

# “Inspect the skin, but don’t forget the brain”

## A case of Giant Congenital Melanocytic Nevus in a newborn

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### CASE REPORT

Newborn male  
Non-consanguineous parents  
No familial history of congenital nevi

**Clinical examination:**  
Giant congenital melanocytic nevus on trunk, genital area, thighs  
Smaller melanocytic lesions, so-called satellite nevi, on face, scalp, left arm/hand  
No proliferative nodules or neuroid overgrowth present

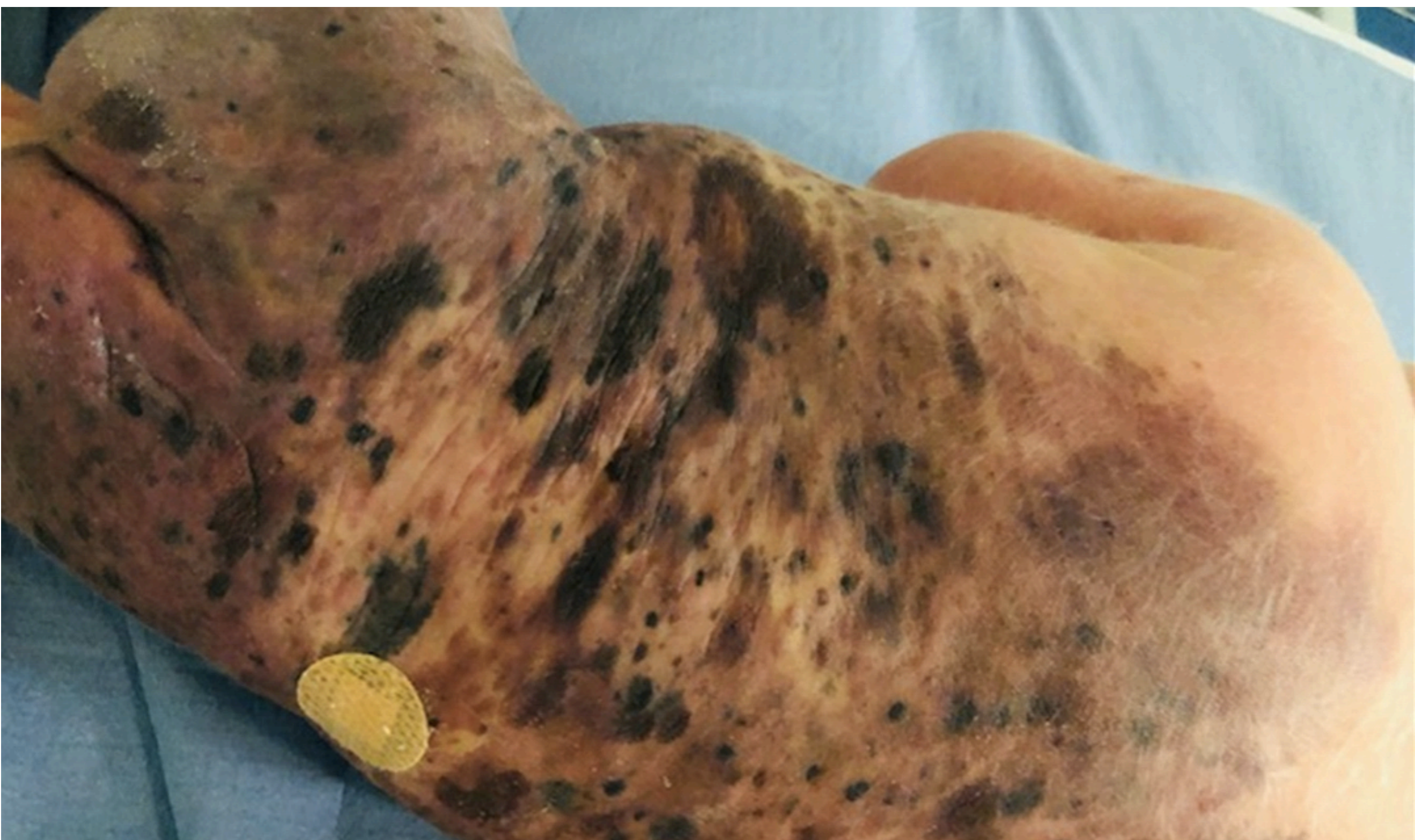
**Diagnostic workup:**  
Biopsies at day 2 (in a dark and a red lesion):

- Histology: congenital melanocytic nevus, no signs of malignancy
- Genotyping: NRAS +, BRAF -

