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Prevalence of eye colobomas in wild birds: insights from a large-scale citizen science study

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Abstract. Colobomas are inherited defects affecting the membranes of the eye, leading to irregularities in the shape of the iris and pupil. While the causes and occurrence of colobomas have been studied extensively in humans and captive or domesticated animals, their prevalence in wild birds (which may represent interesting

model organisms) remains largely unexplored. In this study, we developed a citizen science project which conducted fieldwork to assess the presence of eye colobomas in relation to species and sex, examining 58,651 individuals from 179 wild bird species across Europe. We identified 17 cases of colobomas in nine bird species during the screening. Although the overall prevalence was low (2.90 cases per 10,000 individuals), a notably higher prevalence was observed in several owl species (Strigidae), with rates exceeding 100 cases per 10,000 individuals. Among six sexed Eurasian scops owls *Otus scops* with colobomas, all were females, while in little owls *Athene noctua*, both positive cases were males. Our findings suggest that certain wild bird species could serve as valuable subjects for studying colobomas, though further research involving more extensive sampling and visual inspection, along with cytological and genetic analyses, is necessary to estimate coloboma prevalence across birds accurately.

Key word: birds, community science, Europe, eye defects

Introduction

Colobomas are congenital and sex-linked malformations of the eye's membranes, causing aberrations in the shape of the iris, pupil, lens, retina, or another part of the eye (Abbott et al. 1970, Gregory-Evans et al. 2004, Chan et al. 2021). Colobomas typically arise from abnormal eye development during prenatal development and occur when the optic fissure fails to close (Gregory-Evans et al. 2004, Chan et al. 2021). Failure of optic fissure closure is linked to numerous internal factors, including genetic and chromosomal aberrations as well as external environmental stressors, including toxin exposition, vitamin deficiency and parasitic and viral infections (Gregory-Evans et al. 2004, Verma & FitzPatrick 2007); the effect of factors causing eye coloboma have been explored mainly in humans and captive/domesticated animals (Wilcox 1958, Gregory-Evans et al. 2004, Chan et al. 2021). Coloboma is often associated with microphthalmia, an eye condition where one or both eyes are abnormally small (Gregory-Evans et al. 2004, Verma & FitzPatrick 2007, Aldahmesh et al. 2012). Colobomas may occur in one or both eyes and, depending on their position and size, can be associated with significant visual impairment or even blindness of the affected individual (Chan et al. 2021).

Colobomas are relatively rare malformations in humans, occurring in only one or two per 10,000 newborn babies (Stoll et al. 1997, Morrison et al. 2002). In captive animals such as birds, this defect may be more prevalent and possibly sex-specific (Wilcox 1958, Cardona & Plumer 2004). In contrast, this malformation is rarely described in wild-living birds; Lord (1956) reported two adult individuals of rough-legged hawk *Buteo lagopus* and red-tailed hawk *B. jamaicensis* exhibiting unilateral colobomas which involved a lack of development or atrophy of approximately three-fourths of the iris, lens, retina,

and choroid. It seems that heterogametic sex (i.e. ZW females in birds and XY males in mammals) is more severely affected by colobomas. For example, colobomas were present in 26% of females but only 9% of males in a single flock of rosecomb bantam chickens (Cardona & Plumer 2004). Moreover, the presence of coloboma may cause more serious craniofacial malformations in the skull, beak, and throat in more sensitive sex (Abbott et al. 1970, Robb et al. 2013). Wild birds, as a diverse vertebrate clade, may potentially represent interesting model organisms for the study of eye malformations. In both wild and captive birds, the occurrence of coloboma is rare; hence, large sample sizes are needed to detect it. Accordingly, collaborative citizen science (in broad interpretation, this includes not only general public and amateur experts but also scientists trained in other fields; Shirk et al. 2012) can be a valuable tool to provide large data sets across broad geographic areas, particularly in the case of relatively rare natural phenomena (Sullivan et al. 2014, Mikula et al. 2016, Pesendorfer et al. 2018).

