The ROYAL COLLEGE of OPHTHALMOLOGISTS

COMMENT

Comment on: 'What colour are your eyes? Teaching the genetics of eye colour & colour vision. Edridge Green Lecture RCOphth Annual Congress Glasgow May 2019'

Yossy Machluf[1]✉, Yoram Chaiter[2], Gilad Allon[3] and Eedy Mezer[4,5]

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We read with great interest and pleasure Mackey's article [1] concerning the vast opportunities of leveraging broad genetic and epigenetic concepts, which are essential to genetically literate ophthalmologists, by teaching the subjects of eye colour and colour perception.

Additionally, the author demonstrated the gene-environment interaction in colour blindness mainly by twin study on the #TheDress photograph [2]. A recent retrospective study of Israeli Jewish adolescents further supports this notion and illuminates the intricate relationship between genetics and environmental factors affecting colour blindness [3]. The unique composition of the population enabled to investigate the association of congenital colour blindness prevalence with both ethnicity (place of origin, representing the genetic component) and place of birth (representing environmental factors and habits), and the interplay between them. It provided three main lines of evidence: (I) The prevalence of congenital colour blindness differed among subpopulations of different ethnic background and among subpopulations of different place of birth; (II) differences in the prevalence of congenital colour blindness were obtained among subjects of the same origin, who were born in Israel compared to those who were born elsewhere; and (III) both variables - place of origin ($p < 0.01$) and place of birth ($p < 0.05$) were associated with the prevalence of congenital colour blindness in a multivariable regression model. Thus, congenital colour blindness may be associated with both genetics and environmental factors.

Colour blindness pertains to the cone photoreceptors in the retina. Preservation of cone photoreceptors was shown to be affected by the environment [4]. Epigenetic mechanisms may underly environmental contribution to the pathogenesis of colour blindness, in a similar manner to other congenital anomalies [5]. We anticipate and hope that this evidence may pave the way to future research exploring if and to what extent epigenetic mechanisms are implicated in the aetiology of colour blindness. This may provide novel insights into the consequences of environmental driven in utero modifications of the human genome regulatory architecture on the pathogenesis of colour blindness, which is considered as primarily genetic in origin. This in turn, should be integrated into the genetic education programme of every ophthalmologist, as proposed by Professor David A. Mackey.

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AUTHOR CONTRIBUTIONS

EM, YM and YC conceived the idea of this correspondence. All authors carefully and critically read the original articles by Mackey [1], and collaboratively designed the outline of this article. YM wrote the paper with inputs from all the co-authors.

COMPETING INTERESTS

The authors declare no competing interests.

ADDITIONAL INFORMATION

Correspondence and requests for materials should be addressed to Yossy Machluf.

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[1]Unit of Agrigenomics, Shamir Research Institute, Haifa University, Haifa, Israel. [2]The Israeli Center for Emerging Technologies in Hospitals and Hospital-based Health Technology Assessment, Shamir (Assaf Harofeh) Medical Center, Be'er Ya'akov, Israel. [3]Retina service, Moorfields Eye Hospital, London, UK. [4]Department of Ophthalmology, Bruce and Ruth Rappaport Faculty of Medicine, Technion - Israel Institute of Technology, Haifa, Israel. [5]Department of Ophthalmology, Ruth Rappaport Children's Hospital, Rambam Health Care Campus, Haifa, Israel. ✉email: Yossy.machluf@gmail.com

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