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Guidelines for the Management of Hereditary Retinal Dystrophies.

It has come to the attention of the Academic Advisory Committee of the South African Vitreoretinal Society that there is a problem with the current (1999) management guidelines for hereditary retinal dystrophy, ICD10 code H 35.5. Medical Aids have been unable to provide funding for this PMB condition as the level of care stated in the 1999 guidelines for treatment for condition PMB 904B (H35.5) is " vitrectomy, laser treatment and other surgery". This is clearly incorrect. This condition has been erroneously grouped with other surgical conditions. The condition Hereditary choroidal dystrophy (H31.2) would also fall within this guideline but is inexplicably omitted from PMB 904B. It is also important to note that the Medical Schemes Act states that the minimum standard of care for a condition would be the treatment provided by a state healthcare facility. These management guidelines below would fulfill those criteria.

The patient who presents with a condition which is suspected to be an hereditary retinal dystrophy/degenerative retinal condition (code H31.2, H 35.5) would require the diagnosis to be made by an Ophthalmologist. The Ophthalmologist would perform the following:

- Consult the patient and, following history-taking, perform colour vision testing and biomicroscopic examination.

- The patient would require a visual field.
- Imaging would be required with colour fundus photographs, Autofluorescence imaging, Infrared imaging, Optical Coherence Tomography (OCT) and fluorescein angiography.¹
- Further essential special investigations are an Electroretinography and Electro-oculography. Dark adaptometry would be helpful in certain conditions where available.²

Many retinal dystrophies are difficult to diagnose as the phenotypic appearance may change with time, so the clinical diagnosis may be possible only after repeated examinations and investigations. It is therefore necessary to repeat these investigations initially on a yearly basis until the diagnosis is established. Thereafter the investigations are required where it is necessary to ascertain whether treatment is indicated or to assess the speed of onset of loss of vision in order to guide the patient with respect to visual rehabilitation and occupation related issues.³

Once the diagnosis of a retinal dystrophy is suspected, it is then necessary to involve a registered genetic counsellor. The counsellor would consult the patient and advise the appropriateness of genetic testing, the benefits and limitations of testing and the testing strategy.⁴ I attach a sample of the genetic counselling costs and the costs of genetic testing. (Appendix 1 and 2)

The further management of an hereditary retinal dystrophy will entail the following:

- Completion of the genetic profile may require examination and investigation of the relatives.
- Treatment of the condition will entail oral supplements if they are indicated.
- Cataract, usually of the posterior subcapsular type, presents earlier in this group of conditions and is visually disabling despite the patient's reduced potential visual acuity and requires cataract surgery and intraocular lens implantation.

- As cystoid macular oedema is often a component of these conditions a trial of oral or topical acetazolamide and a topical non-steroidal anti-inflammatory eg Acular, Nevanac would be indicated.⁵
- Another complication that can occur with this group of conditions is the formation of choroidal neovascular membranes. The first line of treatment would be the use of intravitreal injections of anti-VEGF agents. (Please refer to the SAVRS Guidelines for the Management of Exudative Age-related Macular Degeneration for this management).
- The patients would need refraction and low vision management. Low vision management includes access to visual aids eg optical aids, computerized visual aids.
- Where there is advanced deterioration in vision, referral for mobility training will be required.
- Patients can also benefit from registration with Retina South Africa.
- Annual follow-up is required.

References:

1. A Longitudinal Study of Stargardt Disease. Fujinami K et al. Invest Ophthalmol Vis Sci. 2013; 54:8181-8190.
2. Clinical Phenotypes and Prognostic Full-Field Electroretinographic Findings in Stargardt Disease. Zahid S et al. Am J Ophthalmol 2013; Mar,155(3):465-473.
3. Correlations between OCT and VA. Kim Y et al. Invest Ophthalmol Vis Sci. 2013 Feb1;54(2):1303-9.
4. Recommendations for Genetic Testing of Inherited Eye Diseases. Stone EM et al. Ophthalmology 2012; 119:2408-2410.
5. Correlations between Spectral-Domain OCT measurements and Visual Acuity in Cystoid Macular Edema Associated with Retinitis Pigmentosa. Kim YJ. Invest Ophthalmol Vis Sci. 2013;54:1303-1309.

Appendix 1

Mrs Frieda Loubser

BSc Hons Genetics (Stell); MSc Med Genetic Counselling (UCT)

HPCSA Registered Genetic Counsellor

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Genetic Counselling for Retinal Degenerative Disorders:

PRE-TEST GENETIC COUNSELLING:

Detailed family history taking and inheritance pattern identification

Information giving:

- Genetics of Retinal Degenerative Disorders
- Appropriateness of genetic testing
- Benefits, risks and limitations of testing
- Testing strategy

POST-TEST COUNSELLING

Delivery of results:

Identification of known pathogenic mutation/s

or

No mutations identified

or

Inconclusive results

or

Variant of uncertain clinical significance

Implications of results for the individual and for family members (cascade testing)

Liaising with Ophthalmologist for further clinical management

GENETIC COUNSELLING COST:

First appointment (1 hour) : R600

Follow-up (30 min) : R300

Total anticipated cost : R 900

Genetic counselling

ICD10 code: Z31.5

procedure code: 069 (205 for 1 hour)

Appendix 2

Attention: Frieda Loubser

Herewith please find quotation:

AMPATH

Practice number: PR0520005200431

4755	X	1	R 3,709.40
4757	X	1	R 961.50
4751	X	1	R 618.20
4763	X	3	R 1,854.60
4760	X	2	R 3,159.80
4761	X	9	R 4,328.10
4765	X	6	R 4,946.40
4764	X	4	R 4,891.60
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			R 24,469.60

Regards

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