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Case Report

A Case of oculocutaneous albinism in a Maltese

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A 4-month-old female maltese dog was admitted to Veterinary Medical Teaching Hospital of Seoul National University for evaluation of abnormal color of bilateral irises. This patient had the photophobia in the light and exhibited the complete absence of pigment resulting in white hair, pink muzzle, eyelids and foot-pads. Central zone of the irises were yellow in color influenced by tapetal reflex, and peripheral zone were pale blue. The iridal capillaries were transparented on the irises. Ophthalmoscopic examination revealed a yellow tapetal fundus but no pigment in the nontapetal fundus.

Key words: oculocutaneous albinism, Maltese

Abinisms have been described in human and animal, and are classified into three types which are oculocutaneous albinism (OCA), ocular albinism (OA) and cutaneous albinism (CA) [3]. OCA includes tyrosinase-related OCA (OCA1), P gene-related OCA (OCA2) and TRP1 gene-related OCA (OCA3). OCA1 is divided into tyrosinase-negative type (OCA1A) and tyrosinase-positive type (OCA1B). Individuals with OCA1A (classic OCA) have absolutely no pigment in their skin, hair and eyes. They have white hair, milky white skin and translucent irises [4]. Partial OA was reported in Beagle [7], Collie, Shetland Sheep dog and Grate Dane [8]. Partial albinism in the dog is often associated with multiple ocular anomalies as well as with unilateral or bilateral inner ear defects resulting in deafness [5].

Classic albinisms (tyrosinase-negative albinism) were described in human [4], cattle [6] and sheep [1]. However the classic OCA has not been reported in the dog. In this report, we describe typical classic OCA in a Maltese. A four-month old female Maltese was referred to Veterinary Medical Teaching Hospital of Seoul National University because of abnormal color of both irises. On physical examination of the dog exhibited the complete absence of pigment resulting in white hair, pink skin, muzzle, eyelids, oral mucosa and foot-pads. Responses to hearing tests were normal. Laboratory abnormalities were not found in complete blood count and serum chemistry evaluation.

On ophthalmic examination of the dog, there was no clinical signs associated with visual impairment but photophobia under bright illumination. Nystagmus and strabismus were not found. Direct and consensual pupillary light reflexes were normal. Bilateral eyelids appeared pink-color and free borders of third eyelids had no pigment. Slit-lamp biomicroscopy assessment revealed no abnormal findings in cornea, anterior chamber, lens and vitreous in both eyes. Iridal capillaries were seen through the transparent irises. Central zone of the irises were yellow in color under the influence of tapetal reflex, and peripheral zone were pale blue.

Prior to the ophthalmoscopic examination, 1% tropicamide solution (Ocutropic R; Samil Pharm, Korea) was applied to dilate the pupils. Ophthalmoscopically, bilateral eyes showed

Fig. 1. External features on physical examination in a two-year old Maltese with oculocutaneous albinism. Complete absence of pigment resulting in white hair, pink muzzle, eyelids (a), oral mucosa (b), skin (c), and foot-pads (d) was shown.

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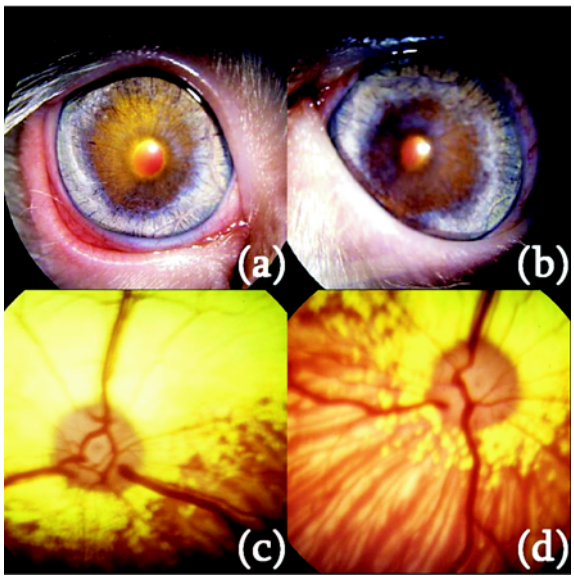

Fig. 2. Oculocutaneous albinism in a two-year old Maltese. Transparent iris (a & b), yellow tapetum (c) and tigroid non tapetum (d) were shown. (a) right eye; (b) left eye; (c) right fundus; (d) left fundus.

tigroid non-tapetal fundus without pigment. Retinal vessels are superimposed on underlying choroidal blood vessels. Normal yellow tapetum was observed in each fundus and optic disc showed no noticeable changes. The lesions of irises and fundi were recorded using the fundus camera (GENESIS ; Kowa , Japan).

After twenty months, physical and ophthalmic examinations were performed again. The dog exhibited no pigment in oculocutaneous and there is no remarkable changes compared to the first examination (Fig. 1 & 2).

OCA is a phenotype for a group of autosomal recessive genetic disorders of melanin synthesis resulting from mutations in several different genes. OCA 1 results from mutations of tyrosinase gene and presents absence of melanin pigment after birth for the life-long (OCA1A, classic OCA) or with the development of minimal-to-moderate amounts of cutaneous and ocular pigment (OCA1B, partial OCA). Other types of OCA have variable amounts of cutaneous and ocular pigment [4]. Individuals with OCA may present a skin burn, nystagmus, photophobia and strabismus in human [2].

This Maltese was manifested no pigment in skin, hair, muzzle, oral mucosa, foot-pads, irises and fundi after birth until 2 years. The dog did not present nystagmus and strabismus but showed photophobia out of doors.

Based on the clinical and ophthalmic findings, this dog was diagnosed as OCA. We need more studies to classify of OCA1, OCA2 and OCA3 related genetic defects.

Microphthalmia, scleral staphylomas, choroidal colobomas, choroidal hypoplasia, retinal dysplasia, retinal detatchments, retinal fibrosis, cataracts, lenticular colobomas, pupillary membranes, iridal colobomas, dyscorias, corectopia, goniodysgenesis and corneal epithelial dysplasia have been described in dogs with partial albinism [5]. Some partial albino dogs appeared cochlear-saccular degeneration resulting deafness [5]. However, these ocular and ear abnormalities were not found except the albinism and photophobia in this case.

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