



# The Norfolk Island Eye Study (NIES): Rationale, Methodology and Distribution of Ocular Biometry (Biometry of the Bounty)

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**Aim:** To describe the recruitment, ophthalmic examination methods and distribution of ocular biometry of participants in the Norfolk Island Eye Study, who were individuals descended from the English Bounty mutineers and their Polynesian wives. **Methods:** All 1,275 permanent residents of Norfolk Island aged over 15 years were invited to participate, including 602 individuals involved in a 2001 cardiovascular disease study. Participants completed a detailed questionnaire and underwent a comprehensive eye assessment including stereo disc and retinal photography, ocular coherence topography and conjunctival autofluorescence assessment. Additionally, blood or saliva was taken for DNA testing. **Results:** 781 participants aged over 15 years were seen (54% female), comprising 61% of the permanent Island population. 343 people (43.9%) could trace their family history to the Pitcairn Islanders (Norfolk Island Pitcairn Pedigree). Mean anterior chamber depth was 3.32mm, mean axial length (AL) was 23.5mm, and mean central corneal thickness was 546 microns. There were no statistically significant differences in these characteristics between persons with and without Pitcairn Island ancestry. Mean intra-ocular pressure was lower in people with Pitcairn Island ancestry: 15.89mmHg compared to those without Pitcairn Island ancestry 16.49mmHg ( $P = .007$ ). The mean keratometry value was lower in people with Pitcairn Island ancestry (43.22 vs. 43.52,  $P = .007$ ). The corneas were flatter in people of Pitcairn ancestry but there was no corresponding difference in AL or refraction. **Conclusion:** Our study population is highly representative of the permanent population of Norfolk Island. Ocular biometry was similar to that of other white populations. Heritability estimates, linkage analysis and genome-wide studies will further elucidate the genetic determinants of chronic ocular diseases in this genetic isolate.

■ **Keywords:** ophthalmology, glaucoma, genetics, genome-wide association, population isolates, mutiny, Bounty, Pitcairn Island

Population studies were initially designed to identify environmental factors for complex diseases but with the development of genetic epidemiology and genome-wide association studies (GWAS), population studies have become useful in identifying disease-causing genes. Population isolates offer advantages for identifying disease-associated genes because founder effects will enrich particular genes within the population, and these groups can also have high rates of participation (Sherwin

et al., 2008a). Large pedigrees have long been useful in identifying disease-related genes. Population admixture,

RECEIVED 06 August, 2010; ACCEPTED 20 October, 2010.

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particularly when diseases and genes have differing frequencies in the founder population, also assists in gene identification (McKeigue, 2005). Adding new phenotypic information to already genotyped populations enhances the efficiency of research, while investigating endophenotypes such as ocular biometry allows use of the entire cohort rather than just individuals with the disease (Charlesworth et al., 2010).

Our previous research into the genetics of the eye disease glaucoma with the Glaucoma Inheritance Study in Tasmania (Mackey, 2008) and the Twins Eye Study in Tasmania (TEST) (Mackey et al., 2009) has utilized several of the strategies above. Of particular note, as the original convict population of Norfolk Island had been relocated to Tasmania (New Norfolk, and Norfolk Plains) and Captain Bligh's *Bounty* had spent two weeks in Tasmania en route to Tahiti (Documented in the Bligh Museum of Bruny Island founded by Tasmanian Ophthalmologist John Bruce Hamilton), our research group has been very interested in the genetic research on Norfolk Island. The rate of glaucoma reported on Norfolk Island is high (Sullivan & Glasson, personal communication), despite glaucoma being uncommon in the Polynesian population; this raised the possibility that one of the European founders carried a glaucoma gene mutation that might be identified through linkage studies. In addition there are ethnic differences between the European and Polynesian populations, similar to those seen with the Aboriginal population of Australia, in some of the glaucoma endophenotypes such as Central Corneal Thickness (CCT) (Mackey, 2007).

### History of Norfolk Island

Norfolk Island was originally populated in either the fourteenth or fifteenth century by Polynesian seafarers who came from the Kermadec Islands located north of New Zealand. Their disappearance from the island after several generations remains a mystery (Macnaughtan, 2001) In 1774, the British explorer Captain James Cook went ashore and took samples of plants back to Britain. Norfolk Island was unpopulated at the time the famous 'Mutiny on the *Bounty*' took place on April 28, 1789. Following the mutiny, Fletcher Christian took the *Bounty* back to Tahiti, where some English chose to remain. However, nine British mutineers, 12 Tahitian women and six Tahitian men went in search for a haven, safe from the British Navy, and chose tiny Pitcairn Island (Clarke, 1986). Pursuit came as soon as 1790 in the form of HMS *Pandora*. Pitcairn Island (4.6km<sup>2</sup>), which previously had a Polynesian settlement and thus established food gardens, was incorrectly charted at that time and not easily found. Thus the Mutineer population remained isolated until rediscovered by the *Topaz* in 1808, when Friday October Christian swam out to meet the American visitors (Wikipedia, 5 August 2010). Although all but one adult male on Pitcairn was killed by 'civil war', the children

flourished, having their own families, and the subsequent population explosion caused problems on the small island, likely similar to those of the earlier Polynesian settlement a century before (Diamond, 2005). A disastrous attempt to relocate to Tahiti in 1831 resulted in 16 deaths, prompting a return to Pitcairn (Williams & Bataille, 2006).

