Visual Cortex Defects in Albinos

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Abstract

All researchers indicate disparities in the organization of the visual system between albinos and pigmented mammals, but nowadays little is known about how albinism distress visual system properties in a whole and visual cortex ones in particular. We have not had efficient proposal of how these exacting findings affect the direction selectivity and receptive fields in visual cortex and how the cortical representation of the visual field is established in albinos, yet. Moreover, there are conflicting reports in different research papers. This brief review highlights unsolved questions associated with albinism and possible perspectives for albino visual cortex research.

INTRODUCTION

Albinism has a profound effect on visual development and visual function. All mammals with hypopigmentation of the retinal pigment epithelium have abnormal visual systems. Albino mammals have been found to have: (1) reduced numbers of uncrossed optic fibres projecting to all visual centres, (2) disorganization of the pattern (lamination) of the dorsal lateral geniculate nuclei, and (3) disorganization of projections from the dorsal lateral geniculate nuclei to the visual cortex (Creel et. al, 1990). It causes reduction and anomalous binocular representations at top levels of the visual system and, as a result, an abnormal retinotopic map in visual cortex (Guillery, 1986; Akerman et al., 2003). Direction selectivity is also broadly disturbed in the motion sensitive cortical areas of albino mammals (Hoffmann et al., 2002). The alterations in retinal projections in albino mammals suppressed severely the visuotopic organisation of cortex. Three cortical projection patterns are associated with albinism: “true-albino”, “Midwestern” and “Boston”. In humans was only found the “true-albino” one, the reasons and consequences of it are listed below.

MAMMALIAN STUDY

In animals investigations of albino visual cortex were mostly performed on mammals. There are relatively few research papers on the rest classes of animals and they are unsystematic, hence, and I will continue with mammals. Albino mutation in mammals has disaster effects on the retina and the visual pathways (Jeffery, 1997). The alterations in retinal projections in albino mammals suppressed severely the visuotopic organisation of cortex. There are three cortical projection patterns, associated with albinism: “true-albino”, “Midwestern” and “Boston”.

(1) Even if the rerouting at the chiasm does not affect the geniculocortical projection, the visuotopic map in albinos will be extremely abnormal: contralateral and ipsilateral hemifields will have a strong representation in the same part of visual cortex. This projection pattern is regarded as “true-albino”. It has been found in tyrosinase-negative albino cats (Leventhal and Creel, 1985; Schmolesky et al., 2000), albino monkey (Guillery et al., 1984), and albino people (Hoffmann et al., 2003).

(2) Representation of the ipsilateral hemifield may be relatively weaker in cortex than in dorsal lateral geniculate nucleus (dLGN); it indicates some cortical suppression. If the geniculocortical projection is anatomically normal, but there is no evidence for any functional input from the misrouted contralateral temporal retina, (cortical suppression of the abnormal input is complete) then it is a case of “Midwestern” pattern. It has been found in most of the Siamese cats studied by Kaas and Guillery (1973) and in albino cats studied by Schmolesky et al. (2000), they conclude that the suppression of abnormal dLGN cell inputs, abnormal cortical cell activity, or both, may underlay reduced numbers of visually driven abnormal cells in cortex relative to the dLGN. Some human albinos also show reduced sensitivity to stimuli obtained by the temporal hemiretina, but not the nasal hemiretina (St John and Timney, 1981). As a consequence each hemisphere responds

rather to stimuli delivered to one visual field and not the another, similar to cortical function in normal animals. So, "Midwestern" pattern looks like the mapping in normal pigmented mammals. This resemblance occurs because the input from the ipsilateral field is physiologically suppressed in cortex.

(3) If there is clear functional input from both temporal and nasal contralateral retina in striate cortex, then it is "Boston" pattern. Geniculocortical projections are reorganized to generate a permanent and consistent mapping of the visual field into the ipsilateral hemifield and through the vertical meridian into the contralateral hemifield. The representation of the vertical midline is no longer at the 17/18 border but is displaced well inside area 17(Akerman et. all, 2003). It has been found in most of the Siamese cats recorded by Hubel and Wiesel (1971) and albino ferrets recorded by Akerman et. all (2003). To form the "Boston" mapping, there must be an additional alteration in the targeting of geniculate axons (Akerman et. all 2003).

Although the presence of these diverse cortical projection patterns in the albino visual cortex is well-known, the factors underlying their genesis is still unclear. The "Boston" pattern appearing, probably, if the representation of the vertical meridian is displaced by only 1–2 mm (Akerman et. all 2003). It was seen in "Boston" Siamese cats (Hubel and Wiesel, 1971; Kaas and Guillery, 1973; Shatz, 1977; Cooper and Blasdel, 1980) and "Boston" albino ferrets (Akerman et. all 2003). These data may correspond to highest possibility of cortex reorganization. One can guess that the "Boston" pattern shall never take place in albino cats, primates or humans, which have a much higher cortical magnification factors.

So, one can conclude that in cases of mammalian albinism, a possible sensory mismatch due to abnormal projection of a part of temporal retina to the visual cortex is avoided either by reorganisation of the thalamocortical connections to give the abnormal input an restricted cortical representation, or by the abnormal input being substantially suppressed.