In this study, through an established regional collaborative research platform for monitoring wild birds, we developed a citizen project utilising trained volunteers across Europe to conduct cross-national and cross-species field surveys to document the occurrence of coloboma among wild bird species. We used only data from systematic field surveys, which simultaneously documented both positive and negative cases, enabling us to estimate the prevalence of this malformation in sampled species.

Material and Methods

Data collection procedure

In February 2023, we emailed a brief description of our project, data requirements and methods to 48 EURING member ringing schemes. In brief, we asked potential

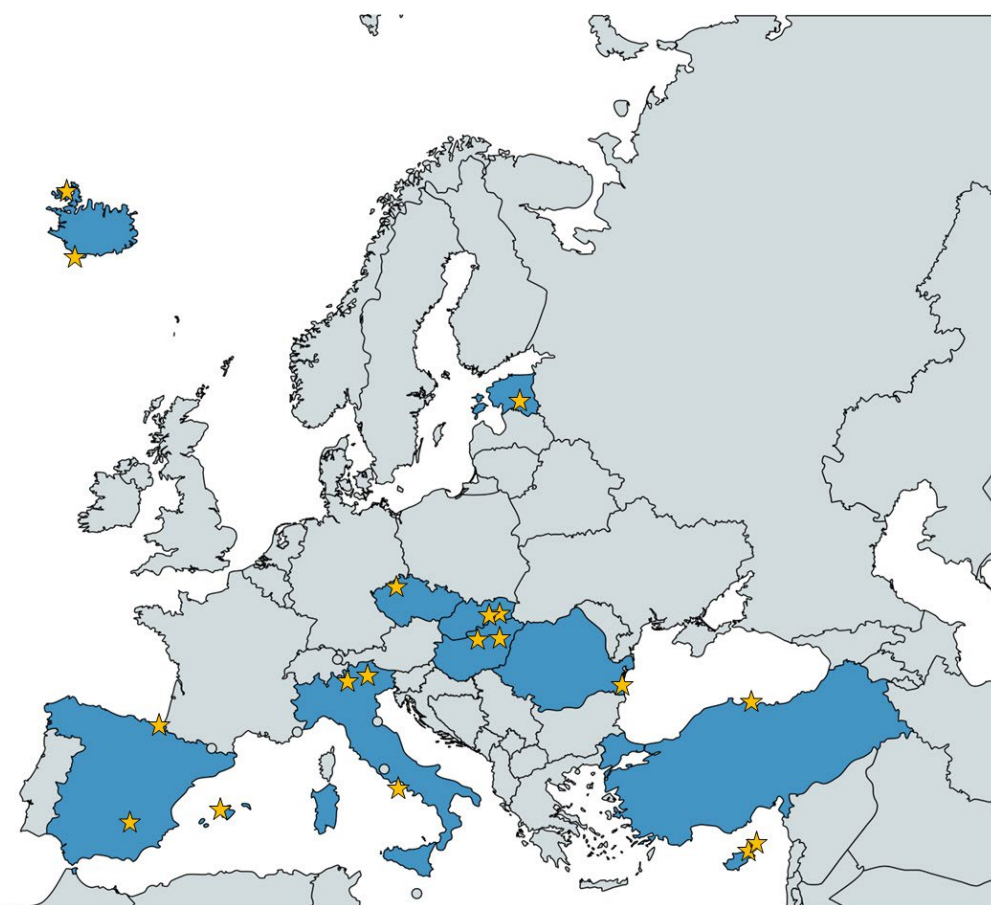


Fig. 1. Distribution of our sampling localities in Europe. Yellow stars depict sampling points, and blue indicates sampled countries. For details on the exact position of localities, see the Material and Methods section. Note that not all points are visible – in two cases (Cyprus and Slovakia), we merged closely positioned points into a single star to improve map clearness.

