IV. 5. Detection of Visual Pathway Abnormalities in Albinism Using Positron Emission Tomography and $^{18}$F-FDG


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Introduction

Albinism is a disorder of the melanin pigmentary system and characterized by congenital hypopigmentation of hair, skin, and eyes. Commonly two types of albinism are known. One is autosomal recessive oculocutaneous albinism, in which skin, hair, and eyes are involved. Another is X-linked ocular albinism, in which mainly eyes are involved.\(^1\) Albinism is one of the most important causes of low vision with nystagmus in children. It is sometimes difficult to make a correct diagnosis of albinism clinically. It is known that all albino mammals have anomalous retinogeniculate projection of fibers in the visual perception system.\(^2\)-\(^4\) The majority of the temporal retinal fibers are uncrossed in the optic chiasma and project to the ipsilateral hemisphere in normal primates. Whereas they are dicussate at the chiasm and project to the contralateral hemisphere in albino animals. Based on this abnormally high percentage of chiasmal crossed fibers, usefulness of one eye stimulated visual evoked response was proposed. We have performed preliminary study to show this abnormal chiasmal projection in albinism by Positron Emission Tomography (PET) with $^{18}$F-FDG.

Patients and Methods

Our study included 3 patients of albinism. One was ocular albinism, two were oculocutaneous albinism. Each patient, whose one eye was closed with a patch, was positioned on the bed of the PET instrument. The examination room was kept lighted visual and the patients were ordered to actively look around themselves. Because scenery in the room is a much stronger visual stimulation to activate visual cortex rather than checker-board or white light stimulation.\(^5\) Twenty minutes after the visual stimulation, 3 to 5 mCi of $^{18}$F-FDG was injected intravenously. We started the collection of emission data 30 minutes after the injection. Data of 14 or 21 planes for 600 seconds of data acquisition time was obtained. Regions of interest (ROI) were set on the metabolic images, generated by blood input curve
and emission data.6)-8) Surface view of the cortex were generated by data reconstruction. Two normal volunteer were also examined in the same way.

Result

First patient was a 15-year-old boy with Nittleship type ocular albinism. He had his right eye closed with an eye patch, opened left eye. CMRGlC of the occipital cortex was asymmetric in this case. (Fig. 1, 2) The CMRGlC of right posterior medial occipital cortex projected from the opened left eye was 26% higher than that of the contralateral region.

Second patient was a 64-year-old oculocutaneous albinism woman. She also revealed asymmetric CMRGlC pattern of the occipital cortex (Fig. 3). She opened only her left eye. Relative value of her right posterior medial occipital cortex was 30% higher than that of the left occipital region. In both patients, all other structures, including lateral occipital cortex, did not show the asymmetric CMRGlC.

Third patient was a 39-year-old oculocutaneous albinism man. His visual acuities were less than 4/200 in each eyes due to retinal detachment and secondary glaucoma. The asymmetric glucose metabolic pattern in occipital cortex wasn't detected in this case(Fig. 4).

Two normal volunteers did not show asymmetric pattern in occipital cortex under monocular stimulation.

Discussion

In normal humans, optic nerve fibers from the temporal retina remain uncrossed in the chiasm to project to the ipsilateral hemisphere. The ratio of crossed to uncrossed fibers is about 53:47 in normal individuals9). In albinism, both oculocutaneous and ocular type, 20 % or more of the fibers from the temporal retina cross the chiasm and project to the contralateral hemisphere. Visual evoked potential (VEP) with monocular stimulation are proposed for use in clinical detection of the visual pathway abnormalities in albinism10)-12). Contralateral emphasis to the stimulated eye are shown in albinism. There has been no other method for detection of this asymmetric responses to the monocular stimulation. PET with 18F-FDG is known to show occipital cortex metabolic activation according to the distribution of optic nerve projection13). This study is the first trial to demonstrate the asymmetric regional activity of visual cortex in albino people. It is realized by PET and 18F-FDG for albinism to have the asymmetric glucose use under monocular stimulation.

Albinism is one of the most important causes of low vision with nystagmus in children. It is characterized by hypoplasia of the fovea, hypopigmentation of the fundus, iris translucency, nystagmus, photophobia and decrease of visual acuity1). Diagnosis of albinism is sometimes difficult, since all these clinical findings may not be present. Especially in mild cases, it is very difficult to be certain whether albinism or other syndromes such as congenital cone dysfunction. So this PET diagnosis should be useful for diagnosis of albinism with detection of this visual pathway abnormality. Cerebral glucose metabolism
study by PET with one eye stimulation gave quantitative informations for visual projection. More patient study is needed to establish this diagnostic technique.

References

2) Guiller R. W. et al., Brain Res 96 (1975) 373.

Fig. 1. Case 1. Cross sections 40 and 47mm above the orbitomeatal line. Cerebral metabolic rate for glucose under left eye stimulation.
Fig. 2. Relative CMRGlc, for 15y., male, ocular type.

AMOC: anterior medial occipital cortex,
PMOC: posterior medial occipital cortex,
LOC: lateral occipital cortex,
FC: frontal cortex,
LN: lentiform nucleus,
TH: thalamus.
Fig. 3. Relative CMRGlc, for 64y., female, oculocutaneous type.

Fig. 4. Relative CMRGlc, for 39y., male, oculocutaneous type with retinal detachment and secondary glaucoma.