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Review article

Inherited retinal disease in global Indigenous populations: A scoping review

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ABSTRACT

Accurate diagnosis is essential for accessing emerging gene-targeted treatments for inherited retinal diseases (IRDs), but many minoritised communities face additional barriers to diagnosis. This scoping review synthesised clinical studies on the prevalence and diagnosis of IRDs among Indigenous Peoples worldwide. Medline, Embase, Global Health, Informit and CINAHL were searched on December 4, 2023. We included articles reporting Indigenous Peoples with IRDs from all global regions published between 1974 and 2023; 73 studies (581 cases) of IRDs in Indigenous Peoples from 24 countries were included, mostly reporting participants indigenous to the Middle East (34 %), Oceania (27 %) and North America (23 %). Studies of specific IRD cases showed geographical or cultural group associations, such as rod-cone dystrophy among the Diné (Navajo Nation) or Bardet-Biedl syndrome in Bedouin populations of the Middle East. With dedicated programs, population-specific IRD gene variants in the Middle Eastern Bedouin populations, New Zealand Māori and other Pacific peoples are the most well-characterised, and this has enabled improved diagnostic approaches. There is limited knowledge of the relative prevalence and support needs for IRDs among most other global Indigenous groups. Engagement, co-designed approaches and collective efforts, including raising awareness, may address challenges limiting equitable access to IRD diagnosis for Indigenous Peoples, facilitating access to emerging treatments.

1. Introduction

Inherited retinal diseases (IRDs) are a group of clinically and genetically heterogeneous diseases. Although individually rare, IRDs collectively affect 1 in 2000 people worldwide and are the leading cause of blindness in working-age adults in many developed countries. [58,82] Onset varies between birth through to late middle age, and most IRDs cause progressive vision loss over years or decades, requiring ongoing vision rehabilitation and support services. Some IRDs are syndromic with systemic features, for example, Bardet-Biedl syndrome (BBS) is

Abbreviations: BBS, Bardet-Biedl syndrome; COFS, cerebro-oculo-facio-skeletal; CORD, cone-rod dystrophy; CSNB, congenital stationary night blindness; FDA, food and drug administration; IQR, interquartile range; IRDs, inherited retinal diseases; IWGIA, international work group for indigenous affairs; LCA, Leber congenital amaurosis; MD, macular dystrophy; NGS, next-generation sequencing; RCD, rod-cone dystrophy; US, United States.

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associated with obesity, polydactyly, kidney disease, and developmental disabilities, and Usher syndrome affects hearing and balance. [131] Because of the heterogeneous clinical presentation, getting a clinical diagnosis can be challenging, resulting in a long diagnostic odyssey for some families. [31]

Genetic testing plays an important role in establishing a definitive IRD diagnosis. To date, over 300 IRD-causing genes have been identified. [N] Having a genetic diagnosis can further assist in family planning and inform eligibility for gene-specific therapies. This is particularly important as in 2017, voretigene neparvovec-rzyl (Luxturna®) became the first Food and Drug Administration (FDA)-approved ocular gene therapy for treating *RPE65*-associated IRDs[118] with many other gene and cell therapies for IRDs under development. [12] As a result, research into these emerging treatments has accelerated, with programs to improve access to genetic testing in many countries; [11] however, access to healthcare and diagnostic rates vary between populations, with many minority groups having lower diagnostic rates attributed to factors like lack of community awareness and limited access to and utilisation of culturally safe healthcare.

There are over 477 million Indigenous Peoples worldwide. [G] Indigenous Peoples are considered to have a historical continuity with pre-colonial, ancestral lands and form a nondominant, distinct community from the societies now prevailing on these lands. [G] With a comparable prevalence to non-indigenous communities, it is estimated that 12–20 million Indigenous Peoples (4 %) are living with rare diseases, and yet often face more challenges in receiving a rare disease diagnosis and accessing care services. [5,96] Many Indigenous populations may have a unique genetic risk profile due to their distinct ancestry and are often poorly represented in publicly available databases of human variation, leading to variant interpretation challenges. [5,20,96,122,149] In addition, there are cultural, ethical, geographical, and trust barriers that may limit access to genomic services, resulting in diagnostic delays and potentially missed treatment opportunities. [29] Diagnosing rare disease in Indigenous Peoples is essential for improving treatment access and health outcomes.

The International Rare Disease Research Consortium global task force has made several recommendations for addressing challenges in advancing rare disease diagnosis for Indigenous Peoples. [5] A key first step is community engagement, where there has previously been little awareness of rare diseases in the community. The prevalence of IRDs in many minority groups is not known, and there has not been a comprehensive evaluation addressing the potential support needs surrounding the diagnosis of IRDs among global Indigenous groups. A clearer understanding of the current landscape can provide insights into regional differences in diagnosis and access, identify shared challenges, and inform strategies to address issues diagnosing rare disease, ultimately enhancing health equity.

This scoping review aims to map the global research landscape of IRDs in Indigenous Peoples and synthesise clinical studies on IRDs in global Indigenous populations.

2. Results

2.1. Summary of the included studies

Database searches yielded 646 nonduplicate citations. Following study screening, 73 studies of Indigenous Peoples with IRDs from 24 countries were included, comprising 60 primary research articles, 10 conference abstracts, 2 government reports, and 1 book chapter (Figs. 1–2). Included studies were published between 1974 and 2023, with all being observational except for 1 interventional study, [141] and 42 % were case studies. Individual studies included between 1 and 34, 869 Indigenous participants (median [IQR]: 23 [7-140]), and across all studies, at least 581 well-defined cases of IRDs in Indigenous Peoples were identified.