collaborators to provide any previously collected data and/or to collect new data for the project during the period February 2023 and September 2024. In the latter case, we asked to check only mist-netted birds and note down both positive and negative cases of sampled birds, accompanied by geographic sampling area species names and coordinates, date and any additional details of sampled birds, such as sex. Of the 48 contacted centres, 13 forwarded our request to their members. We also asked two collaborators with whom our research group has previous personal connections to contribute their data. Our sampling areas cover ringing points in Cyprus (Dipkarpaz 35°36'21'' N, 34°22'55'' E; Karpaz 35°39'34'' N, 34°33'14'' E; Golden Beach marsh 35°38'35'' N, 34°32'14'' E), Czechia (Northern Bohemia 50°23' N, 13°40' E; for more details see Šálek et al. 2016), Estonia (Vaibla 58°24'15'' N, 26°04'30'' E), Hungary (Budapest 47°31' N, 19°05' E; eastern Hungary 47°33' N, 20°54' E; Šálek et al. 2016), Iceland (Westfjords 66°03' N, 22°59' W; Westman Islands 63°26' N, 20°18' W), Italy (Bocca di Caset 45°51'34'' N, 10°41'25'' E; Passo del Brocon 46°06'08'' N, 11°41'50'' E; <https://progettoalpi.muse.it/>; Ventotene Island 40°47' N, 13°25' E),

Romania (Agigea 44°05'11'' N, 28°38'28'' E), Spain (Mallorca 39°35'18'' N, 2°19'46'' E; Albacete 38°46'48'' N, 2°22'40'' W; Hondarribia 43°22'21'' N, 1°48'36'' W), Slovakia (Košice – Horný Bankov 48°44'44'' N, 21°12'55'' E; Rozhanovce 48°46'21'' N, 21°22'18'' E; Drienovec Bird Ringing Station 48°36'59'' N, 20°54'55'' E) and Turkey (Cerneik Bird Ringing Station 41°38'36'' N, 36°05'02'' E) (Fig. 1 and Table S1). Some of the data used in this study ($n = 3,865$ individuals and six species) were collected prior to the launch of the project (1997-2022). In such cases, we ensured that the data collection adhered to the same methodology as the data gathered specifically for this project.

The eyes of sampled birds were inspected visually for any anomalies. As a positive presence of coloboma, we considered cases where the iris and pupil showed an aberrant shape, typically that one or both protruded to the sclera. However, we acknowledge that visual inspection may be less sensible towards positive cases in bird species with dark iris and sclera, potentially underestimating colobomas in these species. Moreover, research on human eyes indicates that a significant proportion of colobomas affect only

