.^[1, 12] Meningeal melanocytomas are histologically characterized as cellular lesions. They have variable amount of melanin pigment in the cytoplasm as intracellular granules. These lesions typically should not have nuclear pleomorphism, atypia or macronucleoli, necrosis and have very occasional mitotic figures (0-1/10 HPF). They usually have low MIB 1 staining (1-2%).

CASE REPORT

On the contrary, melanomas are more pleomorphic, have anaplastic nuclei with prominent macronucleoli. Mitosis is extensive (about 5-7/10 HPF) and MIB-1 labeling index (about 8.1%). A third subset of meningeal melanocytic tumours falls in the intermediate grade because they demonstrate intermediate histologic features. They have sheet like growth patterns, microscopic CNS invasion and occasional mitoses. MIB-1 staining ranges from 1%-4%. On Immunohistochemistry, all the three types are immunoreactive for HMB-45 and S-100 protein and negative for EMA.^[12]

For the diagnosis of primary CNS melanoma, Hayward criteria should be fulfilled that includes absence of melanoma outside the CNS, absence of melanoma in other sites in CNS and histologic confirmation of melanoma.^[13] Studies suggest that primary CNS melanomas grow slow and are less malignant than skin melanomas metastatic to CNS.^[14] Other primary tumours of CNS with melanotic elements originate from non-melanoma cells of CNS. They acquire melanin and these lesions include Meningioma, Medulloblastoma, Astrocytoma, Melanotic Schwannoma, Pituitary tumours and Choroid plexus Papilloma.^[14, 15] In conclusion, primary CNS melanoma is rare and its occurrence in an albino is still rarer that has not yet been reported.

REFERENCES:

1. Brat J, Perry A. Melanocytic lesions. In: Louis DN, Ohgkai H, Wiestler OD, Cavaene WK (ed). WHO Classification of Tumours of Central Nervous System. 4th ed. Lyon: IARC Press; 2007.p.181-3.
2. Schuster LM, Haliska F, Fraker D, Elenitias R. Skin: Malignant Melanoma In: Abeloff MD (ed) Clinical Oncology. New York: Churchill Livingstone, 2000: 1326-32.
3. Baena RR, Gaeliani P, Danova H. Primary solitary intracranial melanoma: Case report and review of literature. Surg Neurol 1992; 38: 26-37.
4. Soderman WA: Inherited diseases caused by deficiency of product. Metabolic Biochemistry in Pathologic Physiology Mechanism of Disease. Saunders 1979: p.91.
5. Casswell AG, Mc Carteny AC, Hungerford JL. Choroidal malignant melanoma in an albino. Br J Ophthal 1989; 73: 840-45.
6. Ozdemir N, Cangir AK, Kutlat H et al. Primary malignant melanoma of the lung in oculocutaneous albino patient. Eur J Cardiothoracic Surg 2001; 20: 864-7.
7. Becker SM, Emanuelli SJ, Sami AW. Primary melanoma of the leptomeninges of the spinal cord. A case report and review of literature. J Med SOC New Jersey 1970; 67: 271-75.
8. Biernat w. Metastatic tumours of the Central Nervous System- a pathological approach. Folia Neuropathol 2009; 47: 228-33.
9. Kiel FW, Starr LB, Hansen JL. Primary melanoma of the spinal cord. J Neurosurg 1961; 18: 616-29.
10. Aichner F, Schuler G. Primary leptomeningeal melanoma: diagnosis by ultrastructural cytology of cerebrospinal fluid and cranial computed tomography. Cancer 1982; 50: 1751-6.
11. Vanzielegheem BD, Lemmerling MM, Van Coster RN. Neurocutaneous melanosis presenting with intracranial amelanotic melanoma. AJNR AM J Neuroradiol 1999; 20: 457-60.
12. Brat DJ, Gianni C, Scheithauer BW et al.. Primary melanocytic neoplasms of the central nervous systems. Am J Surg Pathol 1999; 23: 745-54.
13. Hayward RD. Malignant Melanoma and the central nervous system. A guide for classification based on the clinical findings. J Neurol Neurosurg Psychiatry 1976; 39: 526-30.

CASE REPORT

14. Jaiswal S, Agarwal V, Vij M, Sahu RN, Jaiswal AK, Behari S. Glioblastoma with melanocytic differentiation. Clin Neuropathol 2010; 29: 330-33.
15. Smith AB, Rushing EJ, Smirnio to populous JG. Pigmented lesions of the central nervous system: radiologic- pathologic correlation. Radiographies 2009; 29: 330-3.