Norfolk Island (34.6km<sup>2</sup>) is situated 1,600 km north-east of Sydney, on the Norfolk Ridge that runs from New Zealand to New Caledonia. There were two periods of settlement in the 19th century, when it served as a penal outpost of Sydney. The first took place from 1788–1814 and the second from 1824 until the last convict settlers were transported to Tasmania in the 1850s (Hoare, 1999). Earl Grey and Queen Victoria 'gifted' the island to the descendants of the *Bounty* mutineers and their Tahitian wives who were heading towards crisis on Pitcairn Island, with a population growth rate overtaking available food and water supplies. No copy of the original Victorian document exists, with the island's level of independence from Australia a constant source of disagreement. On June 8, 1856, the Pitcairn Islander descendants of the *Bounty* mutineers and Tahitian women relocated to Norfolk, with a total population 194 (40 men, 47 women, 54 boys and 53 girls; Williams & Bataille, 2006). This small population originated from nine paternal (*Bounty* mutineers), although with only five mutineer surnames: Christian, Young, Adams, Quintal and McCoy (Williams, Martin, Mills and Brown not having direct male-line descendants). Three male Pitcairn settlers arrived later: Evans (@Pitcairn 1823), Buffett (@Pitcairn 1823) and Nobbs (@Pitcairn 1828), and 12 maternal (Tahitian) lineages. There are no descendants from the six Tahitian men who moved to Pitcairn. In 1858 16 individuals returned to Pitcairn and in 1863 a further 30 returned. They were mainly the descendants of Ned Young, who was born in St Kitts and was said to have an English father and West Indian mother. The Pitcairn and Norfolk Island populations were then effectively isolated 6,000 km apart for the next 120 years.

### The Norfolk Island Pitcairn Pedigree

Norfolk Island's family history is particularly well documented, in large part because anthropologists from the island have maintained an exhaustive genealogical history in the form of a single large family pedigree spanning 250 years. In addition to Norfolk Island's obvious geographical isolation, strict immigration and quarantine legislation has restricted new founders from migrating to Norfolk Island (Macgregor et al., 2010). The main Norfolk Island Pitcairn Pedigree contains 5,742 individuals, of whom 1503 were consanguineous. The average inbreeding coefficient, *F* of the inbred individuals was 0.044, with a maximum of 0.16 (0.16 indicates slightly more gene sharing than would result from an avuncular marriage). To explore and quantify the current levels of genetic admixture in the Norfolk Islanders, 128 Ancestry

Informative Markers (AIMs) spread across the autosomes, X/Y chromosomes and mitochondrial DNA genome were recently genotyped (McEvoy et al., 2009). On the basis of autosomal AIMs, the population shows mean European and Polynesian ancestry proportions of 88% and 12%, respectively. However, there is a substantial variation between individuals, ranging from total European ancestry to near total Polynesian origin. There is a strong correlation between individual genetic estimates of Polynesian ancestry and those derived from the extensive pedigree and genealogical records of Islanders. Also in line with historical accounts, there is a substantial asymmetry in the maternal and paternal origins of the Islanders with almost all Y-chromosomes of European origin whereas at least 25% of mtDNAs appear to have a Polynesian origin. On average one-third of the genomes of present-day Islanders (Norfolk Island Pitcairn Pedigree members) are derived from 17 initial founders (Macgregor et al., 2010).

#### Earlier Genetic Research on Norfolk Island

The first 'genetic' study on Norfolk Island was published in 1927. Dr Shapiro 'found these islanders to be of sound physique, taller than the average English and Tahitians, and of good mentality'. The Norfolk Islanders prove that when the stock is sound to begin with, intensive inbreeding makes for no decrease in stamina. Likewise, race mixture, in his opinion, brings no deterioration (no author listed, 1927). Notably, 75 years later, MacGregor et al. found that the more interrelated individuals of Norfolk Island had reduced height (Macgregor et al., 2010)

In 2000, researchers from Griffith University in Queensland began a study of cardiovascular disease (CVD) risk traits on Norfolk Island, with 602 islanders participating in this study (Bellis et al., 2005). The main traits analyzed included cholesterol, triglycerides, high-density lipoprotein (HDL) and low-density lipoprotein (LDL) levels; height; weight; body mass index; systolic blood pressure; and diastolic blood pressure (Macgregor et al., 2010). Hypertension, diabetes and obesity are known to be prevalent in the Polynesian admixture populations of the South Pacific (Abbott et al., 2001). The proportion of Polynesian ancestry in the present-day individuals was found to significantly influence total triglycerides, body mass index, systolic blood pressure and diastolic blood pressure. For various cholesterol traits, the influence of ancestry was less marked but overall the direction of effect for all CVD-related traits was consistent with Polynesian ancestry conferring greater CVD risk. Marker-derived homozygosity was computed and agreed with measures of inbreeding derived from pedigree information (Macgregor et al., 2010).

The Norfolk Island study population is a powerful resource for the localization of complex disease genes because it possesses a unique set of characteristics including founder effect, geographic isolation, limited variation in environmental variation, exhaustive genealogy with

well-documented historical records, and higher levels of linkage disequilibrium (LD) extending up to 9.5–11 Mb (Bellis et al., 2008a). Linkage mapping of 400 microsatellite markers spaced at approximately 10 cM in 600 individuals from Norfolk Island identified a LOD score of 2.01 on chromosome 1p36 for systolic blood pressure (Bellis et al., 2008b). Principal component and linkage analysis support the clustering of CVD risk traits and provide interesting evidence of a region on chromosome 5q35 segregating with weight, waist circumference, HDL and total triglyceride levels (Cox et al., 2009).

Norfolk Islanders travel to Australia and New Zealand for work and study. In addition, the large airport, which was constructed during World War II and described in a chapter of James Michener's 1946 *Tales of the South Pacific*, allows large jets to bring tourists to the remote island. Census data in 2001 reported that there were 1,574 permanent residents, of whom 309 were under age 15 years (Mathews, 2001). The 2006 census indicated 1,576 permanent residents on island, of whom 301 were under 15 years (Matthews, 2006). Thus it might be possible to increase the number of participants from the original 602 individuals studied.

