Congenital choroidal melanoma in an infant

While malignant melanoma is the most common primary ocular tumour in adults, it is exceedingly rare in the pediatric population. A study of more than 8000 patients with uveal melanomas revealed a 0.8% incidence in patients between 3 and 20 years of age.1 Some patients had a history of familial melanoma syndrome.1

We report a case of congenital melanoma in an infant. The 7-month-old male had multiple pigmented skin lesions indicating familial melanoma syndrome. The patient, from rural Yemen, presented in Yemen with a right ocular tumour and underwent enucleation. The patient had multiple atypical cutaneous moles on his face, trunk, and feet, consistent with familial atypical multiple mole and melanoma (FAMMM) syndrome (Fig. 1). Following enucleation and initial review, the slides were forwarded to the Department of Pathology, University of Western Ontario (London, Ont.), for assessment. The material was further reviewed at the Department of Pathology, McGill University (Montreal, Que.).

The tumour had a basal diameter of at least 2.0 cm and a height of 0.6 cm. Details of gross description were not available. Histologically, the tumour was a heavily pigmented invasive malignant melanoma of the choroid, composed of both spindle and epithelioid cells (Fig. 1). The majority of cells were either small or large epithelioid cells. Mitotic figures were identified. No necrosis and no vascular or neuronal invasion were seen. There was a clear invasion of the underlying sclera (Fig. 1). Immunocytochemically, Melan-A stain was positive in the neoplastic cells. Based on the age of the patient and presence of skin lesions, a diagnosis of congenital choroidal melanoma was favoured. Unfortunately, biopsy of the cutaneous lesions was not undertaken, and the patient was lost to follow-up. A literature review found 4 reported cases of congenital uveal melanomas (Table 1).2–5

FAMMM syndrome (B-K mole or dysplastic nevus syndrome) presents with multiple atypical nevi or primary melanomas that affect multiple organ systems, including the skin and the eye. Patients may have hundreds of moles and a significant family history of melanoma.6 There is also an increased risk for other primary cancers, namely pancreatic cancer. With FAMMM syndrome the median age for the development of melanoma is 10–15 years younger than the median age of the general population with sporadic melanoma.6 In a retrospective review of 4600 patients with primary uveal melanoma, 8 had biopsy-verified FAMMM syndrome, and the median age for development of uveal melanoma with FAMMM syndrome was 41 years of age (range, 10–52 years).7 If a patient develops intraocular melanoma with FAMMM syndrome, the patient’s family requires annual ophtalmological examinations.6 Our patient represents the fifth documented case of congenital uveal melanoma, and our patient is likely the youngest documented presentation of FAMMM syndrome.

FAMMM syndrome demonstrates autosomal dominant inheritance with incomplete penetrance. Although melanomas in children respond favourably to chemotherapy, prediction of exact behavior is difficult.5 Molecular biology studies have implicated the 9p21 locus with mutation of the p16/CDKN2A gene.6 In addition, certain variants of the low penetrance gene MC1R act as a genetic modifier.8 In FAMMM syndrome the penetrance of intraocular melanoma is unknown. The sensitivity and specificity of commercially available tests for this mutation have not been

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Table 1—Previously reported cases of congenital uveal melanoma

<table>
<thead>
<tr>
<th>Study</th>
<th>Demographics</th>
<th>Ocular tumour</th>
<th>Skin lesions</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Greer²</td>
<td>Male, birth</td>
<td>Anterior uvea</td>
<td>Multiple melanocytic nevus</td>
<td>No systemic disease, alive at 2 years</td>
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<tr>
<td>Broadway³</td>
<td>Male, birth</td>
<td>Uveal tumor with orbital extension</td>
<td>Multiple melanocytic nevus</td>
<td>Liver metastasis, alive at 2 years, 10 months</td>
</tr>
<tr>
<td>Posnick⁴</td>
<td>Female, birth</td>
<td>Orbital mass, extensive extraocular involvement</td>
<td>None reported</td>
<td>Extension to maxilla, alive at 2 years</td>
</tr>
<tr>
<td>Palazzi⁵</td>
<td>Male, birth</td>
<td>Uveal tumour with extraocular involvement of muscles and optic nerve</td>
<td>Multiple melanocytic nevus, 2 skin melanomas</td>
<td>No systemic disease, alive at 10 years</td>
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established and the American Society of Clinical Oncology does not currently recommend screening for this mutation.6

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