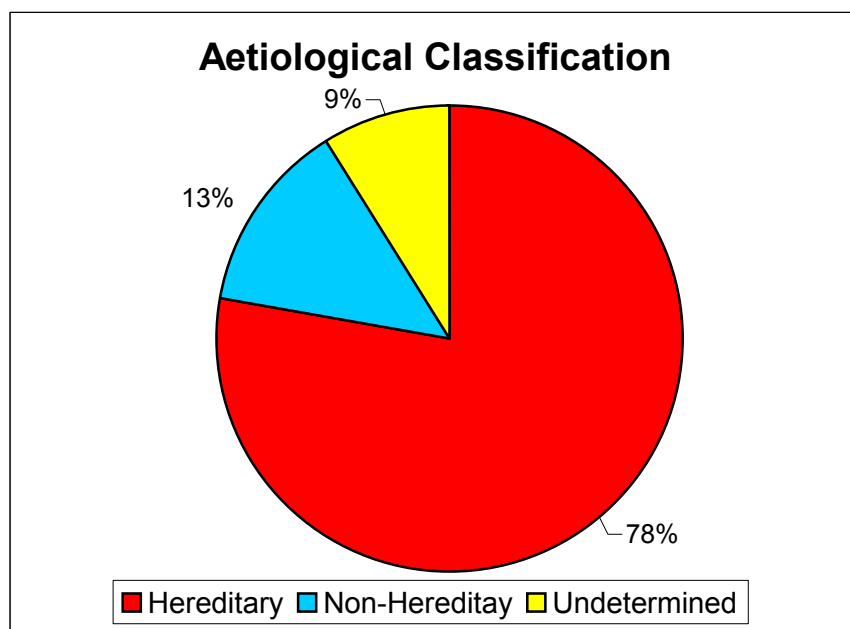


# 17 AETIOLOGICAL CLASSIFICATION AND HEREDITARY CONDITIONS

## 17.1 Aetiological Factors

In this study, among the total number of patients of 669 patients, the aetiologies of the causative condition were as follows:

1. Hereditary: This is the largest aetiological group comprising a total of 519 patients, 243 pedigrees and 310 sibships. (77.6%)
2. Non-Hereditary Conditions: This group comprises 90 patients and sibships with 89 pedigrees. (13.4%)
3. Aetiology Undetermined: This includes 60 patients with the same number of pedigrees and sibships. (9%)



**Figure 17- 1: Aetiological Subgroups in 669 patients**

Patients' distribution in the hereditary and non-hereditary conditions in the WB and GS are shown in Tables 17-1 with their gender highlighted in

Table 17-2. (Also refer to Table 22-6 for comparison with worldwide figures).

		West Bank		Gaza Strip		Others		Total	
Hereditary	<16	163	<b>76.2</b>	141	<b>86.0</b>	6	<b>22.2</b>	310	<b>76.4</b>
	16+	102	<b>76.7</b>	103	<b>82.4</b>	4	<b>66.7</b>	209	<b>79.5</b>
	<b>Total</b>	265	<b>76.4</b>	244	<b>84.4</b>	10	<b>33.3</b>	519	<b>77.6</b>
Non-Hered.	<16	26	<b>12.1</b>	9	<b>5.5</b>	5	<b>18.5</b>	40	<b>9.9</b>
	16+	29	<b>21.8</b>	20	<b>16.0</b>	1	<b>16.7</b>	50	<b>19.0</b>
	<b>Total</b>	55	<b>15.9</b>	29	<b>10.0</b>	6	<b>66.6</b>	90	<b>13.5</b>
UD	<16	25	<b>0.9</b>	14	<b>1.2</b>	16	<b>3.7</b>	55	<b>1.2</b>
	16+	2	<b>1.5</b>	2	<b>1.6</b>	1	<b>16.7</b>	5	<b>1.9</b>
	<b>Total UD</b>	27	<b>7.8</b>	16	<b>5.5</b>	17	<b>51.5</b>	60	<b>9.0</b>
<b>TOTAL</b>		347		289		33		669	
Total 16		214		164		27		405	
Total 16+		133		125		6		264	

Percentages (in bold italics) are of the total number of patients in the cohort

**17-1: Distribution of cases by aetiology by region**

			West Bank		Gaza Strip		Total	
			No.	M/F	No.	M/F	No.	M/F
Hereditary	All series	M	127	<b>0.9</b>	146	<b>1.5</b>	277	<b>1.1</b>
		F	138		98		242	
	Subtotal		265		244		519	
	School Children	M	78	<b>0.95</b>	73	<b>1.3</b>	152	<b>1.1</b>
F		82		55		140		