The Norfolk Island Eye Study (NIES) would have numerous advantages for identifying glaucoma-related genes. CVD risk factors and migraine, which were also studied in the 2000 field trip, are known to be associated with glaucoma and we could cross-link the CVD and ophthalmic data to allow broader analyses. There is a paucity of data on ocular disease amongst Polynesians; it is feasible that a European founder could be identified through the Norfolk Island pedigree. Clustering of glaucoma in nuclear families within larger pedigrees (Sack et al., 1996) suggests recessive modifier genes that may be enriched in a consanguineous pedigree, such as found on Norfolk Island.

Population isolates have a small number of initial founding individuals, isolation with limited immigration, high levels of consanguineous mating, extensive sampling of the target population by different research studies, and a relatively homogeneous environment (Macgregor et al., 2010). Although previous success in using population isolates has involved rare Mendelian monogenic disorders, isolates are likely to be useful in the study of complex multifactorial diseases by using admixture mapping. Admixture mapping in human populations uses the principles that underlie linkage analysis of an experimental cross (McKeigue, 2005). Admixture mapping has greater statistical power than family-linkage studies for detecting genes that contribute to ethnic variation in disease risk. In comparison with association studies, admixture mapping requires far fewer markers to search the genome and is less affected by allelic heterogeneity. Where admixed populations and panels of markers informative for ancestry are available, admixture mapping can be applied to localize genes that contribute to ethnic variation in any measur-

able trait. Looking at the various glaucoma-related traits we felt that the highly heritable measure CCT (Toh et al., 2005), which has a normal distribution but different means in different populations, would be a good candidate for admixture mapping.

## Eye Study Design and Methodology

### Ethical Approval

The original cardiovascular disease study had ethics approval from the Griffith University, Human Research and Ethics Committee in Queensland. This same committee, in addition to the Human Research and Ethics Committee at the Royal Victorian Eye and Ear Hospital in Melbourne, approved the NIES. Consent was obtained to conduct the ophthalmic examination and link this with the earlier cardiovascular and genetic research as well as ongoing genetic eye research.

In addition there was local community consultation with the hospital administration, local doctors, local optometrist and visiting ophthalmologists to check that all concerns were met regarding the possible long-term impact of the study.

### Recruitment of Participants

Any individual aged over 15 years residing on or directly related to Norfolk Islanders could be included in the NIES. In reality we recruited permanent residents of Norfolk Island aged > 15 years, both with and without direct ancestry to Pitcairn Island. Persons did not need to be involved in the previous cardiovascular study, but some genetic information was already available on these individuals. A pilot research team visited Norfolk Island to discuss the study with the local community. Original study participants were mailed invitations to participate in the NIES. Articles were presented in the local newspaper and radio. The resident optometrist agreed to work on the study and refer clinic patients to the study. Word of mouth, particularly within families, helped recruit many other participants. Students at Australian universities were contacted by family or the Norfolk Islander Facebook page, and were seen in Brisbane, Melbourne or Sydney with the TEST fieldwork (Mackey et al., 2009).

As 602 individuals had been seen in the original study, we anticipated two-thirds or 400 would participate in the Eye Study, and planned a 3-week field trip to the island in October 2007. Participants were invited to contact the eye research clinic at the Norfolk Island Hospital personally or by phone to arrange an appointment. Prior to an eye examination, a member of the research team would ensure that the administrative components of the protocol were appropriately addressed. An appointment list of attending participants was reviewed and personal information and study family numbers (if assigned) checked for accuracy. Information about special requests and tests (e.g., DNA collection) was highlighted on the appointment list, par-

ticularly if the individual had not previously attended the cardiovascular study. We set up various clinic rooms at the Norfolk Island District Hospital. Health care services for the geographically isolated community of less than 1,600 permanent residents, plus temporary residents and tourists, are based at the 20-bed hospital enterprise, staffed by 2.5 full-time equivalent medical practitioners. In addition, eye care is available at a private optometry practice. Australian medical students on remote attachments through the John Flynn Placement Program spend a term each year of their training on the island (Sherwin et al., 2008b). The eye examination for the NIES was conducted by a visiting team consisting of ophthalmologists, an ophthalmology trainee, a medical student, orthoptists, optometrists and a medical photographer. During the first three weeks we saw nearly 600 people, turning away over 50, and thus we returned to the island 3 months later to examine almost another 180 participants. A further 20 students were seen in Brisbane, Sydney and Melbourne.

### Questionnaire

Participants were required to fill out a detailed questionnaire. Components of the questionnaire included:

1. Individual demographic data
2. Personal ocular history
3. Family history of ocular disease (especially macular degeneration and glaucoma)
4. General medical history
5. Ultraviolet light exposure history.

### Ophthalmic Examination

With the exception of a slit lamp and automated perimeter at the local optometrist's office, most of the equipment required for the study was borrowed from the TEST and Centre for Eye Research Australia/Royal Victorian Eye and Ear Hospital. We brought a Nidek 3DX, a Kowa portable fundus camera, UV, IOLMaster, tonopen, pachymeter, UV auto fluorescence and an OCT (type on loan from Zeiss).