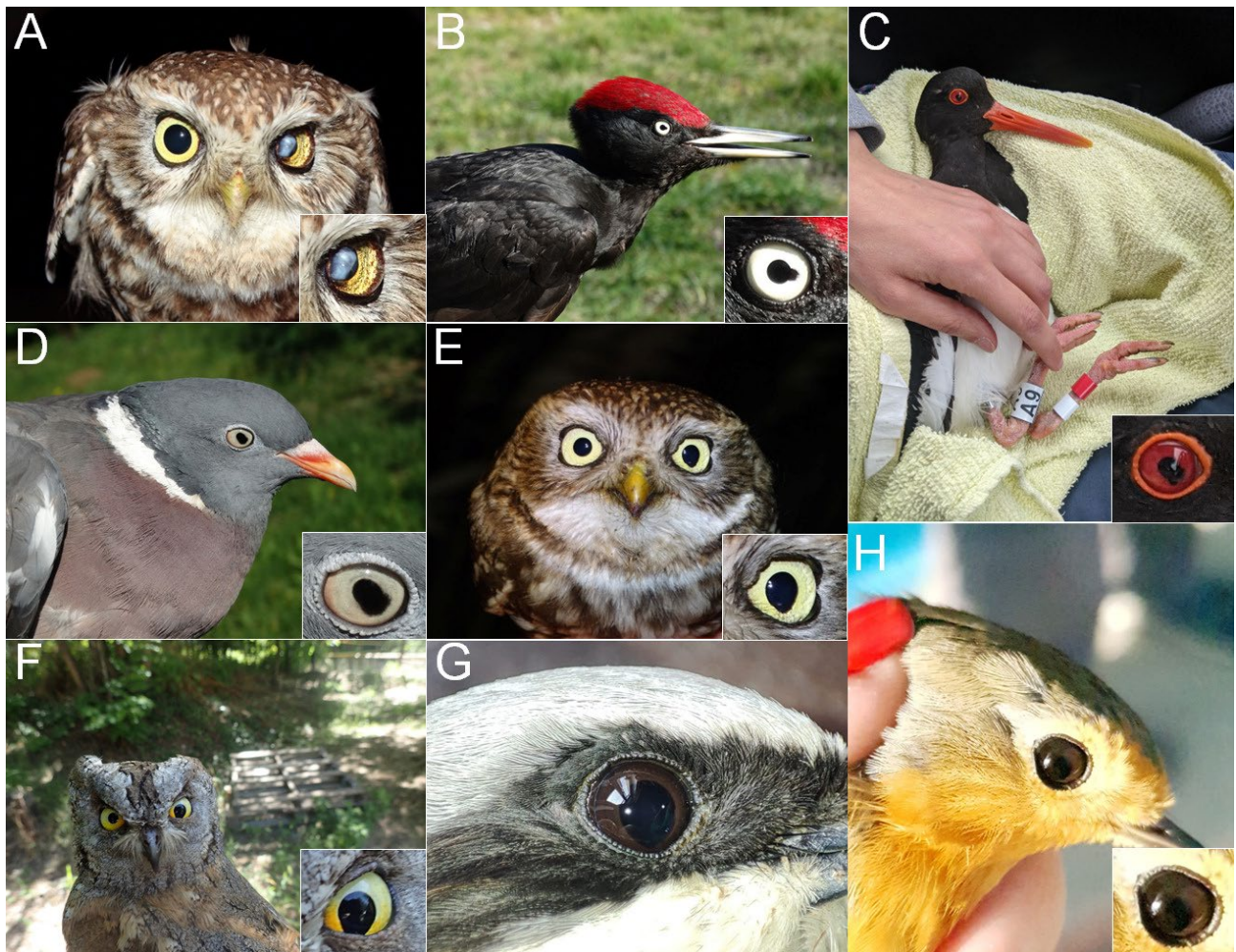


Fig. 2. Examples of (A-D) non-coloboma and presumable coloboma (E-H) eye defects. (A) Pupil defect in the left eye of the little owl *Athene noctua* is presumably affected by mechanical or other non-developmental damage (photo Martin Šálek). (B-D) Examples of three species with common eye flecks which are not related to coloboma, such as black woodpecker *Dryocopus martius* (photo Katalin Odett Lukács), Eurasian oystercatcher *Haematopus ostralegus* (photo Ingrid Bobeková) and common wood pigeon *Columba palumbus* (photo Katalin Odett Lukács), respectively. (E-G) Presumable colobomas in little owl (photo Martin Šálek), Eurasian scops owl *Otus scops* (photo Balázs Juszti), red-backed shrike *Lanius collurio* and European robin *Erithacus rubecula* (both photos Emanuel Baltag). Miniatures in the lower right corner of the picture zoom in the eye in less visible cases.

the posterior eye segment, which cannot be detected through external examination (Nakamura et al. 2011). If this is also the case in birds, our visual sampling method may further reduce the detection rates of colobomas in our sample of wild birds; however, we are not aware of a comprehensive study on this issue across birds. We tried to detect and exclude cases when the iris/pupil structure was affected by non-congenital defects such as mechanical injury or congenital eye defects that can resemble coloboma but are caused by different mechanisms and developmental issues. A coloboma of the iris/pupil can cause an irregular-shaped iris/pupil, which might be mistaken for anisocoria (iris/pupil size difference between the two eyes) if only one eye is affected; hence, we excluded cases when iris/pupil size differed between the two eyes but without significant defect in iris pupil shape.