HUMAN STUDY

In normal people decussation of the optic fibres is loyal to the vertical meridian that passes through the fovea: fibres from the temporal retina project ipsilaterally to the lateral geniculate nucleus and cortex, whereas fibres from the nasal retina project contralaterally. In albinos, however, a higher than normal proportion of fibres from the temporal retina project contralaterally. The line of decussation is, therefore, shifted to the peripheral visual field. Finally line of decussation in the human was detailed as a vertical meridian that is shifted into temporal retina (Hoffmann et.all, 2003). This abnormal projection of a part of the temporal retina to the visual cortex, together with the normally routed fibres from nasal retina, provides a cortical hemisphere with visual input from more than the normal hemifield of visual space. Due to predominantly monocular innervations of the left and right hemispheres, the unilateral activation of the visual cortex (lateralisation) can be detected with visual evoked potentials. It was studied with multichannel pattern onset visual evoked potentials that pathological crossing of the nerve fibres in the optic chiasm is always detectable in oculocutaneous and ocular albinism, but had never been detected in normal humans (Torok, 2001; Kasmann-Kellner et. all; 2003). Thus, lateralisation of visual evoked potentials appeared to be more aware indicator of ocular albinism than other symptoms as macular hypoplasia, hypopigmentation, iris transillumination, nystagmus, reduced visual acuity and so on. But due to the restricted spatial resolution (Morland et. all, 2002), visual evoked potentials analyses cannot make a payment to specific questions of albino cortical organisation such as the representation of the visual field in the early visual areas and the degree of cortical magnification.

Human albinos show congenital nystagmus without perceiving oscillopsia. Consequences of atypical chiasmatic crossing was demonstrated with monocular visual stimulation using functional magnetic resonance imaging (fMRI) (Schmitz et. all, 2004). They observed contralaterally dominated activation of visual cortices correlating to clinical albinism parameters. This confirms albinism as a continuous range of hypopigmentation disorders. Additionally, albinos showed activation of the superior culliculus and of visual motion areas although the stimulus was stationary. One can conclude, therefore, that activations of superior colliculi in albinos are suggestive of their involvement in congenital nystagmus.

Albino misrouting (Apkarian, 1992; Kriss et. all, 1992; Morland et. all, 2002; Schmitz et. all, 2004) is shown clearly during an independent visual stimulation of both hemifields: activity in the occipital cortex in the normal humans is contralateral to the stimulated visual field, whereas it is contralateral to the stimulated eye in the albino, independent of the stimulated visual field (Morland et. all, 2002). Thus, the albino visual cortex is activated not only by stimulation in the contralateral visual field, but also by abnormal input

from the ipsilateral visual field.

There are conflicting reports on cortical activity ipsilateral to the stimulated eye. Morland et. all, 2002, stated that fMRI data directly showed that in the albino it is indeed the occipital lobe contralateral to the stimulated eye that is active during stimulation in each hemifield, whereas the control subject hemispheres were only active during stimulation in the contralateral visual field. Activity ipsilateral to the stimulated eye was strongly reduced in the albino. Hedera et all., documented some cortical activity ipsilateral to the stimulated eye: substantial residual ipsilateral activity in the more anterior portion of the fundus of the calcarine sulcus. The difference between their study may originate from the size of the visual stimulus.

Input from temporal retina is not substantially suppressed in humans and forms a retinotopic mapping that is superimposed on the mapping of the nasal retina in striate and extrastriate areas. The abnormal routing of temporal fibres is not total, with the line of decussation shifting to between 6 and 14° into temporal retina (Hoffmann et.all, 2003). Input to visual cortex in human albinism does not undergo topographic reorganization between the thalamus and cortex and, moreover, is not significantly suppressed in either striate or extrastriate areas. Nasal and temporal retina have their visuotopic representations superimposed in early extrastriate areas. Thus, one may conclude that cells in striate and extrastriate areas that normally respond to binocular input would not develop to act in response to input from mirror-symmetric retinal locations. Topographic mapping in humans does not match to the normally observed patterns in other mammals but takes the form of the “true albino” pattern. This mapping is consistent with that documented in the only other primate studied (Guillery et al., 1984). On one hand it would appear, therefore, that developmental mechanisms that allow the thalamocortical projections to be reorganized in some mammals are not available to the primate. On another hand, abnormal projection of the temporal retina was observed predominantly in the central part of the visual field (Hoffmann et.all, 2003). Hence, the fundamental origin of the albino abnormality appears to be expressed in the same way in primates as in other species.

Perspectives for albino visual cortex research

- Although the presence of diverse cortical projection patterns in the albino visual cortex is well known, the factors underlying their genesis is still unclear.
- Future studies of the albino visual pathways needed to clarify mechanisms used by the central nervous system to protect perceptual function.
- How the topographic cortical representation of the visual field is established as there are conflicting reports on the pattern of the cortical representation in different species.
- Whether cells in striate or extrastriate areas that respond to nasal and temporal retina are segregated into zones (or columns) in humans as they are in the striate cortex of other albino animals.
- The line of decussation in human albinos is shifted to the peripheral visual field. The impact of this abnormal visual projection on the cortical mapping of the visual field in human albinism has to be determined.

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References

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