Our examination protocol consisted of:

1. Visual acuity Log MAR at 3m (Precision Vision, LaSalle, IL, U.S.A.)
2. Binocular vision function tests
  - (a) Cover test
  - (b) Four Dioptre base-out prism test
  - (c) Ocular motility
  - (d) Stereopsis, Lang stereo card (Lang, Forch, Switzerland)
3. Eye dominance
4. Anterior chamber depth (ACD), keratometry, axial length (AL). IOLMaster (Carl Zeiss Meditek, Dublin, CA)
5. Conjunctival UV auto-fluorescence photos

6. Autorefracton, pre- and post-cycloplegia (Nidek, Gamagori, Japan).
7. Intraocular pressure (IOP) Tonopen handheld tonometer (Reichert, Tustin, CA).
8. Central corneal thickness Pachmate ultrasonic pachymeter (DGH, Exton, PA)
9. Dilated stereoscopic optic disc photographs Nidek 3-DX (Nidek, Gamagori, Japan)
10. Disc-centred and macular-centred retinal photographs Genesis D hand-held retinal camera (Kowa, Tokyo, Japan)
11. OCT Cirrus (Carl Zeiss Meditek, Dublin, CA, U.S.A) Three components were assessed; fast retinal nerve fibre layer (RNFL), fast macula and fast optic nerve head.
12. DNA collection, when participants in the original study had inadequate DNA available or for new participants.

The majority of participants were healthy volunteers, freely giving their time to genetic research, but taking advantage of the free comprehensive eye check with equipment that is not normally available on the island. Following completion of the examination session, each participant was given a detailed debriefing of the results of the various tests conducted and a summary of his/her ocular health status and provided with an opportunity to ask any questions. Any patients seen by ophthalmologists or optometrists previously had report letters sent to their eye care providers in Australia, New Zealand or on the island. Any newly diagnosed pathology was referred to the local optometrist (e.g., suspicious discs or pressure) for FDT visual field testing (Carl Zeiss, Dublin, CA). Any individuals from Norfolk Island requiring intervention from an ophthalmologist are usually referred to practitioners based in Australia or New Zealand. Although there is a visiting ophthalmologist to the Island approximately every 12 months, ophthalmic surgery does not usually take place there. Baseline photographs were provided to the local hospital and optometry clinic for all participants' future use. A feedback report outlining the participant's

eye measurements was provided to all participants. These baseline data and ocular imaging are important benefits and useful to participants should they develop an eye condition in the future.

#### Additional Photographic Analysis Data

The Nidek 3-DX stereo images were digitized and analyzed with a stereo Z-screen (StereoGraphics, Beverly Hills, CA) using high-resolution, 16-bit colour images, high-speed modulating stereoscopic panel and passive polarized eyewear. The cup and disc were delineated with custom software (developed by James Morgan; Cardiff, UK) and adjusted for magnification using refraction and Keratometry readings. Further analysis of optic disc stereo photos, retinal vasculature and conjunctival autofluorescence are in process. UV areas of fluorescence were measured using area calculation functions in image processing software (Adobe Photoshop). Details of the exact methods are given in our parallel twin study (Mackey et al., 2009). The main differences between the two studies are the use of the Nidek autorefractor and the Cirrus OCT on Norfolk Island.

#### Statistical Analysis

Chi-square tests were used to assess differences in categorical data and the t-test was used for normally distributed continuous data. The Mann-Whitney test was used to assess differences in non-parametric data. All analyses were performed using the GraphPad Prism v5.00 for Windows (GraphPad Software, San Diego, CA, www.graphpad.com), and results were considered significant for  $P < .05$ . No corrections for multiple testing were performed.

## Results

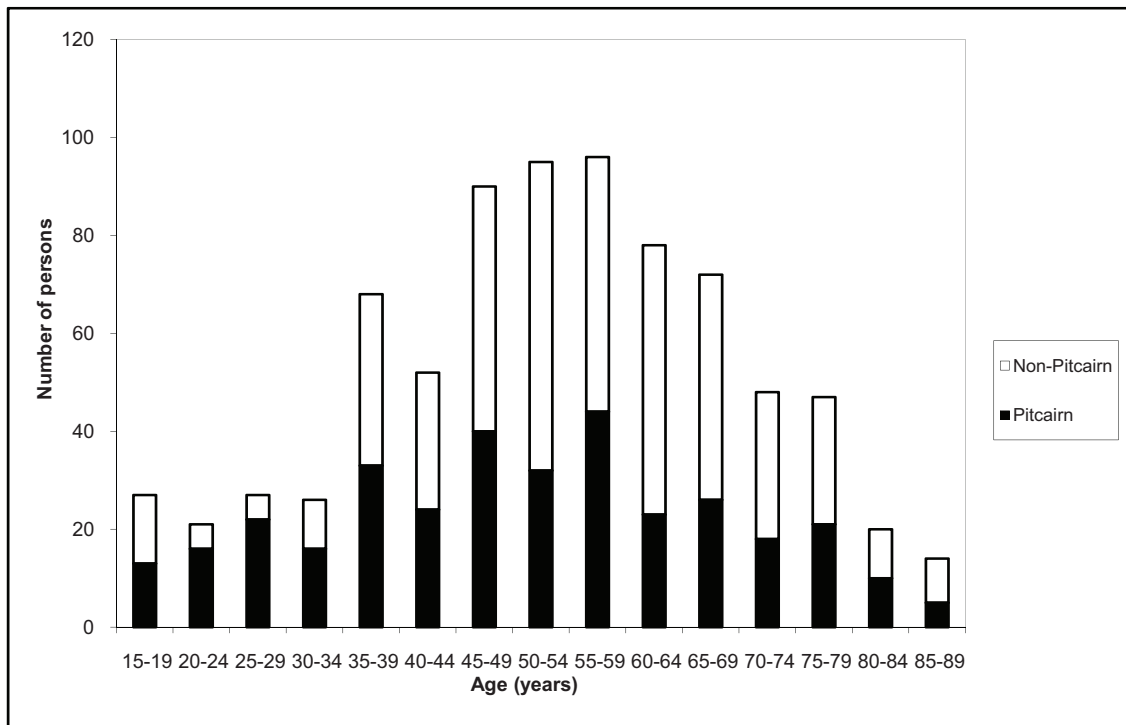
### Participants

781 participants aged over 15 years were seen. 352 (45.1%) were male. The mean age of participants in the NIES was 53.12 years (SD 16.45, range 15–89 years). The age and sex breakdown is detailed in Table 1. The NIES population included 61% of the population compared with the 2006 census data and the age demographics of participants gen-

