

### NEUROCUTANEOUS MELANOSIS: HYDROCEPHALUS AND INTRACRANIAL HYPERTENSION- RARE CAUSE

MELANOSE NEUROCUTÂNEA: UMA CAUSA RARA DE HIDROCEFALIA E HIPERTENSÃO INTRACRANIANA

Carlos dos Reis LISBOA NETO<sup>1</sup>, Raimundo Miranda de CARVALHO<sup>2</sup>, Joel Monteiro de JESUS<sup>3</sup>, José Reginaldo Nascimento BRITO<sup>4</sup>, Simone Mendes ROGÉRIO<sup>5</sup> e Alexandre Maués da COSTA<sup>6</sup>

#### SUMMARY

**Objective:** report a case of neurocutaneousmelanosis in Belém, Pará. **Case report:** a 13-years-old boy, with history 8 months later presented paroxistic absence recurrent seizures controlled with carbamazepine. At three years old, the seizures became generalized type, associated to nistagmus and incipient apendicular ataxia. Until 11 years old, he had neurologic exam stable, without diagnostic, when presented persistent holocranial headache, nausea, morning vomit, oftalmoparesis type III pair lesion on the right and somnolence. At general exam, it was found uncountable melanocytic nevi in his dorsum, buttocks, left arm, thorax and right shoulder. MRI study was performed and suggested CNS melanocytic infiltration. The patient underwent ventriculoperitoneal derivation and presented good evaluation, asymptomatic. **Conclusion:** the early diagnostic is very important, considering the possibility to precocious neurosurgical intervention, allowing the increase of survival in these patients.

**KEY WORDS:**neurocutaneousmelanosis, melanocytic nevi and hydrocephalus.

#### INTRODUCTION

Neurocutaneousmelanosis (NCM) is a very rare neurocutaneous syndrome. It is a phakomatosisnoninherited that was named by Van Bogaert in 1948. The entity occurs in both men and women at an even 1:1 ratio. In NCM, melanomas of the central nervous system are seen in association with congenital melanocytic nevi. The nevi are common on dermatomes distribution and often increase their size together the patient growing. However, they are classified into 3 types based on their final size: small (less than 1.5 cm in diameter), intermediate (1.5-20 cm in diameter), and giant or large (also called garment or bathing trunk nevi, measuring

more than 20 cm in diameter). Giant congenital melanocytic nevi occur in approximately 1 per 20.000 live births, corresponding to two-thirds of NCM, however the incidence of central nervous system involvement in giant nevi cases is unknown.

The syndrome represent a congenital error in differentiation of neural crest cells, which are the origin of melanocytes and of the basal leptomeninges among other tissues. Congenital abnormalities, such as Dandy-Walker malformation, Sturge-Weber syndrome and inferior vermian hypoplasia has been described in association with NCM. Histopathologic examination of the

- 
1. Médico graduado pela UFPA
  2. Residente de Neurocirurgia do Hospital Ophyr Loyola
  3. Neurocirurgião do Hospital Ophyr Loyola
  4. Neurocirurgião do Hospital Ophyr Loyola
  5. Neurocirurgiã do Hospital Ophyr Loyola
  6. Neurocirurgião do Hospital Ophyr Loyola

lesion shows accumulation of melanotic cells in the arachnoid and pia mater of the ventral surface of the mesencephalon, pons, medulla, cerebellum, upper cervical or lumbosacral spinal cord, resulting in leptomeningeal melanosis. Patients with CNS involvement may show lethargy, seizures, vomiting, or signs of raised intracranial pressure from hydrocephalus. Cranial nerve palsies frequently are associated. Once apparent, the symptoms are progressive and usually rapidly fatal, often with development of ataxia and loss of bowel and bladder control.

Hydrocephalus occurs in most patients secondary to meningeal thickening, which causes CSF outflow obstruction, or decreased CSF resorption. It has been calculated to occur in 64% of the patients. Most neurologic manifestations of increased intracranial pressure, mass lesions, and spinal cord compression appear in the first 2 years of life, with a second smaller late peak in the second and third decades of life.

According to Fox et al, diagnosis of neurocutaneous melanosis is established in patients with (1) extensive pigment nevus of the skin larger than 20 cm in diameter, and/or multiple congenital nevi; (2) no malignant transformation of the cutaneous nevus; (3) no primary malignant melanoma; and (4) proven histological central nervous system lesion. Therefore, the criteria were slightly extended, including those cases with cutaneous melanoma but without signs of malignancy in the leptomeninges and those with primary melanoma of the meninges but without cutaneous malignancies.

The color of the nevi is brown-black in raised plaques with small nodules, and an increased hair growth may be present. Neuroimaging with CT or rather with MRI must be performed. MRI findings most commonly described are thickened leptomeninges surrounding the brain and spinal cord, ventricular dilatation, involvement of the cerebral parenchyma and inferior vermian hypoplasia..