Most studies reported on Indigenous Peoples from the Middle East

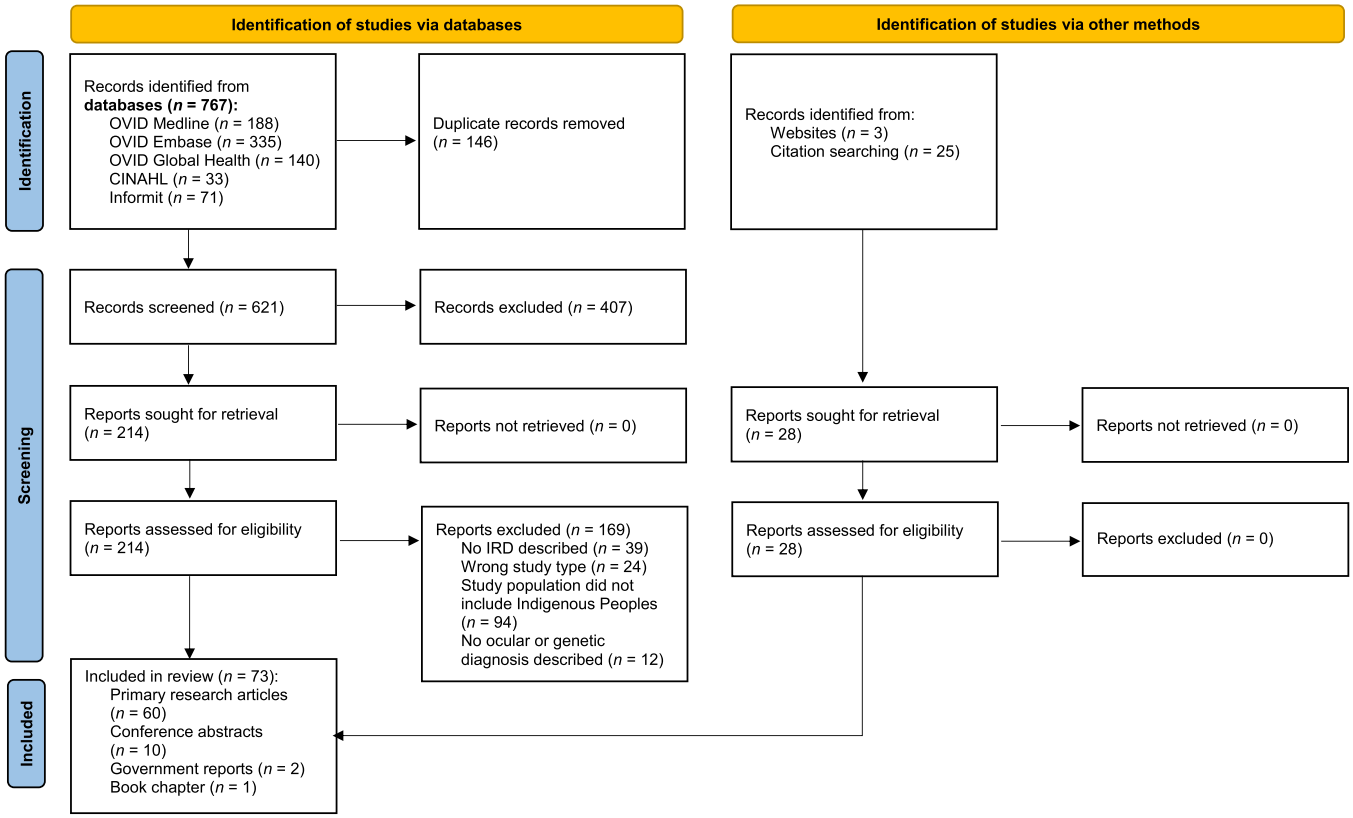

Fig. 1. PRISMA flow chart. abbreviation: IRD, inherited retinal disease.