**TABLE 1**

Age and Gender Distribution in the NIES (The proportion of people reviewed relative to the permanent population of Norfolk Island (As per the 2006 Norfolk Island Census) is also displayed)

Age	Number NIES	(%)	Male	(%)	Female	(%)	Proportion of permanent NI population reviewed
15–29	75	9.70	36	48.00	39	52.00	0.45
30–39	94	12.04	45	47.87	49	52.13	0.46
40–49	142	18.18	50	35.21	92	64.79	0.59
50–59	191	24.46	86	45.03	105	54.97	0.73
60–69	150	19.21	71	47.33	79	52.67	0.71
Over 70	129	16.52	65	50.39	64	49.61	0.67
Total	781	100.00	353	45.20	428	54.80	0.61

**FIGURE 1**

Age distribution of NIES participants showing ratio of Pitcairn to non-Pitcairn ancestry.

**TABLE 2**

Comparison of Number of Persons with Pitcairn Island Heritage in the Census Compared with NIES

	2006 Norfolk Island census				Norfolk Island Eye Study			
	Males	Females	Total	% Total	Males	Females	Total	% Total
Age ≥ 15 years								
Yes	296	252	548	43.0	157	186	343	43.9
No	326	395	721	56.5	195	243	438	56.1
Not stated	3	3	6	0.5	N/A	N/A	N/A	N/A
Total	625	650	1275	100	352	429	781	100.0

**TABLE 3**

Ocular Biometry in the NIES

Ocular Trait	All NIES			Pitcairn Ancestry		No Pitcairn Ancestry		P
	Mean	(SD)	Range	Mean	(SD)	Mean	(SD)	
ACD	3.32	(0.4)	2.16–4.83	3.34	(0.4)	3.3	(0.4)	0.219
AL	23.55	(0.93)	21.12–28.89	23.58	(0.81)	23.53	(1.02)	0.471
CCT	546.34	(33.57)	426–657	544.59	(32.34)	547.72	(34.49)	0.200
IOP	16.23	(3.09)	8–31	15.89	(2.74)	16.49	(3.32)	0.007
Keratometry (H)	42.95	(1.55)	36.75–47.75	42.78	(1.46)	43.08	(1.61)	0.008
Keratometry (V)	43.82	(1.56)	39.13–53.5	43.67	(1.57)	43.94	(1.55)	0.017
SE	0.556	(1.42)	-9–8	0.24	(2.24)	0.48	(1.53)	0.617
CDR	0.405	(0.196)	0–1	0.41	(0.21)	0.4	(0.19)	0.732

Note: The difference between individuals with and without Pitcairn Island ancestry is displayed. ACD, Anterior chamber depth (mm); AL, Axial length (mm); CCT, Central corneal thickness (μm); IOP, intra-ocular pressure (mmHg); H, horizontal curvature (D); V, vertical curvature (D); SE, spherical equivalent (D); CDR, Vertical Cup-to-disc ratio.

erally reflected those of the permanent population (Table 1). 343 people (43.9%) could trace their family history to the Pitcairn Islanders (Norfolk Island Pitcairn Pedigree). Of these, 186 (54.2%) were female, and 157 (45.8%) were male. Compared with the census data, Pitcairn pedigree members were no more or less likely to participate in the NIES (Table 2). The age distribution of participants from both groups is shown in Figure 1.

The population distribution of biometry for ACD, AL, CCT, IOP, keratometry, Spherical Equivalent (SE) and cup:disc ratio (CDR) is shown in Table 3 and Figures 2–6: The IOP was lower in people with Pitcairn ancestry but there was no difference in mean CCT. Mean IOP was 15.89mmHg in those with Pitcairn ancestry compared to 16.49mmHg in non-Pitcairn descendants ( $P = .007$ ). Mean K value (H+V) was less in people with Pitcairn Island ancestry (43.22 vs. 43.52,  $P = .007$ ). The corneas were flatter in people of Pitcairn ancestry but there was no corresponding difference in AL or refraction.

