23 SUMMARY

This is a detailed epidemiological study of the causes and patterns of childhood onset visual impairment in the WB and the GS that was conducted between October 1985 and September 1987. It was aimed primarily at establishing the size of the problem in the target population and it focused initially on blind schools pupils, those on the waiting lists, their relatives and patients referred to the author at the OPC. Later, all other cases on the SJOH register that fulfilled the study criteria and had not been covered in the survey were added. This totalled 699 patients, 347 of whom came from the WB and 289 from the GS. All patients were fully evaluated and examined by the author with the exception of 85 cases who had been examined by various members of the staff and where data were extracted from SJOH casenotes. These patients belonged to 391 pedigrees comprising 458 sibships.

Sixty two percent of patients in the total series had SVI/BL (WB 66.6, GS 56.4, total 61.7). The disparity between the two regions was more marked in the <16 years (WB 60%, GS 47%). However, the prevalence in children was identical in both regions at 28/100,000.

A male preponderance was observed in the total series (1.44:1), particularly in the GS (1.6:1), which diminishes in school age children and in the hereditary conditions to become comparable to that of the general population. In the non-hereditary cases male preponderance was noted in both region (WB 2:1, GS 1.5:1). It was caused in the former by very high male predominance in the acquired optic nerve disorders. Gender differences were also demonstrated in certain conditions with male preponderance found in CC, CG, small eyes and optic nerve disorders and female preponderance in achromatopsia.

The rarity of blind children not in full-time education and the absence of male preponderance in the school age cohort, reflect a culture of valuing education and absence of male favouring education, a problem that has been

encountered in many developing countries. In addition, it shows an effective machinery of recruiting children from remote areas by social services and charitable organisations. Obviously, compulsory schooling for children must be an important factor.

The bulk of the conditions (77%) were hereditary, being higher in the GS (WB 76.4%, GS 84.4%) as a consequence of the high rate of consanguinity; reaching as high as 88.6% in the hereditary conditions (WB 82%, GS 90%). First cousin marriages were the most favoured accounting for up to 70% of the total consanguineous marriages. Such high consanguinity is echoed in the high predominance of the AR mode of inheritance which made up to 89% of the genetically inherited conditions, a figure much higher than in any other country.

The commonest conditions were retinal (47.1%) with retinal dystrophies forming 78% of cases, lens conditions (21.7%), 89.5% of which were congenital cataract/aphakia, congenital glaucoma (10.2%) and small eyes (5.5%). The latter were more predominant in the GS (WB 4.3%, GS 7.6%). Both CC and CG had high ocular and visual morbidity as a consequence of the intractable amblyopia from the neglect of aphakia in the former and a severe form of disease in the latter. The prevalence of these conditions per 100,000 population in the WB and GS was as follows: RD (18 and 23), CC (6.5 and 12) in the <16, CG (3.6 and 6.4) and small eyes (1 and 3.8). In the latter, the mean figure increases to near 9% when all small eyes below 10.5 mm corneal diameter are included. Congenital cataract was present in 41.6% of all the microphthalmic cases; uveal colobomas in 18.3%; and ACS together with Reiger's type of anomaly in 8.3% (section 19).

Wide regional differences existed in the prevalence of RD together with a wide inter and intra familial differences in their phenotypes. In the WB disorders of the rod system (LCA and childhood onset RCD) were the predominant conditions whereas cone dystrophies (cone dysfunction and cone and CD/CRD) were the hallmark of RD in the GS, frequently in symdromatic form.

Our findings have demonstrated similarities shared with the some of the Arab and Muslim countries ie a shift in the causation of blindness from infective to hereditary conditions, as was the case in industrialised countries

before the recent trend of perinatal conditions resulting from complications of prematurity. Care for premature babies had not been developed at the time of the study, hence the rarity of blindness secondary to prematurity.

Treatable blindness in this population is confined to CC as infections have been reduced in recent generations, at least as a cause of bilateral blindness and nutritional factors are non-existent. Consanguinity remains the main factor that needs addressing, especially as there has been a rise in this tendency, particularly in the GS, in the younger generation that has most likely been imposed by geopolitical factors.

Prevention of blindness in the OPT is not an easy task because of the cultural acceptance of consanguineous marriage and it requires long-term measures of health education and premarital carrier counselling by governmental, non-governmental and religious bodies. Recommendations were put forward in 1987 for a multidisciplinary prevention programme [705]. This focused on community education, together with improved primary health care, the need for tertiary paediatric services, and the establishment of a system for continued care and postoperative monitoring and orthoptic services.

The detailed genealogy also presented valuable material for the molecular biologist and led to several new findings including the detection of increased band sharing in DNA fingerprints in inbred populations; a point that should be considered in forensic or paternity cases involving members of inbred communities [741]. More recently a new locus was found on chromosome 2q11 at which recessive AI and CRD were found to cosegregate [768] (See appendices).

Additional findings were that red glasses were found to help in alleviating photophobia and discomfort, and that educational achievement was higher in certain conditions (achromatopsia, cone-rod dystrophy in contrast to rod-cone dystrophies).

It is greatly hoped that the new Palestinian Authority will give attention to the predicaments faced by this community which has long awaited peace and justice especially given the greater decline in health conditions and the increase in suffering since the end of this study.