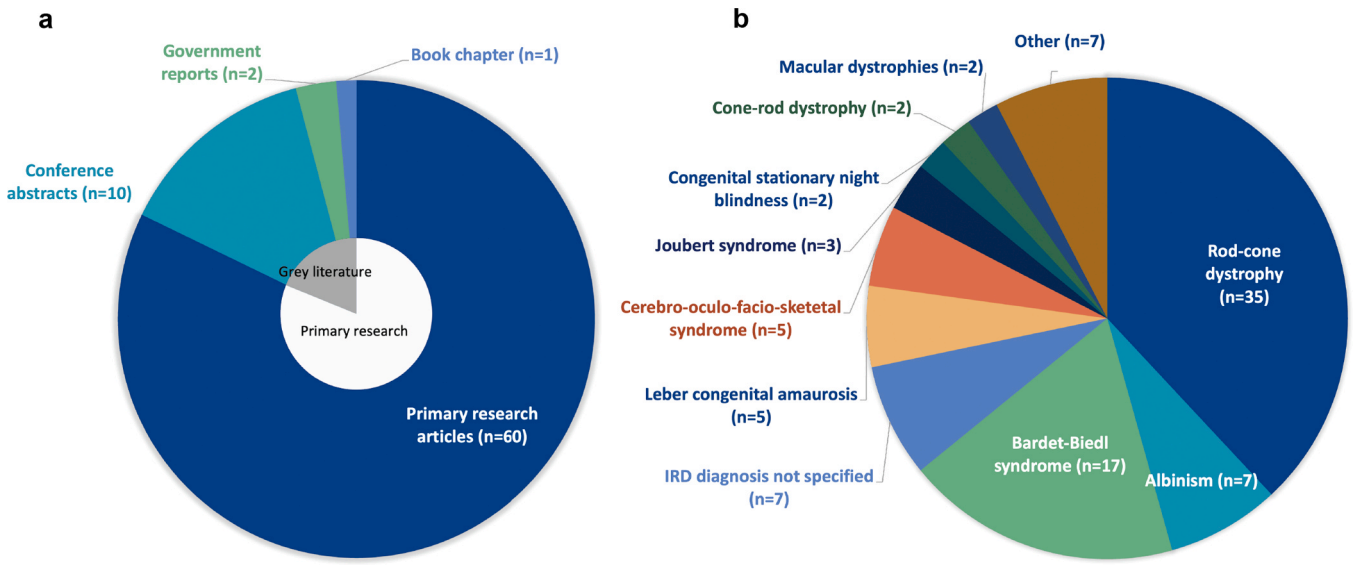

Fig. 2. **a**) The sources of evidence from the $n = 73$ included studies and **b**) the spectrum of inherited retinal diseases (IRDs) reported amongst Indigenous participants, with *n* indicating the number of reports that include each IRD. The total number of studies exceeds 73, as some studies reported on multiple IRDs.

(34 %), Oceania (27 %) and North America (23 %) (Fig. 3), and retinitis pigmentosa/rod-cone dystrophy (RCD) (41 %) was the most studied IRD (Fig. 2b). Characteristics of the included studies and conference abstracts are shown in Supplementary Tables 2 and 3, respectively.

The included studies can be broadly classified into i) population-based studies focussing on vision loss in Indigenous Peoples and ii) small cross-sectional or case studies that investigated specific Indigenous groups and their IRDs. National and subnational population-based studies identified IRDs as contributing causes of vision loss amongst Indigenous Peoples in Australia,[26,43,44,135,B,I] Malaysia, [150] North America, [46,111,142] Asia, [22,63,99] and Argentina. [122] While the majority of studies reported findings in participants who had undergone some form of genetic diagnostics (60 %), including Sanger-sequencing, linkage analysis and karyotyping, fewer described the ocular phenotype (23 %).

We summarised findings at the level of both geographic region and stratified by IRD. We acknowledge that Indigenous Peoples have had complex movements across geographies and that the names of the below spaces represent current name placements that may not be culturally relevant to some Indigenous Peoples; however, we adopted this stratification strategy to account for varying healthcare structures and service accessibility across different geographical areas.

2.2. The Americas

The Americas comprise a large landmass spanning across 2 major continents, North and South America. Native Americans have a diverse and unique ancestry that reflects pre-Columbian geological and ecological conditions[21,40,92,106] and post-Columbian admixing of

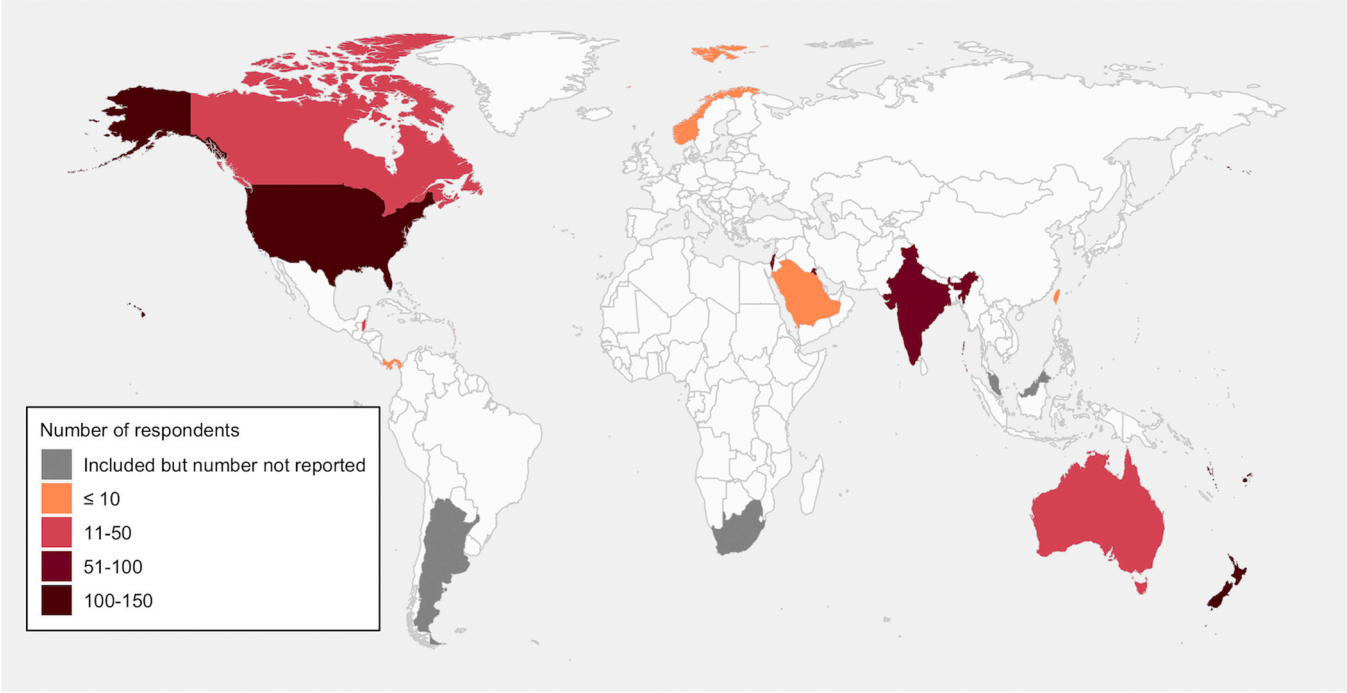

Fig. 3. The number of inherited retinal disease (IRD) cases reported in Indigenous Peoples worldwide.

