Melanoma in a patient with septo-optic dysplasia: a case report

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Introduction

Septo-optic dysplasia (SOD) is a rare congenital disorder characterized by midline neural structural defects. It has a diverse presentation and frequently leads to dysfunction in the hypothalamic-pituitary axis.[1]

Melanoma is a malignancy of the melanocytes in the skin and has several known risk factors, including fair skin, UV exposure, and a family history. Here, we present the case of a 44-year-old Caucasian male with a history of SOD who was diagnosed with pT3b melanoma, a combination not previously described in the literature.

Case Presentation

A 44-year-old Caucasian man presented with a dark mole on his left lateral neck that had been present for years and had recently started to scab.

- PMHx: SOD with adrenal insufficiency and hypothyroidism on prednisone and levothyroxine
- FHx: father had melanoma, no known family history of SOD
- Denies significant history of sunburns, works indoors in IT

- Physical exam: approximately 0.7 x 0.6 cm, red papule-like lesion on the left central lateral neck. No palpable cervical lymphadenopathy noted. Fitzpatrick 1 skin type (see figure 1).

- Dermatology obtained shave biopsy demonstrating ulcerated melanoma with invasion depth of 2.1mm.

- Patient was staged at pT3b, with plans for a wide-local excision (WLE) and sentinel lymph node biopsy (SLN).

Intervention

- Patient underwent WLE and SLN, which yielded 2 lymph nodes. Pathology of WLE was negative for residual melanoma and lymph nodes demonstrated no metastatic disease.

- Given his locally advanced disease, he was referred to medical oncology for adjuvant immunotherapy. He is currently planning on starting pembrolizumab at this time.

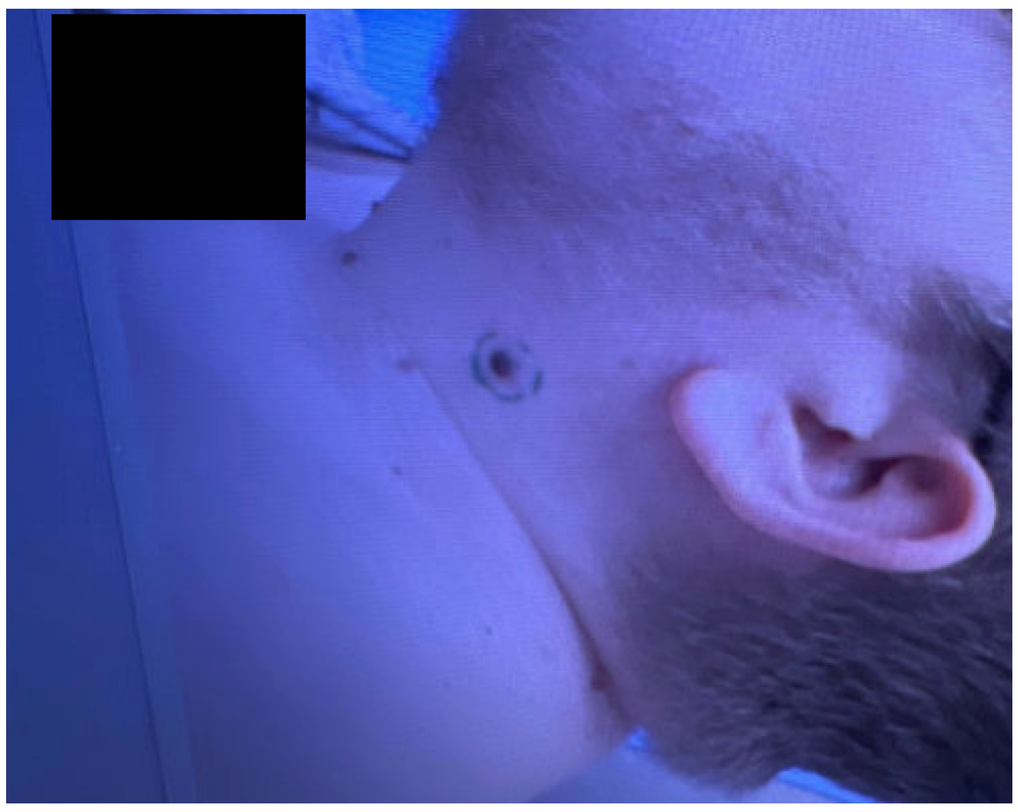

Figure 1: Left lateral neck melanoma marked for surgery. Note Fitzpatrick 1 skin type.

Discussion and Conclusion

SOD occurs in 1 in 10000 newborns and consists of a triad of:
(1) optic nerve hypoplasia, (2) agenesis of the corpus callosum and septum pellucidum, and (3) hypoplasia of the hypothalamic-pituitary axis.[1]

- Our patient was diagnosed at age 19-20 via brain MRI demonstrating absence of the septum pellucidum and hypoplasia of the optic nerves. Hormonal blood testing including thyroid labs, cortisol level, ACTH level, and LH level demonstrated HPA dysfunction.