## CASE REPORT

A 13-years-old boy, student, right hand prefer, with history 8 months later presented paroxistic absence recurrent seizures controlled with carbamazepine. At three years old, the seizures became generalized type, associated to nistagmus and incipient appendicular ataxia. He underwent MRI study that showed Dandy-Walker complex and brief ventricular increase. Until 11 years old, he had neurologic exam stable, without diagnostic, when presented persistent holocranial headache, nausea, morning vomit, ophthalmoparesis type III pair lesion on the right and somnolence. In that occasion was performed cranial CT (neuroimage 1), that showed ventricular dilatation and similar MRI study findings. At general exam, it was found uncountable melanocytic nevi in his dorsum, buttocks, left arm, thorax and right shoulder, the biggest nevi, with 8 cm. He underwent spinal manometry that showed beginning pressure 55cmH<sub>2</sub>O. It was removed 30 ml cerebrospinal fluid and the patient kept asymptomatic for 10 days. Thinking in NCM diagnostic hypothesis, a new encephalic MRI study was performed and showed the lesions already described, this time, associated to focus hypersignal in T2-weighted image in both temporal lobes and diffuse leptomeningeal enhancement, that suggested CNS melanocytic infiltration. The patient underwent ventriculoperitoneal derivation, with medium pressure catheter and presented good evaluation, asymptomatic.

## DISCUSSION

From the point of etiopathogenic view, it is supposed that occur a congenital error in formation and differentiation of neural crest cells during the beginning of embryologic period, when the cutaneous melanocytes precursor cells and leptomeningeal precursor cells, the melanoblasts, are formed. The NCM can be associated with Dandy-Walker complex, like in the case reported, and Sturge-Weber syndrome.

The syndrome generally becomes symptomatic when the CNS lesions affect the CSF flow, presenting signs and

symptoms associated to raised intracranial pressure, like headache, irritability, lethargy and recurrent vomits. The MNC also can show seizures, often before the 2 years old, but can be observed in second decade of life, like in the case reported.

Patients with melanocytic nevi situated in posterior axial region, mainly in lumbosacral place are considered of risk to NCM. It is recommended the CNS lesion screening with encephalic and spinal MRI study.

LCMN have been reported to possess a propensity for malignant degeneration. Clinicians generally concur that LCMN should be prophylactically excised to prevent malignant transformation. One early study reported that roughly 50% of the malignancies that occur in LCMN present within the first 5 years of life.

In relation to prognosis, 70 % of patients die before 10 years old, because of hydrocephalus and intracranial hypertension, or because of the lesions malignant transformation. After the beginning of symptoms, the prognosis becomes bad, with 3 years of medium time survival. Some patients present untreatable hydrocephalus, evolving to death early.

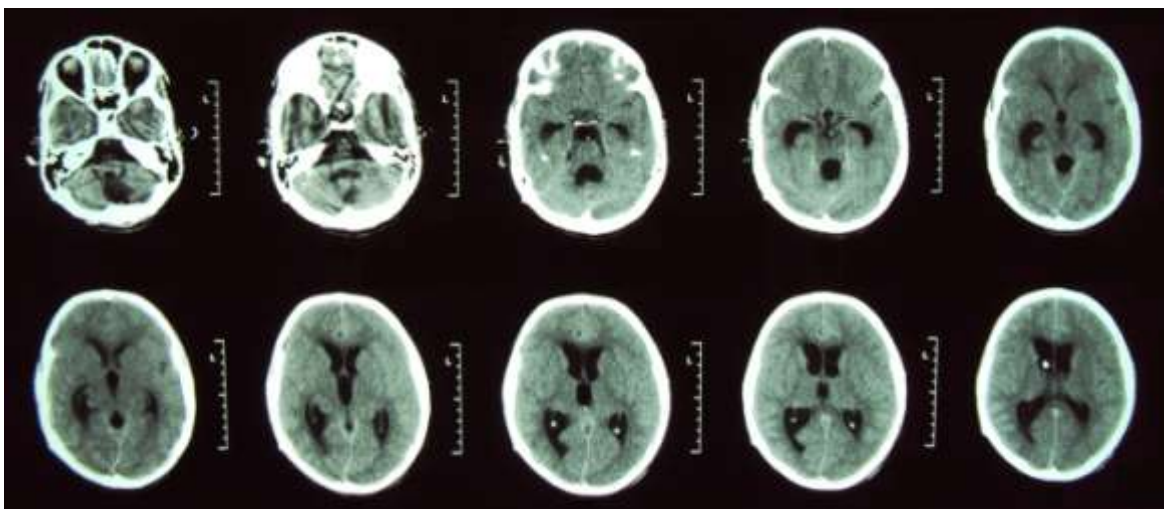
Although the prognosis is poor, early neurosurgical involvement in these

patients may provide tissue diagnosis and the potential for decompression if the process is caught early in its course.