ancestries related to Europeans and Sub-Saharan African groups, as seen in today's Latino populations. [2,89,130] The most recent census data estimates that 1 million Indigenous Peoples are living in Canada and 7 million in the US, making up 5 % and 2 % of the population, respectively. [121,K] A staggering 42–53 million Indigenous Peoples reside in Latin America, making up more than 700 distinct ethnic groups, [J] and accounting for 8 % of the population. [J,F] Among Native American populations, genetic associations with IRDs remain largely uncharacterised. [81,108]

2.2.1. North America

We identified 16 studies, published between 1974 and 2021, that included Native Americans diagnosed with an IRD. Of these, 13 studies recruited participants from Native American communities, and 3 included self-identifying Native Americans living in urban and regional settings. Across 5 case studies and 11 cross-sectional studies, a range of IRDs are reported, including RCD, [28,42,46,59,102,111,132,142] BBS, [102] Stargardt disease, [111] oculocutaneous albinism, [148] and cerebro-oculo-facio-skeletal (COFS) syndrome. [33,66,91,103,104]

2.2.1.1. Rod-cone dystrophy. Six studies investigated RCD in the Diné (Navajo Nation), published between 1981 and 2022. The US federally recognised lands of the Navajo Nation presently span the states of Arizona, Utah, and New Mexico, and with nearly 400,000 enrolled citizens, it is the largest Tribal Nation in the US by enrolment. [S] Of the 6 studies, 3 were population-based studies, [46,111,142] 2 were linkage analyses of the same Navajo family, [28,42] and 1 was an observational study of RCD in 76 Navajo individuals. [59] RCD has been reported to occur at a higher rate in Navajo People, [46,142] with a 2021 study reporting an incidence of 7/1152 (0.61 %) compared to an estimated 1/4000 (0.025 %) in the general US population. [142] While the clinical phenotype of RCD in Diné participants was reported in 2 studies, [46,59] the genetic characteristics are yet to be investigated, owing partially to a long-standing 2002 moratorium on genetic research by the Navajo Nation that is still in effect.

2.2.1.2. Oculocutaneous albinism. One small cross-sectional study investigated oculocutaneous albinism type 2 in 5 Diné participants from the Navajo Nation in Northeastern Arizona. [148] Although albinism has been documented among Native American populations, this is the singular study of a molecular diagnosis, which identified a novel founder mutation in the *OCA2* gene. [148] This 122.5 kb deletion of the *P* gene appeared to be specific to the Diné, as this allele was not detected in 34 other individuals with albinism who listed other non-Diné Native American or other descriptors. [148]

2.2.1.3. COFS syndrome. Five small case series published between 1974 and 2000 reported COFS syndrome in the same paediatric population of the Saulteaux First Nation living on settlements in Manitoba, Canada. [33,66,91,103,104] COFS syndrome is a rare, autosomal recessive, degenerative disorder involving the brain, eyes, and spinal cord, and it was first described in this population. [104] Only 1 of the studies commented on the ocular manifestations of COFS syndrome, reporting mild pigmentary retinopathy to severe optic nerve atrophy. [33] The only study that conducted genetic sequencing identified a novel variant in the COFS-associated *CSB* gene. [91]

2.2.1.4. Other IRDs. Alongside studies including Diné or Saulteaux First Nation members, 1 study investigates IRDs in the culturally unique and geographically isolated Tłı̨chǫ (the Dogrib, a Diné First Nations people) communities living in Rae, in the Northwest Territories, Canada. [102] This case study recorded cases of BBS and retinitis punctata albescens across 3 families and commented on the high carrier frequency in the population. [102] A community-based study from Phoenix, Arizona, also evaluated variants in the *BBS* genes through DNA analysis from over 6800 Native Americans. [32] Several rare, potentially damaging novel variants were identified in BBS genes, including in *BBS1*, *BBS2*, *BBS14* (*CEP290*), *BBS20* (*IFT170*); however, potential ocular associations were not reported in this study. [32]

One study compared the next-generation sequencing (NGS) diagnostic yield among an IRD cohort from the US, which included 6 Native Americans and 1 participant of Hmong ancestry, to a cohort from India. [149] The US cohort showed a lower diagnostic rate (39 % vs 62 % in this study), which the authors discussed to be at least somewhat attributed to the inclusion of participants with more diverse ethnic backgrounds in the US cohort; however, the diagnostic rate by specific populations was not broken down further. [149]

In addition to genetic testing, vision rehabilitation and support are crucial components of IRD management. A 1994 American study evaluating the success of vocational rehabilitation in patients with retinitis pigmentosa found that non-White individuals, including Native Americans, had poorer outcomes. [132] The accessibility of vision rehabilitation for IRDs among Native American populations has not been investigated more recently.

We did not identify large population-based studies reporting prevalence data on the burden of IRDs in Northern Native Americans. While there is ongoing research into the prevalence, treatment, and impact of IRDs in Northern American populations at large, this is yet to be elucidated specifically within Indigenous communities from the region. [6,34,50,79,118]

2.2.2. Latin America

Latin America is generally understood to comprise South America plus Mexico, Central America, and islands of the Caribbean. Between 1998 and 2023, 4 studies examined IRDs such as North Carolina Macular Dystrophy[107] and albinism[4,19] in Native Americans from Belize, [107] the Commonwealth of Dominica, [4] and Panama. [19] In addition, 66 Native Americans were included in a large Argentinian genetic analysis of inherited ocular disease, but the diagnoses were not presented according to ancestry. [122] Distinct novel variants in *OCA2* were identified in both the Kalingo population of the Commonwealth of Dominica[4] and the Kuna population of Panama, [19] which are themselves distinct from the *OCA2* mutation identified in the Diné in North America, [148] underscoring the importance of investigating Tribe- and geographic-specific genetic drivers. Although only 4 eligible studies specifically included Indigenous populations, other large population-based IRD studies have expanded the understanding of genotype-phenotype correlations and allelic heterogeneity in this region. [3,15,30,49,84,93,101,120,133,137,152]

