

not unusual for a pet boa constrictor to strike its owner; however, it is usually the fault of the keeper for disregarding the natural instincts of the snake. The boa constrictor is active at dusk and during the night when it lies in wait for its prey. If the boa keeper tries to handle the snake after the vivarium light has been switched off they may be bitten. The same can happen if the keeper's hand smells of boa food, for example after handling a rat or mouse. Furthermore, there exist very aggressive specimens, which frequently strike their owner. There are also certain periods when even docile snakes are more aggressive, for example when shedding their skin. The boa has two types of bite—the prey bite and the defence bite. In the former, the jaws lock shut on the prey while the snake coils itself around its victim to suffocate it by constriction. The defence bite is an open-mouthed strike against a threat, and usually the snake recoils after the attack (Hermann Stöckl, personal communication).

Our patient was attacked as he reached into the vivarium to change the water, but he had been preparing a rat to feed to the snake just prior to this. The pattern of teeth marks on our patient's face suggests an open-mouthed defence strike. Unfortunately for the boa, and our patient, the snake's fangs became impaled on the patient's eye and face and caused the injuries described above.

Penetrating eye injuries following animal bites are unusual, even in the presence of extensive adnexal trauma.^{8,9} In this case, despite minimal adnexal trauma, the eye was seriously injured. The pattern of teeth marks crossing the medial canthus towards the globe is the sign that should alert the clinician to involvement of the eye.

Dog bites cause large, complex corneo-scleral lacerations with prolapse of, or severe damage to, the intraocular structures.⁷ In contrast, cat bites result in puncture wounds that may inoculate infective organisms deep into the tissues.⁶ The pattern of globe injury caused by the snake bite is therefore most similar to a bite from a cat, with puncture wounds from needle-like teeth. Fortunately for our patient, there was no sign of infection.

We have presented the first case of penetrating eye injury caused by a snake bite. The attack was probably precipitated by poor handling techniques on the part of the owner. All keepers of unusual or dangerous animals should understand the behaviour of their chosen pet.

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Sir,

Consanguinity and disease coincidence

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Increased homozygosity leads to the expression of recessive genes in the children of consanguineous unions. There is also the possibility of noninherited genetic disease in such patients, and this should not be overlooked.

Case report

An 11-month-old girl was initially referred to us in 1988 with nystagmus, recently noticed by her 35-year-old mother. She was systemically well as were her five elder siblings. Her parents are second cousins, but there was no family history of ocular or systemic pathology (Figure 1).

Examination confirmed the presence of horizontal jerk nystagmus and revealed bilateral iris transillumination.

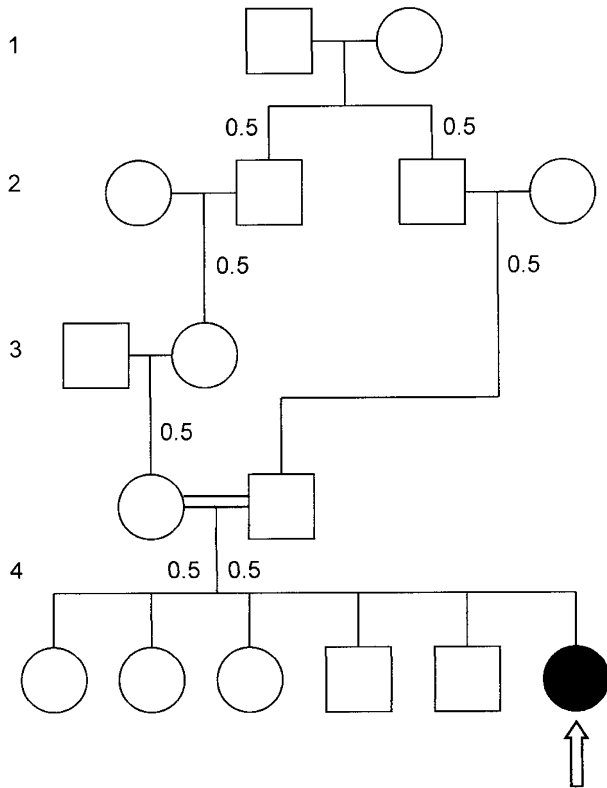


Figure 1 Family tree demonstrating recessive allele transmission probabilities.

Her family is of Pakistani origin, and her irides were a paler brown colour than might have been considered the usual norm. No hypopigmentation of her skin was noted.

She was followed up over the next few years and found to have hypoplastic maculae (Figure 2a).

By the age of 3 she was noted to have developed a right posterior pole lens opacity (Figure 2b). The vision in each eye was 6/60 with a small hypermetropic astigmatic correction, achieving 6/36 binocularly, and it was felt that there would be no benefit to be gained from surgery for this cataract. A diagnosis of ocular albinism was made, presumed to be autosomal recessive, with a coincidental left lens opacity. We have not as yet been able to perform the visual evoked potential examination that would be necessary to demonstrate the abnormally increased decussation of retino-geniculate axons at the chiasm, which would be expected to confirm the clinical diagnosis of ocular albinism.

