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The hourglass facial deformity as a consequence of orbital irradiation for bilateral retinoblastoma

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Abstract We report a case illustrating characteristic facial deformities following irradiation during infancy for bilateral retinoblastoma. CT and MR imaging revealed distinctive features of the resulting "hourglass facial deformity": hypotelorism, enophthalmos, depressed temporal bones, atrophy of the temporalis muscles, and a depressed nasion. This case also displayed premature metopic craniosynostosis and frontal sinus aplasia; these may be additional hallmarks of this deformity.

Introduction

Radiation changes are well recognized in endochondral bone of the spine, iliac crest, and long bones [1], but little attention has been paid to radiographic evidence of growth retardation in developing membranous bones caused by orbital irradiation. We report a case illustrating characteristic facial deformities following irradiation during infancy for bilateral retinoblastoma. This includes hypotelorism, enophthalmos, depressed temporal bones, atrophy of the temporalis muscles, and a depressed nasion. These distinctive features are termed the "hourglass facial deformity".

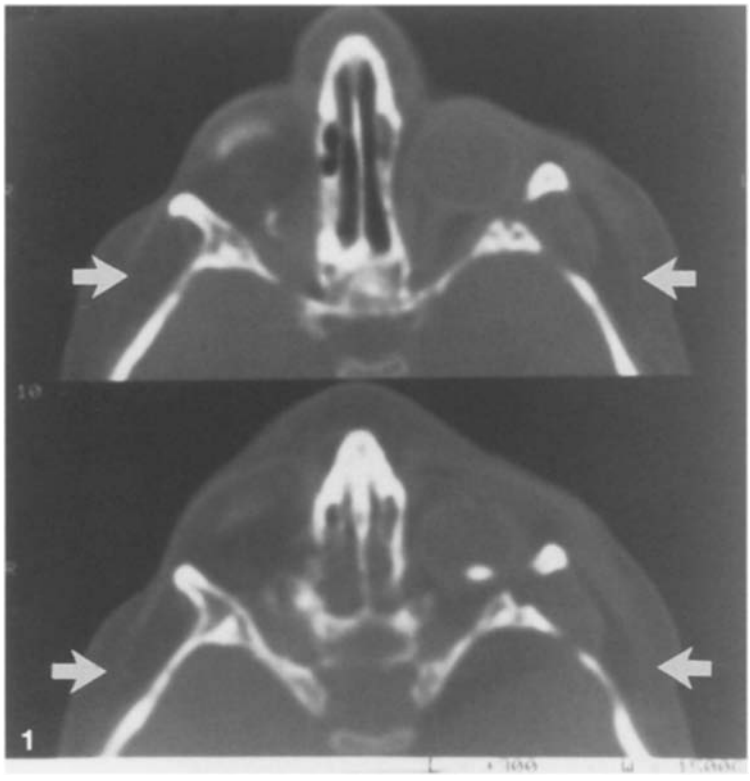
Case report

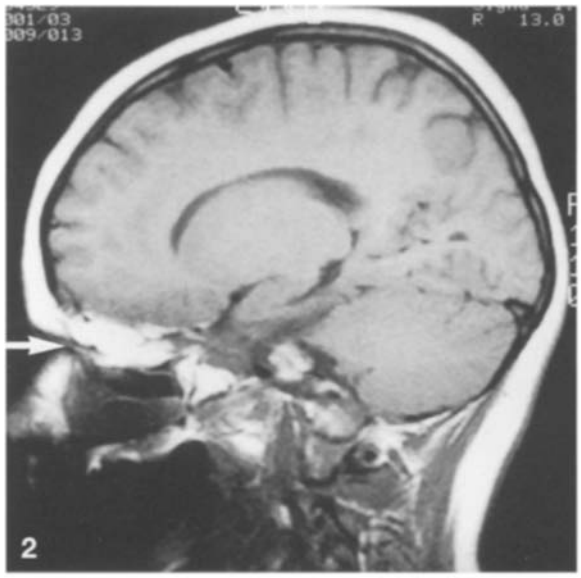
Our patient is a 14-year-old girl who was diagnosed with bilateral retinoblastoma at the age of 5 months and was irradiated at that time with 4000 rads (40 Gy) to the posterior right orbit and the posterior left globe after right-sided enucleation. She was fitted with a prosthesis in her right globe at the age of 2 but was unable to wear the prosthesis by the age of 7 due to "shrunken orbits and temporal bones" (as described in the clinic notes). A CT scan of the orbits (Fig. 1) was performed at the age of 9 which showed enophthalmos, hypotelorism, temporalis muscle atrophy, and depression of the temporal bones. MR imaging of the brain was performed at the age of 13, which showed progression of the orbital deformity with depression of the nasion in profile (Fig. 2), as well as the characteristic hourglass configuration of the face and calvarium (Fig. 3).