2.3. Middle East

The Middle East is a geopolitical region that, while contentious, can be considered to encompass the Arabian Peninsula, extending to Egypt, Turkey, and Iran. According to the International Work Group for Indigenous Affairs (IWGIA), the Indigenous Peoples of the Middle East include the Arab-Bedouins of the Negev Desert in Israel and the Bedouin Jahalin, al-Kaabneh, al-Azazmeh, al-Ramadin and al-Rshaida peoples of Palestine. [M] Traditionally semi-nomadic, Bedouins had high birth rates and levels of consanguinity. [128] Consequently, Bedouin tribes have represented small genetic isolates with high rates of homozygosity for autosomal recessive genes inherited from a common ancestor. [159] For example, according to data published by the Israeli Ministry of Health between 2011 and 2013, 1 in 8 Bedouins carry a *BBS* variant. [112,128] These unique genetic features have facilitated studies of IRDs in Bedouin populations over the last 30 years.

Between 1993 and 2022, 23 studies examined IRDs in Bedouin populations. Of these, 16 studied Bedouin populations from Israel, with the remaining studies including Bedouins from Palestine, Kuwait, and Saudi Arabia. A diverse range of IRDs were investigated, including BBS, [17,18,24,25,38,41,77,95,97,119,127,147,155,156] Hermansky-Pudlak syndrome, [124] RCD, [7,100,146] cone-rod dystrophy (CORD), [100] macular dystrophy

(MD), [100] Leber congenital amaurosis (LCA), [51,100,154] Joubert syndrome, [154,155] Alström syndrome, [72] achromatopsia, [151] and a spectrum of familial exudative vitreoretinopathy, persistent foetal vasculature, and Norrie disease. [48] Eighteen of the 23 studies recorded consanguinity in participants' family histories.

Of the 14 studies investigating BBS in Bedouin populations, [17,18,24,25,38,41,77,95,97,119,127,147,155,156] 7 studied the same large Bedouin kindred. [17,25,38,77,95,97,127] Eleven studies included genetic sequencing such as homozygosity mapping, linkage analysis, karyotyping, and more recently, Sanger and whole exome sequencing. As a result, many of the 21 known BBS-causing genes[45] were identified in these studied populations, for example, *TRIM32*, [24] *ARL6*, [25] *BBS2*, [97] and *BBS4*, [95] underscoring the value of evaluating relative IRD prevalence among minoritised Indigenous groups. Compared to the genetic profiles, ocular presentations of BBS in Bedouin peoples remain largely underreported, having only been described in 1 study. [147]

Several other IRDs have also been described in the Bedouin Peoples. Three studies described data from the Israeli national genetic database on autosomal recessive diseases and their causative mutations amongst the different ethnic groups in Israel and Saudi Arabia. [154–156] This database was launched in 2006 and serves as a repository of information on monogenic disorders in various Israeli populations. [160,R] Since 2006, several novel variants have been discovered, for example, in congenital vitreoretinal disease, [48] familial Alström syndrome, [72] RCD, [146] and LCA. [51] Furthermore, several studies reported founder mutations in *RP1* (associated with autosomal recessive MD, RCD, and CORD, showing phenotypic diversity), [100] *GUCY2D* (associated with LCA), [51] and *RPGRIP1* (associated with LCA/early-onset severe retinal dystrophy)[100] in this population. This research informs the Israeli Government's bespoke, funded, carrier screening program for family planning, which, since its launch in 2013, has been used to screen for targeted disease-causing variants, for example, in LCA, Stargardt disease, and BBS, based on ancestry and region. [57,156–158]

2.4. Oceania

Oceania is a region comprising Australasia, Melanesia, Micronesia, and Polynesia. The Indigenous Peoples of Oceania are genetically distinct and made up of Aboriginal and/or Torres Strait Islanders (respectively hereafter referred to as Indigenous Australians), the Papuans, and the Austronesians (Melanesians, Micronesians and Polynesians including Māori). [47] Indigenous Australians have continuously occupied their lands for over 40,000 years[60,87,110] and display astonishing genetic diversity, [87] greater than that seen between people living in Europe and Asia. [35,61,88] Contrastingly, the Māori people of New Zealand arrived from Eastern Polynesia in the late 13th to early 14th century and demonstrate reduced genetic diversity, with genetic signatures owing to a history of founder effects and genetic drift. [36,47,94,145]

2.4.1. Australia

Published between 2009 and 2023, 7 studies and 2 government reports included data on Indigenous Australians with IRDs. Seven were population-based studies on vision loss[26,43,44,123,135,B,I] and 2 were small cross-sectional studies investigating RCD[116] and oculocutaneous albinism. [67] From the population-based studies, at least 2 confirmed cases of RCD were recorded in the National Indigenous Eye Health Survey[135,B] and a diabetic retinopathy cohort study in Western Australia. [26] Four Indigenous Australians with IRDs were included in a recent study evaluating healthcare and societal costs of IRDs in Australia, but their IRD diagnoses were not specified. [123] Based on these studies, further research is needed to understand the relative prevalence of IRDs in Indigenous Australians.