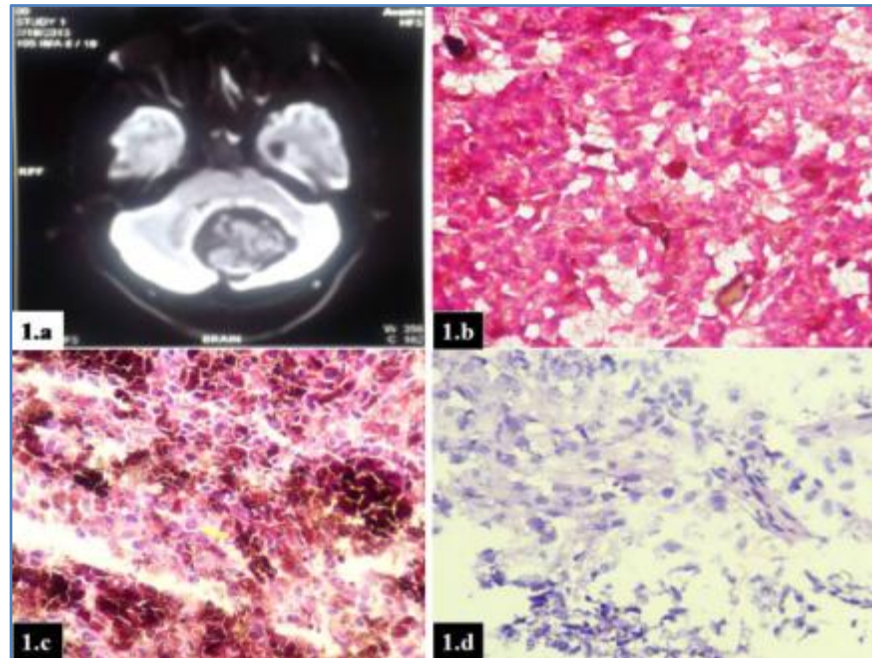


Figure 1

- a. MRI film showing a tumour in posterior fossa.
- b. Intraoperative squash smear showing elongated cells exhibiting hyperchromatic nuclei with prominent nucleoli (H & E X400).
- c. Frozen section showing sheets of malignant cells and both intracellular and extracellular melanin (H & E X400).
- d. Frozen section after bleaching resulting in removal of melanin pigment (potassium permanganate, x200).

CASE REPORT

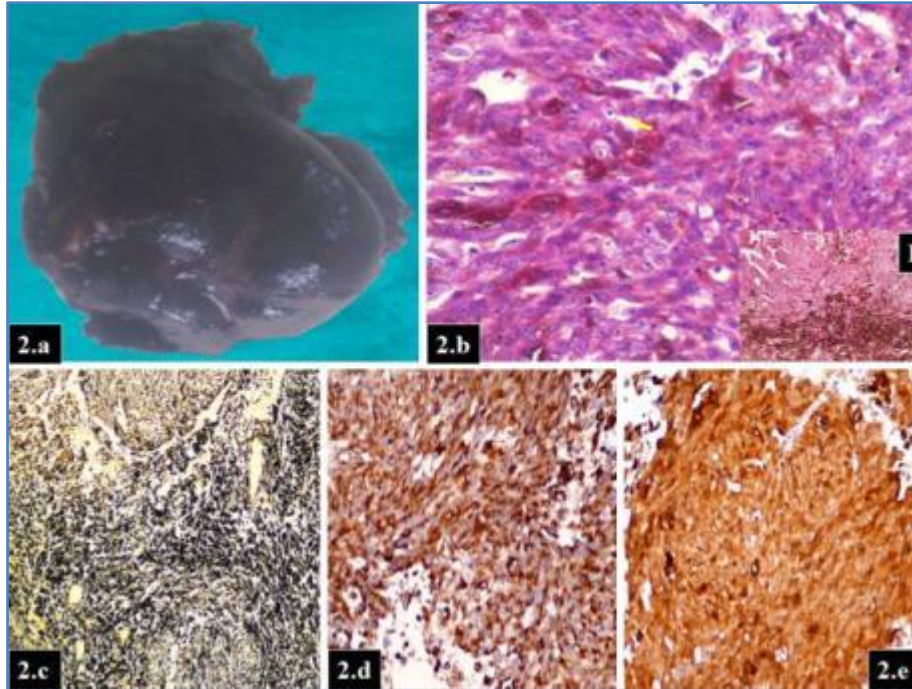


Figure 2

- a. Gross specimen of dark brown to black tumour mass measuring 5.5x5x4 cm.
- b. Section showing tumour cells in sheets and loose nests of epithelioid cells showing prominent macronucleoli (H & E X400); Inset (I) showing abundant intra & extracellular melanin pigment (H & E X100).
- c. Tumor cells showing abundant dark black pigment in cytoplasm (Masson Fontana x200).
- d. Diffuse S-100 immunoreactivity in the tumour cells (x400).
- e. Diffuse HMB-45 immunoreactivity in the tumour cells (x400).

AUTHORS:

1. Kishore Sanjeev
2. Bhardwaj Aparna
3. Gupta Mohit
4. Seema Acharya
5. Kudesia Sandip

PARTICULARS OF CONTRIBUTORS:

1. Professor, Department Pathology, SGRRIM & HS, Dehradun.
2. Associate Professor, Department of Pathology, SGRRIM & HS, Dehradun.
3. IIIrd Year Junior Resident, Department of Pathology, SGRRIM & HS, Dehradun.
4. Professor, Department of Pathology, SGRRIM & HS, Dehradun.

5. Professor & Head, Department of Pathology, SGRRIM & HS, Dehradun.

NAME ADDRESS EMAIL ID OF THE CORRESPONDING AUTHOR:

Dr. Aparna Bhardwaj,
Department of Pathology,
SGRRIM & HS, Dehradun,
Uttarakhand, India.
Email: aparnapande1977@gmail.com

Date of Submission: 12/06/2014.

Date of Peer Review: 13/06/2014.

Date of Acceptance: 27/06/2014.

Date of Publishing: 07/07/2014.