<b>Non-Hereditary</b>	All Series	M	25	<b>0.9</b>	19	<b>1.9</b>	49	<b>1.2</b>
		F	30		10		41	
	Subtotal		55		29		90	
	School Children	M	21	<b>2</b>	9	<b>4.5</b>	32	<b>2.5</b>
F		10		2		13		
TOTAL			347		289		669	

**Table 17-2: Gender in hereditary and non-Genetic cases**

## 17.2 Distribution of Aetiological Groups

In the WB, the percentages of both the hereditary and non-hereditary conditions in the northern regions (Jenin, Qalqilya, Tul-Karem and Nablus) are equal. There is preponderance of hereditary conditions in the central regions (Ramalla, Jerusalem, and Bethlehem) and of the non-hereditary conditions in the southern regions of Hebron (Table 17-3).

	North		Centre		South		UD		Total	
	No.	%	No.	%	No.	%	No.	%	No.	%
Non-hereditary	18	<b>32.7</b>	20	<b>36.4</b>	12	<b>21.8</b>	5	<b>9.1</b>	55	<b>100</b>
Hereditary	85	<b>32.1</b>	124	<b>46.8</b>	39	<b>14.7</b>	17	<b>6.4</b>	265	<b>100</b>
Total	103	<b>32.2</b>	144	<b>45.0</b>	51	<b>15.9</b>	22	<b>6.9</b>	320	<b>100</b>
% Non-Hereditary	<b>17.5</b>		<b>13.9</b>		<b>23.5</b>		<b>22.7</b>		<b>17.2</b>	
	least				highest					

**Table 17-3: Distribution of cases in the West Bank by aetiology by subregion**

There are wide variations in the proportions of hereditary and non-hereditary conditions according to the type of locality (i.e urban, rural or camps). Over 50% of both the hereditary and the non-hereditary cases live in villages (Table 17-4).

	Villages		Towns		Camps		UD		Totals	
	No.	%	No.	%	No.	%	No.	%	No.	%
<b>West Bank</b>										
Non-hereditary	31	<b>56.4</b>	20	<b>36.4</b>	3	<b>5.5</b>	1	<b>1.8</b>	55	<b>100</b>

Hereditary	148	<b>55.8</b>	79	<b>29.8</b>	24	<b>9.1</b>	14	<b>5.3</b>	265	<b>100</b>
Subtotal	179	<b>55.9</b>	99	<b>30.9</b>	27	<b>8.4</b>	15	<b>4.7</b>	320	<b>100</b>
% non- hereditary		<b>17.3</b>		<b>20.2</b>		<b>11</b>		<b>6.7</b>		<b>17</b>
<b>Gaza Strip</b>										
Non-hereditary	6	<b>20.7</b>	7	<b>24.1</b>	15	<b>52</b>	1	<b>3.4</b>	29	<b>100</b>
Hereditary	73	<b>29.9</b>	95	<b>38.9</b>	66	<b>27</b>	10	<b>4.1</b>	244	<b>100</b>
Subtotal	79	<b>28.9</b>	102	<b>37.4</b>	81	<b>30</b>	11	<b>4</b>	273	<b>100</b>
% non-hereditary		<b>7.6</b>		<b>6.9</b>		<b>19</b>		<b>9.1</b>		<b>11</b>
<b>All OPT *</b>										
All aetiologies	269	<b>40.2</b>	226	<b>33.8</b>	111	<b>17</b>	63	<b>9.4</b>	669	<b>100</b>
% non-hereditary		<b>14.3</b>		<b>13.4</b>		<b>16.7</b>		<b>7.7</b>		<b>14.2</b>

\* Including others from undetermined regions. Figures in *italic bold* are percentage

**Table 17-4: Distribution of aetiological cohorts by dwelling**

### 17.3 Hereditary Factors

Heredity formed the major bulk of cases in this survey. Of the 669 patients with bilateral visual disorders, the aetiology in 519 is hereditary in origin. Of these, 265 patients (51%) are in the WB and 244 (47%) in the GS. A further 10 (2%) are patients whose families live in Israel or are from either the WB or the GS but where the exact origin is uncertain.

The M/F ratio for the total WB hereditary series (n= 163) is identical to the sex ratio in the general population at 1:1 in the <16. In the 16+ cohort the ratio 0.79:1 (n=102) - Table 17-2. The latter ratio resulted from a female excess of 12 patients. In the GS, the ratio in the <16 is 1.39:1 (n=141) and in the 16+, 1.64:1 (n=103). This is the result of an excess of 23 and 25 male patients in each cohort respectively. The overall ratio, however, in the total hereditary group is 1.14:1 combining the <16 at 1.17:1 and the 16+ at 1.1, similar to the ratio in the general population.