The largest study was a longitudinal case report of a three-generation Indigenous Australian family with *PRPF31*-associated RCD, including 12 affected members and 2 asymptomatic carriers. [116] *PRPF31* is implicated in up to 10 % of autosomal dominant RCD with incomplete penetrance. In this family, disease was caused by a heterozygous nonsense variant (c.1205C>A; p.Ser402Ter), which was also absent in variant population databases, and has since been reported in only 1 other individual with RCD in a later article published in 2021. [27] The final study documented 1 Indigenous Australian with oculocutaneous albinism among a cohort of 132 Indigenous participants from Oceania. [67]

2.4.2. New Zealand

Six studies and 1 book chapter included data on Polynesian (New Zealand Māori, Cook Island Māori, Samoan, Tongan, Niuean, Tahitian, Tuvaluan) and Melanesian Peoples (Fijian and ni-Vanuatu) with IRDs. [62,65,67,76,139,140,141] One was a participant of Native Hawaiian or Pacific Islander origin included in the natural history of the progression of X-linked RCD study (XOLARIS) as a subset of a clinical trial (NCT04926129). [141] A range of IRDs have been studied, including oculocutaneous albinism, [67] RCD, [76,139–141] X-linked incomplete congenital stationary night blindness (CSNB), [62,139] LCA/early-onset severe retinal dystrophy, [139] MD, [76,139] and CORD. [76,139]

Six studies from the region characterised the molecular diagnoses, [62,65,67,76,139,140] identifying several novel disease-causing variants in Māori and Samoan patients with *RP2*- and *RPGR*-associated X-linked IRDs[76] and a large Māori family *CACNA1F*-associated CSNB. [62] Novel founder mutations were also identified, including in *OCA2* in Tuvaluan with oculocutaneous albinism[67] and in *PDE6B*, which accounts for up to 16 % of recessive IRDs in Māori. [140] Despite these advances, many New Zealand Māori and other Polynesians with IRDs remain genetically unsolved. [139] It has been estimated that over half of the Māori and Polynesian patients with inherited RCD had no pathogenic variant(s) detected with targeted NGS, supporting novel IRD variants in this population. [140]

2.4.3. IRD programs in Oceania

While IRDs in Oceanic Indigenous populations are relatively understudied, several countries are establishing programs for equitable eye health research in IRDs and indigenous genomics. Three national population-based surveys have reported epidemiological data on vision loss and eye disease in Indigenous populations from Australia[44,135] and Fiji,[109] and several IRD-specific registries aim to improve access to research and emerging treatments. [13,140,L] In addition, the National Centre for Indigenous Genomics was established to protect Indigenous Australian genomic data sovereignty. [D] Researchers from the University of Auckland are setting a global standard by recognising the importance of understanding regional ancestry and iwi (tribe) in the diagnostic process, improving genomic care for Indigenous Peoples with IRDs in this region. [37,140]

2.5. Africa

Africa consists of 54 countries with over 2000 ethnolinguistic groups. [16,O] Being the most ancient of all populations, Africa harbors the highest levels of human genetic variation[16,90,125] due to population admixture, migration, and environmental exposures. [16] While there are local proponents of the concept, 'indigeneity' in Africa is complex and contentious. [M] Many African states still either deny the existence of Indigenous Peoples or argue that everyone is indigenous. [86,144,M]

Using the IWGIA definition of Indigenous groups, [M] only 1 case study from Africa was eligible for inclusion. [52] Published in 1999, this study included Indigenous Khoe-San South African participants and non-indigenous participants diagnosed with RCD, and newly described the Pro-347-Leu mutation in *RHO* in this population. [52]

The concept of indigeneity aside, there is an evolving body of research on IRDs in African populations. A significant body of this work was undertaken in this region as part of the Retinal Degenerative Disorders project, established in 1990, and now with over 3000 members, serves as an invaluable resource. [85,113,114,115,126] As a part of this work, several founder mutations in IRDs have been identified in these

populations, including *MYO7A*-related Usher syndrome, [113] *SCA7*-related spinocerebellar ataxia, [53,129] and *ABCA4*-related macular dystrophies. [126]

2.6. Asia

Asia is the largest continent in the world and is home to 260 million, or two-thirds, of the world's Indigenous Peoples. [H] The concept of indigeneity in Asia is complex. Some countries, like Cambodia, Malaysia, and Taiwan, officially recognise Indigenous Peoples, while others, such as China and India, use alternative terms like "ethnic minorities" or "scheduled tribes", respectively. [M] Four epidemiological studies, published between 2002 and 2021, reporting on the prevalence of ocular disease or vision impairment included Indigenous participants with IRDs. [22,63,99,150] From Eastern Taiwan, the Amis study reported up to 6 cases of RCD among over 2300 Amis Peoples from the Hualien-Taitung Valley, [22,63] and from Eastern India, the Tribal Eye Disease Study reported 70 cases of RCD among over 34,800 tribal people from Odisha. [99] Cases of RCD ($n = 4$) and albinism ($n = 1$) were identified as causes of blindness and low vision from the 1996 Malaysia National Eye Survey of 18,027 participants, including 1740 Indigenous participants. [150] The prevalence of IRDs among Indigenous participants was not reported separately; however, Indigenous participants, particularly females, had higher rates of blindness overall compared to Chinese, Indian, and Malay ethnic groups. While there are several published studies on the genotypic and phenotypic spectrum of IRDs in Asian populations, [10,23,54,73] this is yet to be done in Asia's Indigenous populations.

2.7. Europe

Most Indigenous Peoples in Europe live in the Arctic region, such as the Sámi Peoples in Norway, Finland, Sweden, and Russia's Kola Peninsula, and the Kalaallit, the Greenlandic Inuit ethnic group. [M] In contrast to the modern European gene pool made up of an admixture of 3 ancestral populations, [80] the origin and genetic landscape of these Indigenous groups are less well-defined. In this context, it was not unexpected that we found only 1 study, which described a linkage analysis of a large Norwegian Sámi family with 5 confirmed cases of Usher syndrome Type IIB. [39] The Sámi are often referred to as 'extreme genetic outliers' amongst European populations[68,134] and exhibit distinct features such as reduced genetic heterogeneity and high levels of linkage disequilibrium attributed to genetic drift. [69,71,78,117] As such, further studies are warranted to investigate the prevalence, diagnosis, and genetic profile of IRDs in Sámi peoples and European Indigenous Peoples at large.