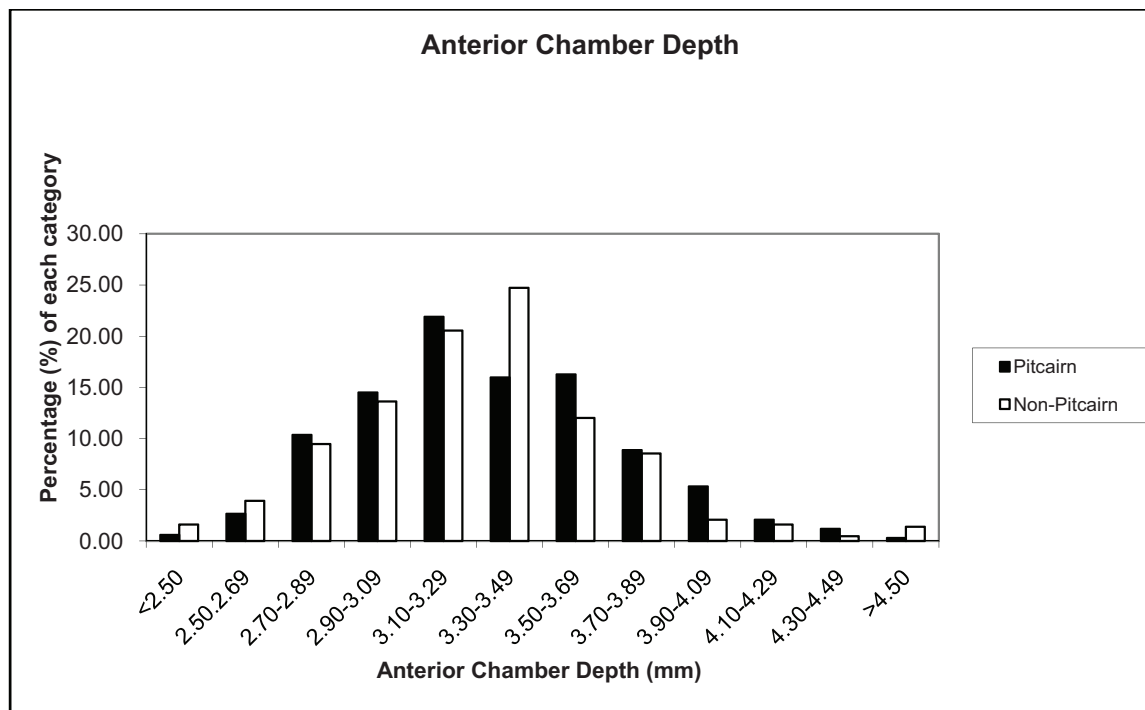
## Discussion

The study recruited 61% of the population age over 15 years, although recruitment of 70% of those aged over 50 years is comparable to other population-based eye studies. The ethnic breakdown of NIES — 43.9% Pitcairn descent — matches 43.0% Pitcairn descent in the census. The cohort provides an opportunity for both genetic and non-genetic research with the data collected, as minimal ocular

health data are available for populations in the Pacific Islands (especially people of Polynesian descent). Although there had been a previous cardiovascular study, there was no prior population-based ocular survey of the island. Providing free comprehensive eye examinations in co-operation with the optometrist aided recruitment. The data obtained will be useful for planning of future eye services on Norfolk Island. Like many regional areas of Australia, there is a higher percentage of older participants as many younger islanders need to leave the Island for education and work.

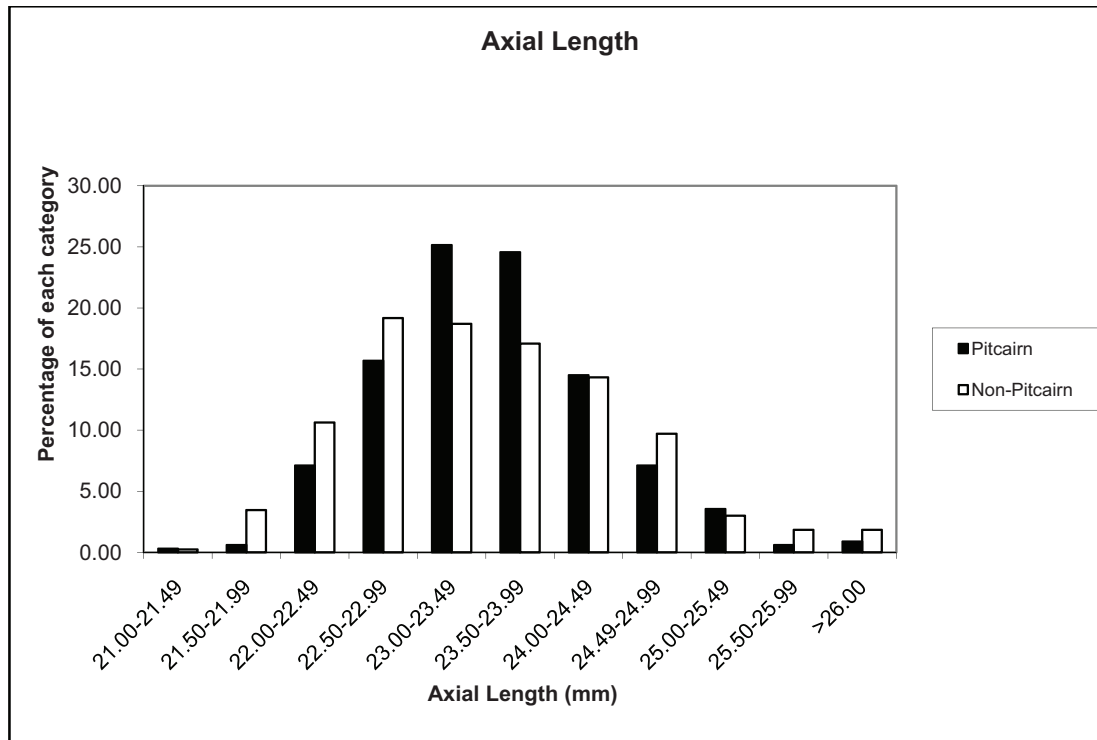
## Population Genetics

Previous research had shown that the 44% of islanders belonging to the Norfolk Island Pitcairn pedigree were genetically 88% European and 12% Polynesian (McEvoy et al., 2009). The majority of the non-Pitcairn population are from either Australia or New Zealand (White Caucasian) although some other Melanesian, Polynesian or Chinese residents live on the island. The Norfolk Island Census (2006) indicated country of birth of permanent population as Australian 32.6%, New Zealand 22.9%, United Kingdom 2.6%, or other 4.8%. In total, 62.9% were not born on Norfolk Island but this includes islanders whose parents went to Australia or New Zealand for antenatal care. The current citizenship of permanent population is: AUS 81.9%, NZ 15.0%, UK 0.8%, other 1.3%.