Of note, patient's central adrenal insufficiency appears to result in reduced MSH production.
- May contribute to patient's pale skin, increasing risk for melanoma

Several genes are implicated in the pathogenesis of SOD, one of which is SOX2.[2]
- Downregulation results in pituitary hypoplasia is seen in SOD
- Normal adult melanocytes express low levels of the gene
- Increased expression in melanoma has been linked with more aggressive tumor behavior[3]
- Our patient may have low baseline SOX2 expression, which would be protective against metastatic spread and aggressive tumor behavior

This unique interplay of factors has never been documented before in the literature.

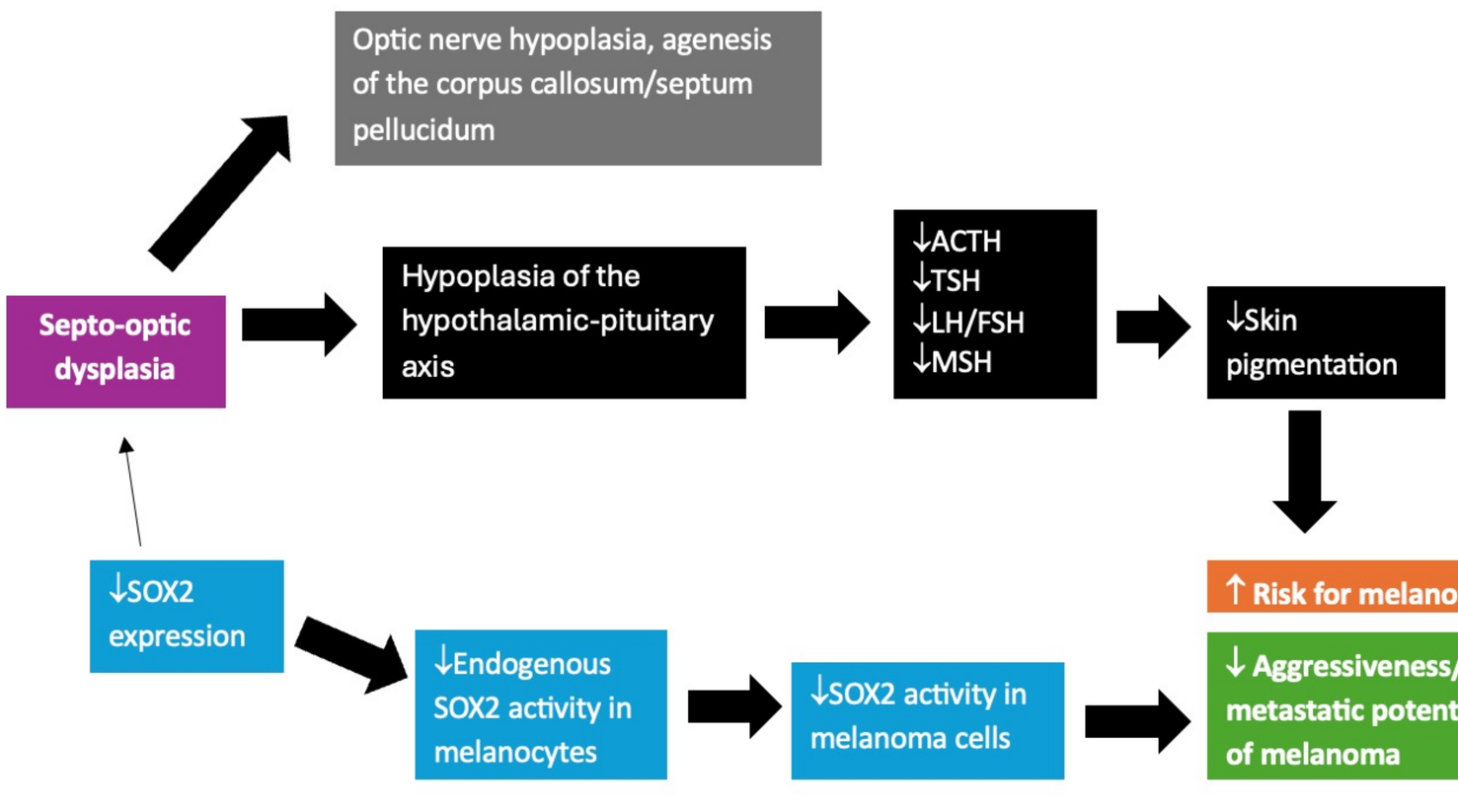

Figure 2: Flowchart demonstrating the interplay between this patient's unique characteristics contributing to melanoma pathogenesis

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References

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3. Utikal, J., et al. (2009). "Sox2 is dispensable for the reprogramming of melanocytes and melanoma cells into induced pluripotent stem cells." Journal of Cell Science 122(19): 3502-3510.

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