3. Discussion

This scoping review comprehensively synthesised 73 studies of IRDs in global Indigenous groups from 24 countries. The research on this topic is era- and region-dependent. Specific IRDs have been widely studied in populations where they are geographically and/or culturally concentrated, for example, BBS in the Middle East, RCD in Diné of the Navajo Nation, and COFS syndrome in the Saulteaux First Nation, Manitoba, Canada. With specific research programs for targeted diagnosis and understanding founder mutations, IRDs in Indigenous Peoples from New Zealand and the Middle East were comparatively the most well-studied. National epidemiological data from Australia and Malaysia have reported cases of IRDs as a cause of vision loss among Indigenous Peoples, but data on the relative prevalence of specific IRDs is lacking across most global regions.

Overall, there is a paucity of data on the ocular presentations of many IRDs among Indigenous Peoples, as well as specific data on the socioeconomic impact of IRDs in these populations. Raising awareness and understanding the disease burden is a first step towards bridging the gap in the current state of IRD care for Indigenous Peoples worldwide, to ultimately address global health equity for rare disease diagnosis in Indigenous populations. [5]

3.1. Implications for clinical care

This study revealed gaps in diagnosing IRDs and disparities in care access among Indigenous populations worldwide. Minoritised populations face additional barriers in obtaining an IRD diagnosis, including limited access to culturally safe specialised healthcare services and vision rehabilitation programs. [8,9,29] Geographic isolation often compounds these issues, making it difficult for individuals to receive consistent and comprehensive care. Cultural differences, language barriers, and limited awareness of available resources can also hinder effective communication and vision care delivery, impacting patient outcomes. [29] Recognising the prevalence of IRDs in Indigenous communities and addressing barriers to diagnosis are essential for improving clinical care and ensuring equity of access to emerging treatments. The first steps to addressing gaps in diagnosis include engaging communities to raise awareness, increasing culturally safe access to genomic services, and supporting community-led, self-determined initiatives to put indigenous eye care into indigenous hands.

Existing population variant databases are largely composed of individuals with European ancestry. As a result, Indigenous Peoples are underrepresented in global databases of human variation. While the current diagnostic yield of NGS for IRDs is between 52 % and 74 %,[11] it is almost always lower in minority ethnic groups. The lack of population-specific variant data makes determining the significance of novel variants challenging. [20,122,149] Additionally, there is likely underreporting of potential disease-causing variants from minority populations due to limited access to genetic testing and difficulties in generating sufficient evidence for assessing novel variant pathogenicity. Without a genetic diagnosis, individuals living with IRDs, including Indigenous Peoples, cannot participate in gene-specific clinical trials and treatments.

3.2. Implications for research

Several countries are working to establish and support community-led national indigenous biorepositories, for example, the Australian National Centre for Indigenous Genomics. [D] Research into Indigenous Peoples must always consider, protect, and enhance indigenous data sovereignty under the CARE (collective benefit, authority to control, responsibility and ethics) principles, [64,A] or the right of Indigenous Peoples to govern the collection, ownership, and application of data about indigenous communities, peoples, lands, and resources. [C] At its worst, research has been conducted 'on' and not 'with' indigenous groups, and historically has been highly exploitative. [55] Except for a few notable studies, for example, the Australian national prevalence surveys, [43,44,135,136,B,I] few studies explicitly reported consulting with indigenous communities or seeking approval from relevant indigenous peak bodies and councils. Co-designing research is essential to ensure that future research is community-driven, prioritises community values, and generates translatable outcomes that primarily benefit indigenous communities.

Bearing these concepts in mind, supporting rare disease research in minoritized groups can deepen the global understanding of IRD pathogenesis, identify population-specific allelic differences and phenotypes, and ultimately improve diagnosis. IRDs are rare diseases, and the more people with a genetic diagnosis, the more likely it is that treatments can be developed. Indeed, many studies postulated whether their participants harboured previously unreported IRD variants, and that studying indigenous populations could facilitate understanding of additional associated gene variants for diseases such as IRDs. [114,140]

Particularly in developed countries, where progress is being made to address the gap in some preventable eye diseases, a clearer

understanding is needed of the prevalence and impact of IRDs in many indigenous groups, including potential differences in phenotypic expression of the same disorder or variant in Indigenous and non-indigenous communities. As demonstrated in New Zealand and Israel, a better understanding of IRD genetic and phenotypic presentations in indigenous populations could enable population-specific genetic testing approaches, facilitating earlier diagnosis and minimising variants of uncertain significance.

3.3. Strengths and limitations of the review

We chose the design of a scoping review to systematically map and synthesise all the clinical literature on this topic and identify areas where future research is required, using a rigorous and transparent approach. A comprehensive and inclusive search strategy was devised alongside information specialists[14] and followed recommendations from a recent review on searching the literature for global Indigenous groups.[56] We searched widely indexed international databases and Informit, the largest online indigenous literature database, using a combination of region-specific indigenous search filters, including non-English terms (Appendix A);[Q] however, alternative and non-English spellings could have been missed. Additionally, we acknowledge that we did not specifically search non-English databases due to our team's limited ability to assess these sources. Many of the early studies were anthropological in nature and reported on Indigenous Peoples living on reserves, or in isolated villages, with varying levels of sampling bias.