By the age of 8, systemic features brought her to the attention of the clinical genetics department with learning difficulties, height below the 1st centile, head circumference on the 10th centile, a single subcutaneous nodule on her chest wall, enlargement of her left labia majora and cutaneous lesions. These consisted of five raised, hairy lesions, 5–15 mm in diameter on her arms,

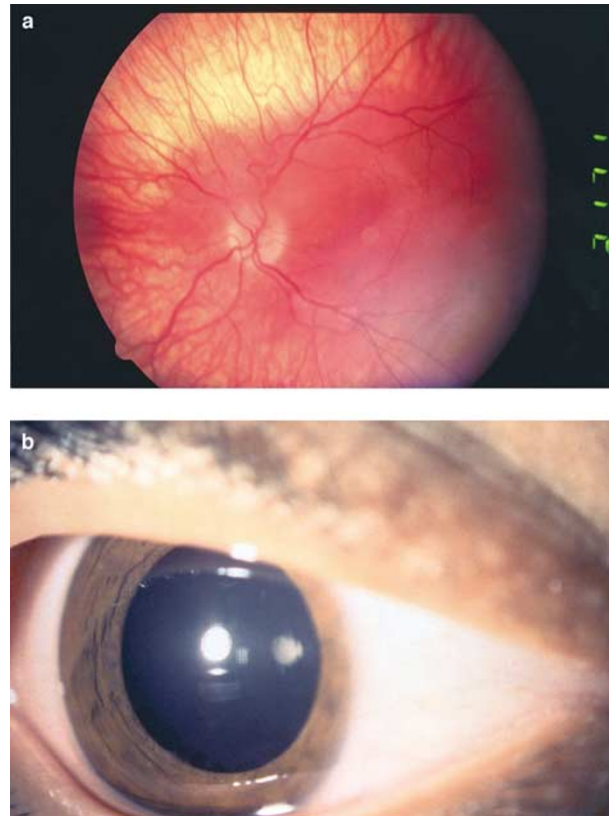


Figure 2 (a) Bilateral hypoplastic maculae. (b) Right posterior pole cataract.

with 10 small pale macules across her trunk and thighs, each 2–3 mm in diameter. She also had two larger pale patches on her buttocks, of 3–4 cm diameter.

The chest wall nodule was biopsied and found to be a plexiform neurofibroma, which led to neuro-axial MRI scans being performed. Bilateral acoustic neuromas and multiple spinal cord tumours were found, thus establishing the diagnosis of neurofibromatosis type 2 (NF2). Mutation screening for NF2 was performed and a mutation affecting the conserved splice donor site of intron 4 of the NF2 (schwannomin) gene, 447+2T>C, was found. Although this is not a classic NF2 mutation, it was felt to account fully for the phenotype. The conjunction of clinical NF2 and ocular albinism raised the possibility of a chromosomal abnormality; the karyotype was normal however.

She is now 14 years old with stable visual acuities of 6/60 right eye, 6/36 left eye and persistent manifest horizontal jerk nystagmus. There has been no progression of the cataract. Her parents and siblings have been examined fully in the Ophthalmology Department and none of them have been found to show any signs of either NF2 or ocular albinism. Family interview revealed no history whatsoever of premature loss of hearing or

any other neurological manifestations of NF2, nystagmus or poor vision.

Comment

The coincidence of autosomal recessive ocular albinism and NF2 in the same individual has not, to our knowledge, been described before. The clinical diagnosis of ocular albinism was made on the basis of the iris pallor and transillumination, nystagmus, hypopigmented fundus and hypoplastic maculae. Although autosomal recessive ocular albinism is rare, the consanguineous parentage would support this diagnosis.

Either parent in generation 1 could be carrying the recessive allele for ocular albinism (Figure 1). The probability of a recessive allele being expressed in subsequent children of this second-cousin marriage would be $0.5 \times 0.5 \times 0.5 \times 0.5 \times 0.5 \times 0.5 = 1/128$. The same would apply for any recessive allele from either parent in generation 1.

The NF2 is most likely a new mutation as 95% penetrance for bilateral vestibular schwannomas would be expected, and this would have almost certainly appeared phenotypically in the family history.¹ Of the NF2 cases, 50% present with no antecedent family history, so it is not unusual to find this as a new mutation. Genetic counselling was therefore reassuring, as the risk of either conditions appearing in subsequent children is very low.

The first clinical manifestation of NF2 in our patient was the posterior subcapsular cataract, a finding in around 80% of cases of NF2.² The subcutaneous plexiform neurofibroma found in this case is not typical of NF2, and does not carry the risk of malignant degeneration expected in similar lesions of NF1.³

Ocular abnormalities are often the first sign of NF2,⁴ the coincidence of the cataract and the ocular albinism

perhaps leading to less attention being paid to the cataract than might otherwise have been the case.

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