We also checked for cases of aniridia (i.e. a partial or complete absence of the iris), Peters' anomaly (affects primarily cornea, which is clouded, and there is an abnormal attachment between the cornea and the iris or lens) and sclerocornea (cornea becomes opaque and blends with the sclera) but we did not detect them. We excluded from our analysis species that exhibit a high prevalence of eye structures resembling colobomas but are, in fact, dark eye flecks that have different origins and development than colobomas (see also Guzzetti et al. 2008). These include shorebirds (Charadriiformes), such as the Eurasian oystercatcher (*Haematopus ostralegus*), as well as the black woodpecker (*Dryocopus martius*) and the common wood pigeon (*Columba palumbus*), including these species could have significantly but falsely overestimated the frequency of colobomas.



Fig. 3. Distribution of coloboma across the phylogenetic tree of birds covering 179 species sampled in this study. Orange bars show the presence of coloboma across species; grey bars show sampling effort; the height of the bars is proportional to the number of observations (note that values with grey bars were log-transformed due to unequal sampling effort, range = 1-9,776 observations per species). Pictures show species with a positive discovery of coloboma (clockwise): (1) red-backed shrike *Lanius collurio*, (2) Eurasian wryneck *Jynx torquilla*, (3) Eurasian pygmy owl *Glaucidium passerinum*, (4) little owl *Athene noctua*, (5) Eurasian scops owl *Otus scops*, (6) European robin *Erithacus rubecula*, (7) European pied flycatcher *Ficedula hypoleuca*, (8) spotted flycatcher *Muscicapa striata*, (9) garden warbler *Sylvia borin*. Photos were taken by Jan Grünwald (photos: 1, 3, 6-8), Peter Mikula (2), Martin Šálek (4), Balázs Jusztin (5), and Ľuboš Korytár (9).

Finally, we calculated the prevalence of coloboma in wild birds by dividing the number of positive cases by the total number of sampled birds. We also calculated the binomial proportion confidence interval for each species using the *binom.confint* function in the *binom* v. 1.1-1.1 package (Dorai-Raj 2014). We used the 'exact' method of confidence interval estimation, which is particularly suitable for small sample sizes.

Results

Data were provided from ten countries distributed between 33° in the south, 67° in the north, 36° in the east and 23° in the west (Fig. 1 and Table S1). We identified 17 cases of colobomas (2.9 cases per 10,000 sampled birds; 95% CI (1.7-4.6)) in nine wild bird species during our screening of 58,651 individuals

from 179 species (Figs. 2 and 3). Among species with at least one positive coloboma finding, the prevalence per 10,000 individuals was as follows (in decreasing order): 160 (43.8–404.6) in little owl *Athene noctua* (four cases in 250 individuals), 149 (3.8–803.8) in Eurasian wryneck *Jynx torquilla* (one in 67), 111 (40.9–240.3) in Eurasian scops owl *Otus scops* (six in 540), 83 (2.1–455.6) in Eurasian pygmy owl *Glaucidium passerinum* (one in 120), 17 (0.4–95.0 and 0.4–91.7, respectively) in spotted flycatcher *Muscicapa striata* (one in 584) and European pied flycatcher *Ficedula hypoleuca* (one in 605), 14 (0.3–76.9) in garden warbler *Sylvia borin* (one in 722), nine (0.2–51.4) in red-backed shrike *Lanius collurio* (one in 1,081), and two (0.1–10.4) in European robin *Erithacus rubecula* (one in 5,379). Notably, no colobomas were detected in several well-sampled species, including barn swallow *Hirundo rustica* ($n = 9,776$), common chiffchaff *Phylloscopus collybita* ($n = 4,412$), willow warbler *P. trochilus* ($n = 3,993$), and other bird species (Table S2). In cases where we had information on the sex of coloboma-positive individuals, all six coloboma-positive Eurasian scops owls were females (six individuals sexed in total; all females), while the two cases in little owls were males (173 individuals sexed, 29 females and 144 males); owls were caught and sexed in the field based on a head shape, wing length, body mass, brood patch, and later also using genetics (see also van Nieuwenhuyse et al. 2008, Valterová R. et al., unpublished data).