We were surprised to find few reports of more common IRDs, such as Stargardt disease, in the indigenous populations included in this review. Stargardt disease is likely present in indigenous populations due to its prevalence; however, relevant cases may not have been identified either because studies did not specify whether participants were indigenous, or the regions where this was done did not meet the indigenous criteria included in our review. Research has been undertaken evaluating *ABCA4* variants, including founder mutations, in Tunisia and African populations,[114,115] and these findings have been discussed in other reviews.[98] Further investigation, however, is needed to understand the prevalence of *ABCA4* in indigenous groups included in the present review.

Our scope was narrow, including only studies of IRDs in indigenous groups. The concept of 'indigeneity' is nuanced and is dependent on patterns of European colonisation. We have, therefore, not represented minority groups from countries such as China (Uighurs, Tibetans, *etc.*) and Africa, that do not fall into the definition of 'Indigenous populations' yet are likely populations in which IRDs may not be well studied.[1,70,83,85,113,114,143,153] Our study did not address the separate issue that all people from countries with limited genetic testing, whether indigenous or not, face challenges obtaining adequate diagnosis and support services for IRDs.

4. Conclusion

Engaging communities and raising awareness of IRDs are important steps for improving the care of these debilitating conditions, and our study underscores the importance of recognising their presentation in Indigenous Peoples. Ophthalmology is currently at the forefront of a genetics revolution, and efforts must be made to ensure that the whole community benefits from this rapidly advancing field. Failure to address these issues will exacerbate the health disparities that already exist between global indigenous and non-indigenous communities. Engagement, co-designed approaches and collective efforts, including raising awareness, could open pathways to address challenges limiting equitable access to IRD diagnosis for Indigenous Peoples, facilitating access to emerging treatments.

5. Methods

This scoping review was undertaken according to an *a priori* protocol (doi.org/10.17605/OSF.IO/YNU9K)[E] and follows the recommendations described in the Joanna Briggs Institute methodological guidance for the conduct of scoping reviews.[105] Reporting was conducted according to the Preferred Reporting Items for Systematic reviews and Meta-Analysis extension for Scoping Reviews (PRISMA-ScR).[138]

5.1. Literature search strategy

A comprehensive search strategy (Appendix A) was devised in consultation with information specialists, following recommendations by Harding et al.[56] The strategy additionally incorporated a combination of population and region-specific keywords tailored to identify literature related to Indigenous populations globally, developed by researchers at the University of Alberta.[14,Q] The following databases were searched on 4 December 2023: Ovid Medline, Ovid Embase, Ovid Global Health, Informit and CINAHL. References lists of review articles identified at the full-text stage were manually searched. No date, language, or publication restrictions were set.

5.2. Study selection

Eligible studies included articles describing adult or paediatric Indigenous populations with IRDs, including syndromic conditions (Supplementary Table 1). We included Indigenous populations according to regions defined by the International Work Group for Indigenous Affairs[M] and definitions by the United Nations Permanent Forum on Indigenous Issues.[P] If the study did not state whether participants were indigenous but included specific keywords (village, tribe, elder, clan, communit*, ancest*), the corresponding author was contacted for further information about the study population and given 4 weeks to respond, or the study was excluded. We excluded anthropologic studies on albinism that did not describe an ocular phenotype or genetic diagnosis, and these studies have been reviewed elsewhere.[74,75]

Duplicate entries from database searches were identified and removed. Titles and abstracts were independently screened by 2 reviewers (ETC and ACBJ) in Covidence (Veritas Health Innovation, Melbourne, Australia). Potentially relevant sources were retrieved, and full texts were screened by 2 independent reviewers (ETC and ACBJ), who were masked to each other's decisions. Reasons for the exclusion of sources of evidence at full text were recorded. Any disagreements between reviewers at each stage of study selection were resolved through discussion and consensus.

5.3. Data extraction and presentation

Data was extracted in Covidence by 2 reviewers for the first 10 papers (ETC, ACBJ), and by 1 reviewer (ETC) for the remaining and checked by a second reviewer (ACBJ). Information included i) study information and design, ii) participant characteristics including country they are indigenous to, iii) IRD and ocular phenotype and iv) genetic testing method.

We stratified the included studies into colonial geographic distributions that participants were indigenous to and provided a descriptive summary of the key findings. Study characteristics were also presented in tabulated formats (Supplementary Table 2 and 3).

CRediT authorship contribution statement

Alexis Ceecee Britten-Jones: Writing – review & editing, Writing – original draft, Supervision, Resources, Project administration, Methodology, Funding acquisition, Data curation, Conceptualization. **Emma C. Tovey Crutchfield:** Writing – review & editing, Writing – original draft, Methodology, Investigation, Data curation, Conceptualization. **Andrea**

L. Vincent: Writing – review & editing, Conceptualization. **Hugh R. Taylor:** Writing – review & editing, Conceptualization. **Mitchell D. Anjou:** Writing – review & editing, Conceptualization. **Krystal S. Tsosie:** Writing – review & editing, Conceptualization. **Shaun Tatipata:** Conceptualization. **Lauren N. Ayton:** Writing – review & editing, Supervision, Resources, Project administration, Conceptualization. **Livia S. Carvalho:** Writing – review & editing, Conceptualization.

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Declaration of Competing Interest

The authors declare the following financial interests/personal relationships which may be considered as potential competing interests: ACBJ reports financial support was provided by the University of Melbourne. LNA reports financial support was provided by the NHMRC. KST co-founded and serves in a non-compensatory board of director role for the Native BioData Consortium, an Indigenous-led biological and data repository operating with the jurisdiction of a US Tribal Nation. KST and NativeBio are unaffiliated with ocular-related primary research. If there are other authors, they declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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Appendix A. Supporting information

Supplementary data associated with this article can be found in the online version at doi:10.1016/j.survophthal.2025.06.005.

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