Aetiological Groups	Pedigrees			Sibships		
	WB	GS	Total <sup>a</sup>	WB	GS	Total <sup>a</sup>
Hereditary	148 <b>64.3</b>	86 <b>65.6</b>	239 <b>65.3</b>	175 <b>68.1</b>	126 <b>73.7</b>	306 <b>70.7</b>
Acquired	55 <b>23.9</b>	29 <b>22.1</b>	84 <b>23</b>	55 <b>21.4</b>	29 <b>10</b>	84 <b>19.4</b>
UD	27 <b>11.7</b>	16 <b>12.2</b>	43 <b>11.7</b>	27 <b>10.5</b>	16 <b>9.4</b>	43 <b>9.9</b>
Total	230 <b>100</b>	131 <b>100</b>	366 <b>100</b>	257 <b>100</b>	171 <b>100</b>	433 <b>100</b>
% Hereditary	% <b>73</b>	<b>75</b>	<b>74</b>	<b>76</b>	<b>81</b>	<b>78</b>

<sup>a</sup> Includes additional 5 pedigrees and sibships and 2 patients from either region

## 17.4 Hereditary Conditions

As hereditary conditions form 77.6% of the total series, analysing the data will follow closely the findings demonstrated in the total series addressed previously. Apart from 2 cases which are most probably chromosomal, hereditary conditions are genetic in aetiology.

### 17.4.1 Hereditary Retinal Conditions

Hereditary retinal conditions constitute 97% of the retina cases with the commonest conditions being retinal dystrophies, albinism and high myopia 4.8% (n=22) (Table 17-8). In the GS, this was as high as 99% of all the retinal conditions (Tables 17-8, 17-9). There is an obvious male preponderance in the GS (ratio 1.5:1) with the reverse observed in the WB (ratio 0.7:1), averaging out in both regions at 1:1.

Sites	Clinical Conditions	<16		16+		Total	
Retina	Retinal Dystrophies	147	<b>46.1</b>	97	<b>48.5</b>	244*	<b>47.0</b>
	Albinism	21	<b>6.6</b>	6	<b>3.0</b>	27	<b>5.2</b>
	Degenerative myopia	12	<b>3.8</b>	12	<b>6.0</b>	24	<b>4.6</b>
	Others	5	<b>1.6</b>	4	<b>2.0</b>	9	<b>1.7</b>
	Subtotal	<b>185</b>	<b>58</b>	<b>119</b>	<b>59.5</b>	<b>304</b>	<b>58.6</b>
Lens	Congenital cataract	46	<b>14.4</b>	35	<b>17.5</b>	81	<b>15.6</b>
	Ectopia lentis	11	<b>3.4</b>	2	<b>1.0</b>	13	<b>2.5</b>
	Subtotal	<b>57</b>	<b>17.9</b>	<b>37</b>	<b>18.5</b>	<b>94</b>	<b>18.1</b>
Whole Globe	Congenital Glaucoma	39	<b>12.2</b>	18	<b>9.0</b>	57	<b>11.0</b>
	Micro/anophthalmos	20	<b>6.3</b>	13	<b>6.5</b>	33	<b>6.4</b>
	Others	0	<b>0.0</b>	3	<b>1.5</b>	3	<b>0.6</b>
	Subtotal	<b>59</b>	<b>18.5</b>	<b>34</b>	<b>17.0</b>	<b>93</b>	<b>17.9</b>
Others Sites	Unspecified	18	<b>5.6</b>	10	<b>5.0</b>	28	<b>5.4</b>

### 17.4.3

Hereditary retinal conditions constitute 97% of the retina cases with the commonest conditions being retinal dystrophies, albinism (5.2%, n=27)

	West Bank				Gaza Strip				Total Series			
	M	F	Total	M/F	M	F	Total	M/F	M	F	Total <sup>a</sup>	M/F
All Retina	72	95	167	0.8:1	84	58	142	1.4:1	156	153	317	1:1
Genetic	68	92	160	0.7:1	83	57	140	1.5:1	151	149	306	1:1
% Genetic	<b>94</b>	<b>97</b>	<b>96</b>		<b>99</b>	<b>98</b>	<b>99</b>		<b>97</b>	<b>97</b>	<b>97</b>	

<sup>a</sup> Including others mainly from either region      Percentages are in italic