Discussion

This study is the first of its kind in Europe and globally to document the prevalence of coloboma in wild-living bird species, offering new insights into the occurrence of this condition in avian populations. It is also the largest study in terms of geographical scope, encompassing a wide range of habitats across Europe, samples including approximately 60,000 individual wild-living birds and species diversity covering 179 bird species. It represents a pioneering application of citizen science in the study of coloboma, utilising contributions from bird ringers to enhance data collection and broaden the study's reach. We documented 17 cases of coloboma occurrence in nine wild-living species of European birds. The general prevalence of coloboma among birds was low, occurring in approximately three in 10,000 sampled birds. This rarity is similar to coloboma prevalence in humans (Stoll et al. 1997, Nakamura et al. 2011, George et al. 2020). We detected the highest prevalence of coloboma in some species of owls (Strigidae, where in two species, > 1% individuals were coloboma positive) and woodpeckers (Picidae).

Pourlis (2011) reviewed ocular malformations of birds and stated that such deformities are rarely reported in wild birds, with microphthalmia being the most frequently reported malformation and mentioning only two cases of coloboma originally reported by Lord (1956). Similarly, Kinney et al. (2017) reported only a single case of iris coloboma among 21 zoo-housed American white pelicans *Pelecanus erythrorhynchos*. Compared to other species surveyed, we detected a disproportionate occurrence of coloboma in little owls. The little owl population in Europe has declined significantly since the mid of 20th century, which resulted in the substantial contraction of its distribution range and local extinctions in Central Europe (Šálek & Schropfer 2008, van Nieuwenhuyse et al. 2008, Chrenková et al. 2017). Individuals living in small and isolated populations may be more prone to inbreeding (Pellegrino et al. 2015), potentially resulting in increased congenital diseases such as eye coloboma. In particular, a significant level of inbreeding was detected only in the study area in the Czech Republic, while it was not detected in the Hungarian population (Valterová R. et al., unpublished data), where the species still achieves a high population density (Šálek et al. 2013, 2025). However, since coloboma in little owls was detected in both populations (two cases in Hungary and one in Czechia), this topic warrants further research.

The absence of coloboma detection in most species is most likely related to relatively low sampling in most species (median = 14 observations for species; see also Fig. 3). When taking all species together, coloboma prevalence was generally low. This rarity is in accordance with previous studies showing a low occurrence of coloboma among wild animals. For example, Schmidt & Toft (1981) described an overview of ophthalmic malformations in animals from zoological collections diagnosed via biopsy or necropsy, detecting 24 and 12 such cases in birds and other vertebrates (mammals, non-avian reptiles and fish), respectively, but without a single case of coloboma. Schmidt (1971) necropsied 223 non-human primates and found four cases of eye coloboma in olive baboon *Papio anubis* (two cases), rhesus macaque *Macaca mulatta*, and chimpanzee *Pan troglodytes*. However, we found a high prevalence of coloboma in some owls and woodpeckers. This finding may suggest that the prevalence of coloboma is genuinely high in these species, making these species potentially interesting model species for further study of this phenomenon, as well as its causes and impacts. Reliable baseline estimates of coloboma prevalence among most wild-living birds



would require 1) a more intense sampling effort and 2) visual inspection supported by cytologic and genetic evaluation. For example, the two owl species with the highest rates of coloboma are both nocturnal predators, often using woodland or urban habitats. These species may be particularly prone to injury due to collisions while hunting at night (Seruca et al. 2012, Šálek et al. 2019, 2023). Therefore, genetic techniques could help confirm specific eye conditions according to the field evidence observed. Inspection for coloboma could be integrated into long-term monitoring of national ringing schemes to allow long-term monitoring and is recommended at least for owls and woodpeckers.