Discussion

Orbital irradiation is commonly utilized as an adjuvant treatment regimen after enucleation in children with orbital retinoblastoma. The radiation fields often cover both retinae because of the possibility of multiple tumors in one or both eyes [2]. Retardation of bony and soft tissue growth of the mid face, which includes both orbits, ethmoid bones, and the nasal bridge, is typically a delayed finding in affected children. The radiologic findings of hypotelorism, enophthalmos, depressed temporal bones, atrophy of the temporalis muscles, narrow and deep orbits, and a depressed nasion can first be seen at the end of the first decade and progress through adolescence.

This constellation of findings was first described as a consequence of orbital irradiation by Ju et al., who noted that the appearance is so characteristic that all the patients "bear a strong resemblance to each other, as if they were siblings" [3]. Recently, Guyuron has aptly termed this the "hourglass facial deformity" (Fig. 3) [4]. We have also noted a slightly triangular deformity of the forehead in our patient due to premature metopic craniosynostosis, as well as aplasia of the frontal sinuses.





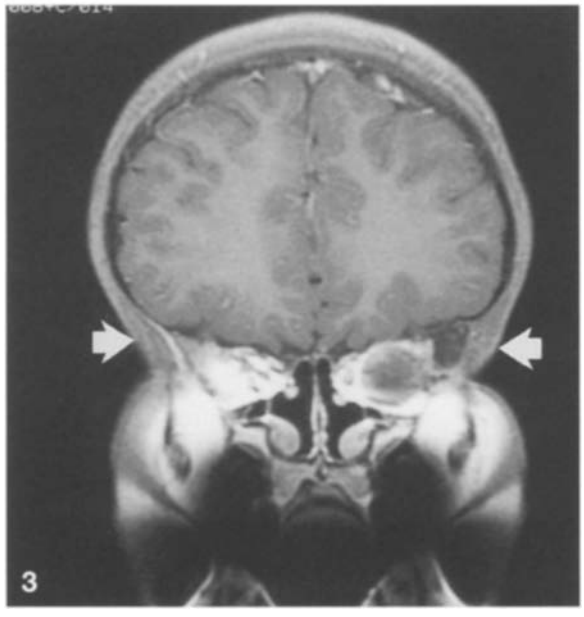


Fig. 1 CT scan of the orbits shows enophthalmos, hypotelorism, and temporalis muscle atrophy *(white arrows)*. Narrowing and deepening of the right orbit has caused the socket to become nonfunctional. Incidentally noted is calcification of the left retina from treated retinoblastoma

Fig. 2 Sagittal T1-weighted MR image (TR/TE 520/18) shows characteristic depression of the nasal bridge *(open white arrow)*, which is included in the radiation field. The nasal alae are spared

Fig. 3 Coronal T1-weighted MR image (TR/TE 520/18) shows mid-face hypoplasia with hypotelorism and depressed temporal bones, as well as temporalis atrophy *(white arrows)*. The resulting hourglass configuration is well demonstrated

Although radiologists are familiar with secondary bone tumors and cataract development in long-term survivors of orbital irradiation, little is mentioned in the radiographic literature on growth retardation in these patients. The early literature indicates hypoplasia of the treated orbit, along with varying degrees of growth arrest of the adjacent maxillary, nasal, and temporal bones [1]. Although relatively little information is available on its effect on developing membranous bone, radiation is thought to effect small blood vessels in the richly vascularized precursor connective tissue, causing irregular production of osteoid and faulty bone production. Harmful doses for soft tissue and bone are estimated to occur at 400 and 3000 rads (4 and 30 Gy), respectively [5]. Radiation-induced arrest of bone growth can lead to asymmetry in size and volume of the bony orbit. Scarring and contracture of the intraorbital soft tissues narrow the socket, and periorbital soft tissue contractures make the enophthalmos more pronounced. Continued deepening of the patient's orbit with growth ultimately results in the eye socket becoming nonfunctional [4]. Growth retardation of the mid face (orbits, ethmoidal bones, and nasion) contributes to hypotelorism, while growth retardation of the frontal bones is related to the triangular deformity of the forehead as well as frontal sinus aplasia. Frontal sinus aplasia has not previously been noted in the literature as a consequence of orbital irradiation, but the two radiographs available from an

extensive literature search also show this finding in the mature cranium [1, 6]. This is probably due to the fact that frontal sinuses develop late, long after the usual age of irradiation for retinoblastoma. We postulate that this may be a common finding in bilateral orbital irradiation.

Due to earlier detection and improved treatment regimens, the mortality of retinoblastoma has decreased from 60 % in the 1920s to 10 % today [7]. Treatment approaches are now more concerned with morbidity as well as cosmetic and functional results. Since radiotherapy involving doses of 3500–5000 rads (35–50 Gy) is still often used [2], new patients with this deformity will continue to be identified. Early detection of radiation-induced mid-face hypoplasia is an important step in improving the cosmetic prognosis of these patients. Various surgical techniques exist to correct the deformity, but early detection is essential for a good cosmetic result. Therefore, it is important for radiologists to be aware of this deformity to aid in a timely diagnosis and treatment.

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