Because of the importance of NCM early diagnostic, it is very important that clinicians make the CNS lesions screening with encephalic and spinal MRI study, when observe the melanocytic nevi during the physical examination. It is also important remember the large variety of neurocutaneous syndromes that affect people, however stay without diagnostic during big period, worsening the prognosis and chance of appropriated treatment.

### CONCLUSION

The NCM is an extremely rare entity, with little studies published in literature, around 150 cases reported. The early diagnostic is very important, considering the possibility to precocious neurosurgical intervention, specially in CNS malignant melanoma and hydrocephalus, allowing the increase of survival in these patients. Periodic following is indispensable to identification of malignant lesions.



**Neuroimage 1** – Cranial CT showing ventricular dilatation compatible with hydrocephalus.

## RESUMO

**Objetivo:** relatar um caso de melanoseneurocutânea em Belém, Pará. **Relato de caso:** adolescente, 13 anos de idade, gênero masculino, com história de crises convulsivas paroxísticas, tipo ausência, recorrentes, controladas com uso de carbamazepina. Aos três anos de idade, as crises tornaram-se do tipo generalizada, associada com nistagmo e ataxia apendicular incipiente. Até 11 anos de idade, o paciente teve quadro neurológico estável, sem diagnóstico, quando apresentou cefaléia persistente holocraniana, náusea, vômitos pela manhã, sonolência e oftalmoparesia por lesão do III par craniano à direita. No exame geral, foram encontrados incontáveis nevos melanocíticos no dorso, nádegas, braço esquerdo, tórax e ombro direito. A RNM sugeriu infiltração melanocítica do SNC. O paciente foi submetido a derivação ventrículo-peritoneal e apresentou boa evolução, ficando assintomático após o procedimento. **Conclusão:** o diagnóstico precoce é muito importante, considerando a possibilidade de intervenção neurocirúrgica precoce, permitindo o aumento da sobrevida nesses pacientes.

**DESCRITORES:** melanoseneurocutânea, nevo melanocítico e hidrocefalia

## REFERENCES

1. Chu, WCW; Lee, V; Chan, Y; Shing, MMK; Chik, K; Li C; Ma, K. Neurocutaneous Melanomatosis with a Rapidly Deteriorating Course. AJNR Am. J. Neuroradiol, n 24: 287 – 290, Feb 2003
2. D'Argenio, A; David,.; Engohan, C; Hennequin, Y; Balériaux, D; JissendI, P. Neurocutaneous melanosis in a newborn with giant congenital melanocytic nevus. Journal of Neuroradiology, n 43: 272-275, 2007.
3. Burstein, F; Seier, H; Hudgins, PA; Zapiach, L. Neurocutaneous Melanosis. The journal of craniofacial surgery, vol 16, n 5: Sep 2005.
4. Heffel, DF; Thaller, S. Congenital Melanosis: An Update. The journal of craniofacial surgery, vol 16, n 5: Sep 2005.
5. Pavlidou, E; Hagel, C; Papavasilliou, A; Giouroukos, S.; Panteliadis, C. Neurocutaneous Melanosis: Report of Three Cases and Up-to-date Review. Journal of Child Neurology, vol 23, n 12: 1382-1391, Dec 2008.
6. Noronha, L; Sampaio, GA; Netto, MRM; Reis-Filho, JS; Faoro, LN; Raskin, S; Bleggi-Torres, L. F. Neurocutaneous melanosis. Jornal de Pediatria - Rio de Janeiro Vol. 75, Nº4, 277-280, 1999
7. Makin, GWJ; Eden, OB; Lashford, LS; Moppett, J; Gerrard, MP; Davies, HA; Powell, CVE; Campbel, AN; Child, HF. Leptomeningeal Melanoma in Childhood. CANCER, Vol. 86, n 5, Sep. 1999
8. Demirci, A; Kawamura, Y; Sze, G; Duncan, C. MR of Parenchymal Neurocutaneous Melanosis. AJNR 16:603–606, Mar 1995 0195-6108/95/1603–0603 q American Society of Neuroradiology
9. Ahuja, SR; Karande S; Kulkarni, MV. Multiple giant congenital melanocytic nevi with central nervous system melanosis: A case report. Neurol India, 51:541-3, 2003
10. Ko, SF; Wang, HS; Lui, TN; Ng, SH; Ho, YS; Tsai, CC. Neurocutaneous melanosis associated with inferior vermian hypoplasia: MR findings. J Comput Assist Tomogr. 17:691–695, 1993.

### Endereço para correspondência:

Carlos dos Reis Lisboa Neto  
Tv. Lomas Valentinas 1412 – Marco – Belém – Pará  
Fone: +55 (91) 3276-9515  
E-mail: [carlosnetolisboa@gmail.com](mailto:carlosnetolisboa@gmail.com)

Recebido em 10.11.2011 – Aprovado em 02.02.2012