Most individuals with coloboma in the eye were females. This finding agrees with a previous study on chickens, showing that coloboma is sex-specific (Abbott et al. 1970, Cardona & Plumer 2004). However, our results on sex-links in coloboma must be taken with caution because of very low sampling.

Several wild bird species, such as some shorebirds (Charadriiformes) and pigeons (Columbiformes), have regularly distinct and dark eye 'flecks', which might be mistaken for coloboma during visual inspection. Interestingly, these eye flecks seem to be sex-linked, as shown in a study on sex-determination using eye flecks in black oystercatchers *Haematopus bachmani* (Guzzetti et al. 2008), species closely related to Eurasian oystercatcher *H. ostralegus* included in this study. Authors were able to correctly sex oystercatcher individuals in 94% of cases based solely on the size of eye flecks, with full eye flecks (which had a coloboma-like appearance, see Fig. 2C here or Fig. 2 in Guzzetti et al. 2008) being much more prevalent in females than in males (Guzzetti et al. 2008).

Citizen science projects such as the current study allow data to be collected on large scales with large sample sizes. However, this data availability may result in reduced data quality due to larger associated observer error, where the field teams are less experienced than the research leads. To optimise future work, the integration of guidance materials/training for regional and national ringing schemes on identifying and reporting eye defects is recommended.

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Ethics: The birds were captured and handled by licenced ornithologists; in Slovakia, under exemption No. 3320/2019-6.3 from Act. No. 543/2002 of the code on nature and landscape protection, granted by the Ministry of Environment of the Slovak Republic (ringing licence SR 5040; E. Korytár); in Hungary, all activities were conducted under the Hungarian national license PE-KTF/97-13/2017, and in compliance with personal

certifications TMF-3/2008, TMF-14/2010, TMF-320/2017, TMF-556/2007, TMF-24/2009 and TMF-514/2004 as well ringing licence numbers 623 and 643 issued by the Ministry of Agriculture and MME Birdlife Hungary (Á. Monoki, K.O. Lukács and B. Juszti); in Italy, all activities have been carried out in compliance of art. 4.1, 4.2 and 7 of national Law 157/92 and under ringing licenses 0601 and 2624 issued by the Italian Institute for Environmental Protection and Research (Istituto Superiore per la Protezione e la Ricerca Ambientale; ISPRA) and under project Progetto Alpi permitted by ISPRA and the Autonomous Province of Trento (Ufficio Foreste e Fauna) (L. Serra and A. Franzoi); in Turkey, bird handling was performed with the permission of the Ministry of Agriculture and Forestry, General Directorate of Nature Conservation and National Parks, according to the protocol number 21264211-288.04-10658562 and under the Ethical Principles in Animal Research approved by the Ondokuz Mayıs University Animal Ethical Committee with the protocol number: 604.01.04-2300045202 and the 2023/17 ethical number (K. Erciyas-Yavuz and N. Yavuz); in Czechia, work was performed under ringing licence 984 issued by the Bird Ringing

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Author Contributions

P. Mikula established the original idea, did formal analyses and visualisations, and wrote the first version of the manuscript. P. Tryjanowski supervised the project. M. Šálek, K. Adojaan, A.D.L. Alonso, E. Baltag, D. Beton, I. Bobeková, R.A.M. Catalán, A. Franzoi, K. Erciyas-Yavuz, E.S. Hansen, L. Korytár, P. Linhart, V. Méndez, Á. Monoki, A.-M. Pintilioaie, L. Serra, R.T.E. Snape and N. Yavuz collected field data. All authors read and approved the final manuscript.

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Supplementary online material

Table S1. List of sampling localities, sampling periods, number of sampled species and individuals, and data collectors.

Table S2. The number of positive coloboma cases in sampled birds. We also mention the number of negative cases, birds with distinct eye flecks, the total number of sampled individuals, the prevalence of coloboma, and its 95% confidence intervals in sampled bird species.

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