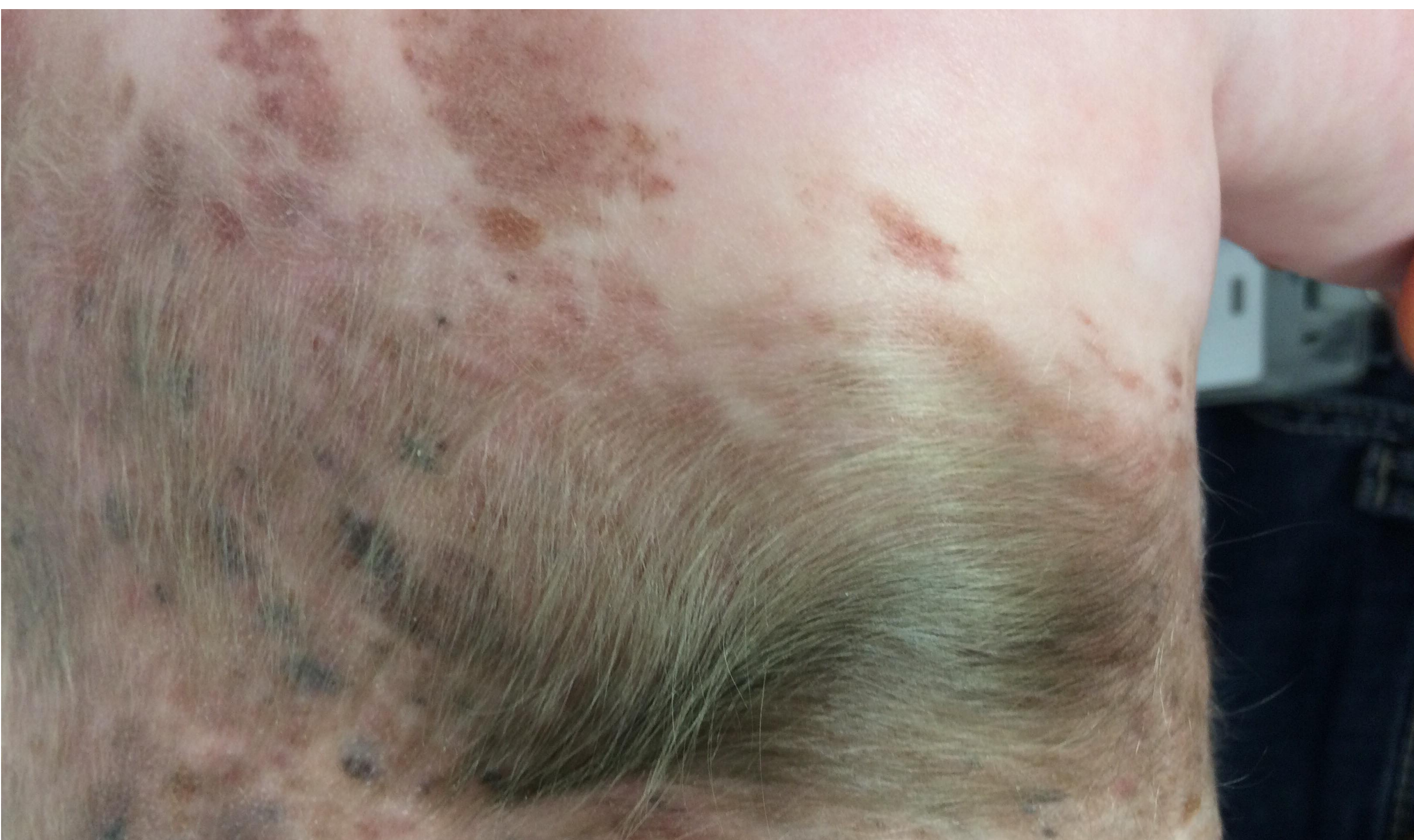
MRI brain & whole-spine at day 4: no central nervous system abnormalities



Day 1: red - blue - purple macules and plaques



After 2 weeks: evolution to brown - black macules and plaques



After 5 months: brown - black macules and plaques with pronounced hypertrichosis with darkly pigmented terminal hairs

### DISCUSSION

- **Giant Congenital Melanocytic Nevi:**  
Melanocytic lesions at birth that become  $\geq 20$  cm in projected adult size (*other definitions are available, but many authorities currently favor this definition*). The estimated incidence is 1 in 20.000 to 500.000 live births.
- **Melanoma risk:**  
Very low in small single congenital melanocytic nevi, but the lifetime risk is up to 10-15% in giant congenital melanocytic nevi of  $> 40$  cm in projected adult size with several smaller congenital melanocytic nevi. Melanoma are most common in the first 5 years. One-third of these melanoma patients develop a primary central nervous system tumor. The melanoma risk is higher in those with congenital central nervous system abnormalities.
- **Workup at birth:**
  - **Skin examination:** advisable with high quality photography material for comparison.
  - **MRI brain & whole-spine:** recommended in every patient with  $\geq 2$  congenital melanocytic nevi at birth independent of size/location before the age of 6 months because of the better visualization before full myelination. The strongest risk factor for all-site melanoma is an abnormal MRI screening of the central nervous system in the first year of life.
- **Genotyping:**  
NRAS mutations are a very common cause of congenital melanocytic nevi, especially with projected adult size  $> 60$ cm. The clinical outcomes are not different among various genotypes (*clear data for neurological disease, suggestive data for melanoma risk*). Genotyping should be performed when melanoma is suspected and certainly when treatment is required, but not as part of routine clinical care.

### CONCLUSION

- **Clinical examination** might be challenging at birth due to the erythematous and bluish skin in newborns.
- Prompt referral for **MRI** is indicated for central nervous system screening before the age of 6 months.
- **Genotyping** (NRAS/BRAF) should be done when melanoma is suspected and treatment is needed, but not routinely.

### REFERENCES

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**MARIA  
MIDDELAES**

Gezondheids**Zorg** met een Ziel