**Table 17-8: Patients' numbers, percentages and gender in the genetic retinal series**

	Pedigrees						Sibships					
	WB		GS		Total		WB		GS		Total	
All Retina	96	<b>91</b>	44	<b>42</b>	147	<b>100</b>	113	<b>60</b>	68	<b>36</b>	188	<b>100</b>
Genetic Retina	89	<b>84</b>	42	<b>40</b>	136	<b>100</b>	106	<b>60</b>	66	<b>37</b>	177	<b>100</b>
% Genetic	<b>93</b>		<b>95</b>		<b>93</b>		<b>94</b>		<b>97</b>		<b>94</b>	
S'ship/Pedig <sup>a</sup>	<b>1.18</b>		<b>1.55</b>		<b>1.28</b>							

Percentages are in italic

<sup>a</sup>: ratio of sibships/pedigrees

**Table 17-9: Pedigrees and sibships in the genetic retinal series**

An account of the clinical aspects of the common hereditary retinal conditions namely; retinal dystrophies, albinism and degenerative myopia is given in section 16. Table 17-10 compares the distribution of patients, their gender, the important genetic retinal conditions and Table 17-11 demonstrates the pedigrees and sibships in these conditions.

	West Bank				Gaza Strip				Total Series			
	M	F	Total	M/F	M	F	Total	M/F	M	F	Total	M/F
Dystrophies	60	79	139	0.8	55	46	101	1.2	115	125	245	0.9
Albinism	2	6	8	0.3	7	12	19	0.6	9	18	27	0.5
Myopia	4	4	8	1.0	13	4	17	3.3	17	8	25	2.1
Retinoblastoma	0	1	1	-	0	0	0	-	0	2	2	-
Others	2	0	2	-	2	1	3	2.0	4	1	5	4.0
Total	68	90	158	0.8	77	63	140	1.2	145	154	304	0.9

**Table 17-10: Gender in the common genetic retinal conditions**

Retinal Conditions	Pedigrees						Sibships					
	WB		GS		Total OPT		WB		GS		Total OPT	
Dystrophies	77	<b>87</b>	25	<b>60</b>	106	<b>78</b>	94	<b>89</b>	46	<b>70</b>	144	<b>81</b>
Albinism	3	<b>3</b>	7	<b>17</b>	10	<b>7</b>	3	<b>3</b>	7	<b>11</b>	10	<b>6</b>
Myopia	5	<b>6</b>	8	<b>19</b>	13	<b>10</b>	5	<b>5</b>	10	<b>15</b>	15	<b>8</b>
Retinoblastoma	1	<b>1</b>	0	<b>0</b>	2	<b>1</b>	1	<b>1</b>	0	<b>0</b>	2	<b>1</b>
Others	3	<b>3</b>	2	<b>5</b>	5	<b>4</b>	3	<b>3</b>	3	<b>5</b>	6	<b>3</b>
Total	89	<b>100</b>	42	<b>100</b>	136	<b>100</b>	106	<b>100</b>	66	<b>100</b>	177	<b>100</b>

Percentages are in bold

R'blastoma: Retinoblastoma

**Table 17-11: Pedigrees and sibships in genetic retinal conditions**

### 17.4.2 Hereditary Lens Disorders

The second commonest genetic group is lens disorders with 94 cases forming 18.1% of the total genetic cohort. This leaves 14 (9.6%) cases of non-hereditary aetiology and 37 (25.5%) cases of undetermined aetiology.

Excluding the latter, hereditary conditions form 87% of the lens cases. The hereditary group comprises primarily genetic congenital cataract with 81 patients (86%) followed by ectopia lentis with 13 patients (14%). An account of the clinical aspects of this group is found in section 16.1.

	All Types		Hereditary		Non-hereditary		UD	
<b>All WHO Lens Categories (WHO categories L14, 15, 16)</b>								
All ages	145	<b>21.7</b>	94	<b>65</b>	14	<b>9.6</b>	37	<b>25.5</b>
<16	105	<b>26</b>	57	<b>54.3</b>	11	<b>10.5</b>	37	<b>35</b>
<b>Cong. Cataract (WHO Category L14, including aphakia L15)</b>								
All ages	132	<b>19.7</b>	81	<b>61.4</b>	14	<b>10.6</b>	37	<b>28</b>
<16	94	<b>23.2</b>	46	<b>49</b>	12	<b>12.7</b>	37	<b>39.4</b>
<b>All Cataract (primary and secondary pathology)</b>								
All ages	153	<b>22.8</b>	100	<b>63.4</b>	15	<b>9.8</b>	38	<b>34</b>
<16	111	<b>27.4</b>	61	<b>55</b>	13	<b>11.7</b>	38	<b>34</b>

**Table 17-12: Proportions of lens conditions and congenital cataract**

### 17.4.3 Other Genetic Conditions

These include congenital glaucoma (section 16.2), small eyes (microphthalmos and anophthalmos (section 16.3), and congenital hereditary corneal oedema (section 16.4).