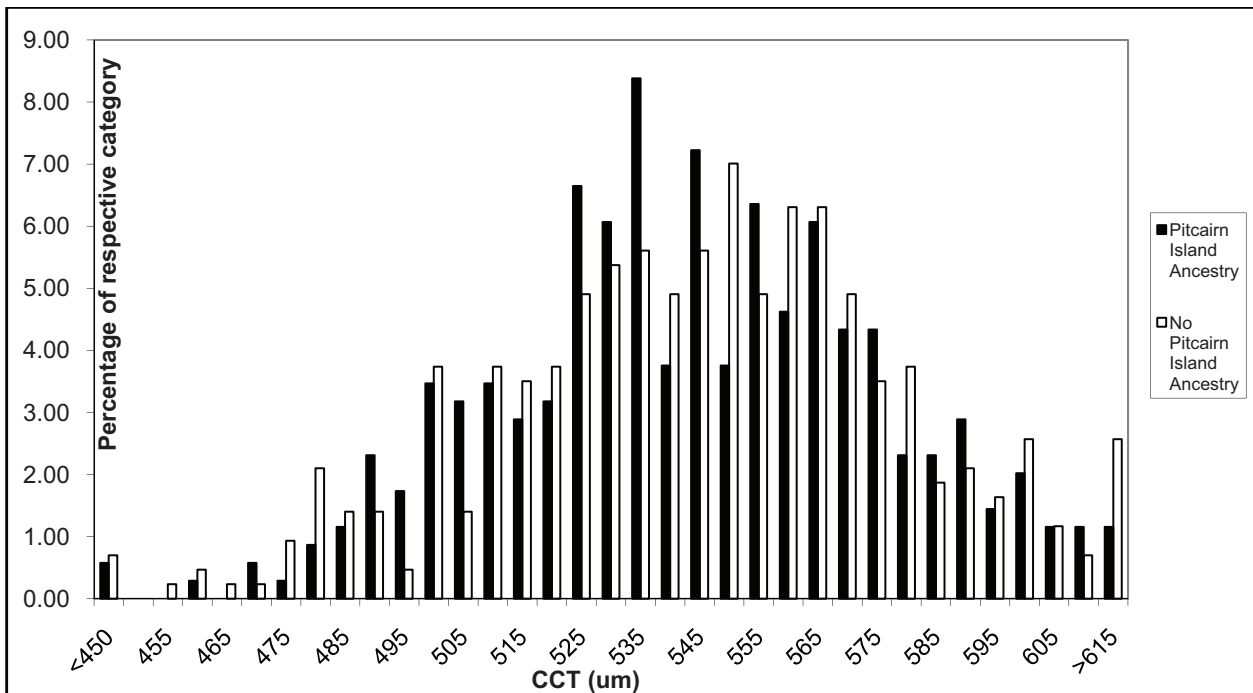


**FIGURE 2**

Distribution of anterior chamber depth according to Pitcairn Island ancestry.

