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Summary of Results

This is a detailed epidemiological study of the causes and patterns of childhood onset visual impairment in the West Bank (WB) and the Gaza Strip (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 via the out-patients. 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 where data was extracted from the casenotes of patients examined by various members of the staff.

The total number of cases fulfilling the criteria of childhood onset visual impairment was 669; they belonged to 391 pedigrees comprising 458 families. Of these, 61.7 had SVI/BL (66.6% of the Wb cohort, 56.4% of GS cphort). The disparity between the two regions was more marked in the < 16 years (WB cohort 60%, GS cohort 47%). However, the prevalence of SVI/BL in children was identical in both Palestinian 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 as the conditions were predominantly recessive traits. In the non-hereditary cases male preponderance was noted in both the WB and GS (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 congenital cataract (CC), congenital glaucoma (CG), microphthalmos (MC), and optic nerve disorders with a female preponderance in achromatopsia.

The bulk of the conditions in the total series was hereditary (77%) and was higher in the GS at 84.4% than the WB at 76.4%. The consanguinity rate reached as high as 88.6% in the hereditary disorders, being 90% in the GS and 82% in the WB. 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 autosomal recessive (AR) mode of inheritance which made up to 89% of the genetically inherited conditions, a figure 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 CC/aphakia; CG (10.2%) and small eyes (5.5%). The latter were more predominant in the GS (7.6%) in comparison to the WB (4.3%). 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 commonest retinal dystrophies were Lebers congenital amaurosis (LCA), childhood onset rod-cone dystrophies (RCD), and cone disorders (achromatopsia and cone/cone rod dystrophies (CD/CRD). combined with inter and intra phenotypic variability The efficacy of red-filters in cone disorders was demonstrated, [(1)] together with the better educational performance in cone disorders and CG and the reverse in LCA. [(2)]

Wide regional differences existed in the prevalence of retinal dystrophies. In the WB disorders of the rod system (LCA) and RCD were predominant whereas cone disorders

(achromatopsia and CD/CRD were the hallmark of retinal dystrophies in the GS, frequently syndromatic.

The prevalence of the above conditions per 100,000 population in the WB and GS respectively was as follows: retinal dystrophies (18%; 23%), CC (6.5%; 12%) in the <16, CG (3.6%; 6.4%) and MC (1%; 3.8%). In the latter, the mean figure increases to near 9% when all small eyes below 10.5 mm corneal diameter are included. In the small eyes series, CG was present in 41.6%; uveal colobomas in 18.3%; and anterior cleavage syndrome with Reiger's type of anomaly in 8.3%.

The 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 in the WB and GS, hence the rarity of blindness secondary to prematurity.

Avoidable 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 (the author had encountered frequent cases of unilateral blindness caused by unilateral corneal infection and trauma). Consanguinity remains the main factor that needs to be addressed, 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 Occupied Palestinian Territories is not an easy task because of the cultural acceptance of cousin marriages and it requires long-term measures of health education and premarital carrier counselling by governmental, NGO and religious bodies. Recommendations were put forward in 1987 for a multidisciplinary prevention programme. **(3)** 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; [(4)] a point that should be considered in forensic or paternity cases involving members of inbred communities.

Several publications have arisen from this work and more recently the gene of one extended family with a syndromatic CRD has been found. [(2), (5), (6), (7), (8), (9) (10) (11), (12) (13) (14)]

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). **(1)**

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.

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