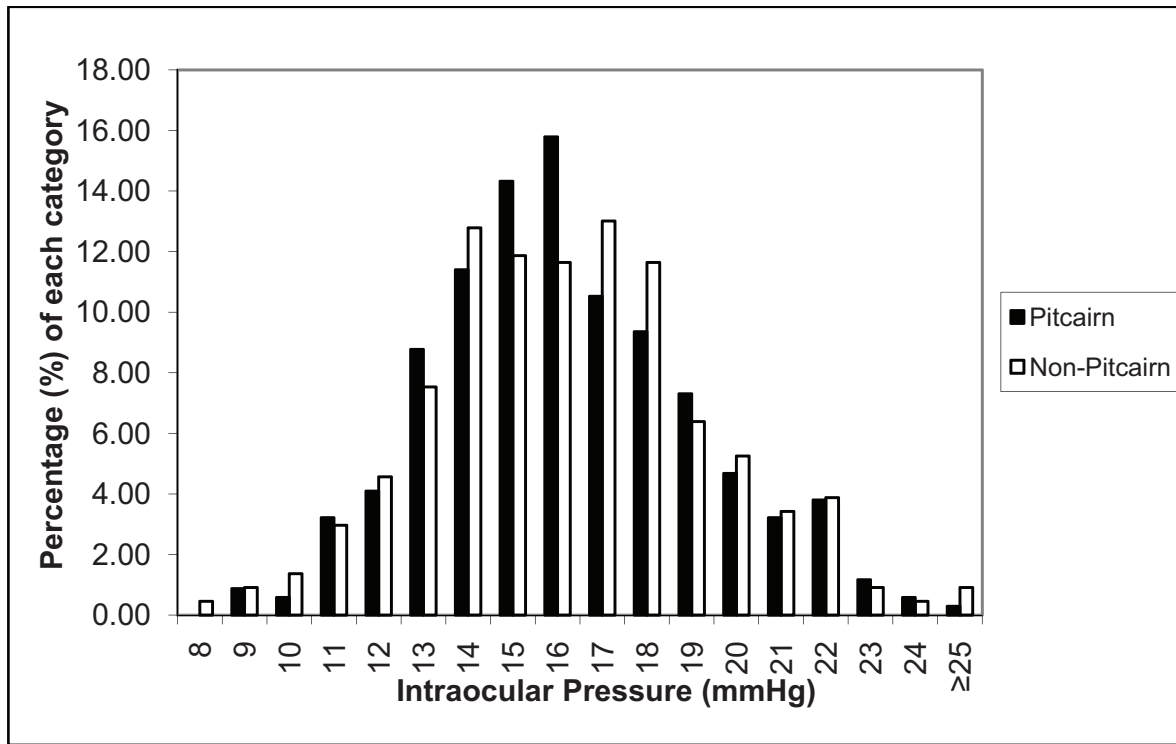


**FIGURE 3**  
Distribution of axial length according to Pitcairn Island ancestry.

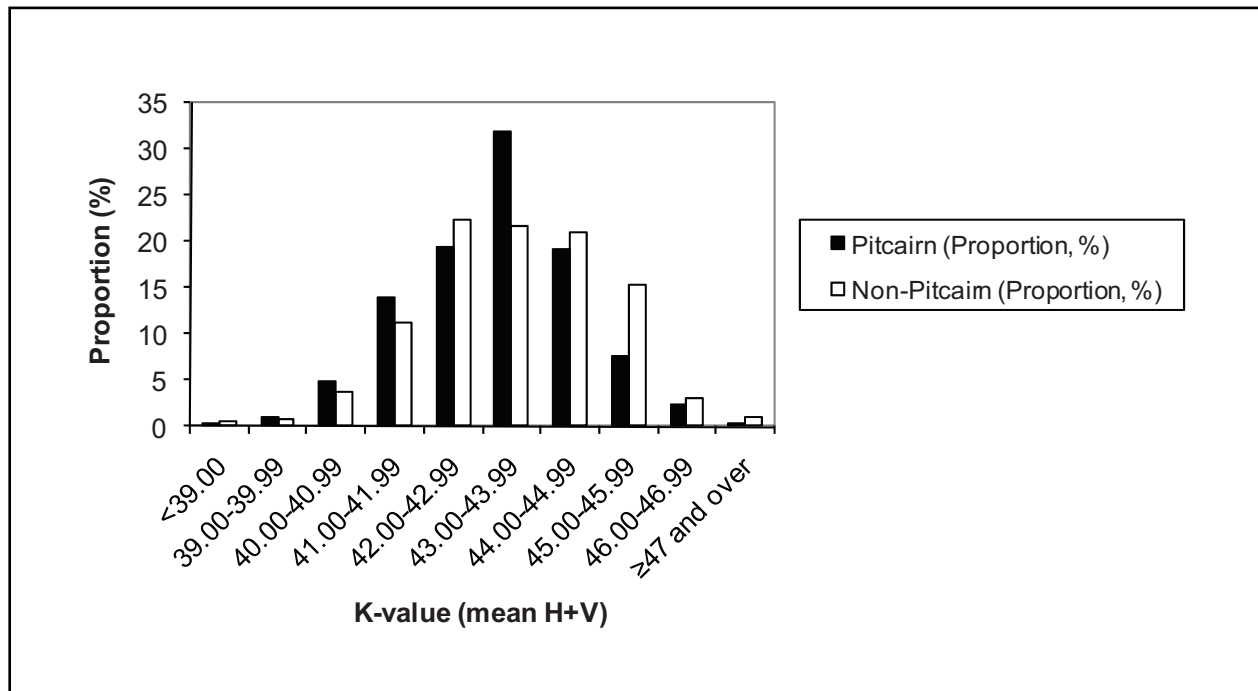


**FIGURE 4**  
Distribution of central corneal thickness according to Pitcairn Island ancestry: In these graphs, each category PIT or Non-PIT = 100%. This table has 5 um intervals.





**FIGURE 5**  
Distribution of intraocular pressure according to Pitcairn Island ancestry.



**FIGURE 6**  
Distribution of mean (H/V R/L) keratometry in dioptres according to Pitcairn Island ancestry.

## Ocular Biometry

Of the five main biometric measures in this study: ACD, AL, CCT, IOP, and keratometry readings, we found significant differences in the IOP and keratometry. Refractive measures and ophthalmic diseases are discussed in other papers (Sherwin submitted). There is racial variation in virtually all chronic ocular diseases. Having 12% Polynesian ancestry in addition to consanguinity may influence the distribution of biometry and ocular diseases in Pitcairn Islander descendants. Racial variation in the prevalence of glaucoma (Rudnicka et al., 2006), as well as in glaucoma-related ocular biometry (Shimmyo et al., 2003), is well described.

The mean ACD of 3.32mm was similar to that reported from the Blue Mountains Eye Study (3.10mm), which also utilised an IOL Master (Fotadar et al., 2010). Mean IOP was 16.23mm Hg; this was slightly higher than the mean IOP of 15.1mmHg in non-glaucoma subjects in the Melbourne Visual Impairment Project (VIP) project in which IOP was also measured with the Tonopen (Le et al., 2003). The mean Keratometry (K) reading of K Horizontal 42.95D and K Vertical 43.82D was similar to the mean K 43.42 D of the BMES (Fotadar et al., 2010).

Ocular biometry suggested a lower IOP in those of Pitcairn descent but there was no difference in CCT. However, the distribution of CCT on the island did not seem to follow the normal distribution seen in other populations. Those of Pitcairn descent had flatter corneas but no corresponding differences in AL or refraction. The next phase of NIES work will be to study the genetic associations of these biometric measures. The Norfolk Island population offers an opportunity to investigate heritability and linkage analyses in a unique population with founder effects from English and Polynesian ancestors. Future genetic studies, both linkage and GWAS, may allow identification of genes that differ between the two ancestral populations. In addition, in depth analysis of UV photos, stereo disc photos, retinal photos and OCT will be used to determine genetic factors involved in conjunctival, optic nerve and retinal biometry.

## Acknowledgments

We thank the Norfolk Islanders for welcoming us into their community and for their participation and assistance in this study. Financial support was provided by the RVEEH research committee, the Peggy and Leslie Cranbourne Foundation, DAM is a recipient of the Pfizer Australia Senior Research Fellowship. CERA receives operational infrastructure support from the Victorian government.

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