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The ROYAL COLLEGE of OPHTHALMOLOGISTS

COMMENT

Eponymous women in ophthalmology: syndromes with prominent eye manifestations named after female physicians

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Many eponymic syndromes were characterized when few women had entered medicine. Nearly simultaneous with the growth of women in ophthalmology has been a decreased emphasis on eponyms in favor of descriptive [1, 2] or biochemical [2–4] terminology. The decline in eponym use has been fueled by calls to eliminate the possessive form [2], concerns regarding inaccuracy as knowledge of disease pathogenesis has evolved [3, 4], worries that using different eponyms across languages stymies global scientific discussion [1], and the realization of inhumane activities by some individuals to whom eponyms were previously attributed [1, 3]. Nonetheless, it is of important historical relevance to identify and celebrate the women who distinguished themselves by helping to characterize syndromes associated with their names.

For this report, we culled print and electronic media for eponymous eye syndromes named after female ophthalmologists. Public records, obituaries, images, and other resources were compared to determine gender. Only syndromes listed in OMIM were reviewed to avoid inclusion of arcane eponyms. Syndromes in which the only ophthalmic-related features are craniofacial abnormalities, optic atrophy, and/or cortical blindness were excluded in order to focus inclusion on eye-related syndromes.

Two eponymous female ophthalmologists were identified: Ester Elisabeth Gröenblad and Mette Warburg. Gröenblad-Strandberg disease is pseudoxanthoma elasticum (PXE), characterized by ectopic mineralization and fragmentation of elastic fibers primarily in the eye, skin, and cardiovascular system. Ophthalmic findings include angioid streaks, *peau d'orange* fundus, and choroidal neovascularization. Gröenblad, a Swedish ophthalmologist, established the syndromic association of angioid streaks with the skin findings in collaboration with dermatologist James Strandberg.

Walker-Warburg syndrome causes brain abnormalities and retinal dysplasia. Neurosurgeon Arthur Earl Walker reported a case of hydrocephalus, lissencephaly, microphthalmos, and retinal dysplasia in 1942; additional authors subsequently presented cases. Warburg, a Danish ophthalmologist, suggested in 1971 that the findings represented a yet-uncharacterized syndrome. Today, the syndrome is referred to as muscular dystrophy-dystroglycanopathy with brain and eye anomalies, and genetic sequencing has led to subtype classification.

During our search for eponymous ophthalmologists, we identified several female non-ophthalmologist physicians who characterized syndromes with prominent eye manifestations (Table 1).

As is common among eponyms, the syndromes identified herein were not all named after the original discoverers, a phenomenon described as Stigler's *Law of Eponymy* [5]. The skin and eye findings of PXE had been described before Gröenblad and Strandberg established the syndromic association. Cases of Walker-Warburg syndrome were reported by Walker and others as many as 30 years prior to Warburg's papers, but eponymous credit is given to her due to her summative assessments of the syndrome. Warburg also described Norrie syndrome but named it in tribute after Gordon Norrie, a Danish ophthalmologist.

The plethora of historical eponyms leaves open the possibility that our report is incomplete. Additionally, binary gender categories may unintentionally exclude or mischaracterize some individuals.

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Table 1 Eponymous female non-ophthalmologist physicians

Name	Nationality and specialty	Syndrome name(s)	OMIM number	Description
Denise Louis-Bar	Belgian neurologist	Louis-Bar syndrome, Ataxia telangiectasia	#208900	Autosomal recessive disorder characterized by progressive cerebellar ataxia, telangiectasia, variable immunodeficiency, and predisposition to malignancy Eye findings: conjunctival telangiectasia
Gertrud Hurler	German pediatrician	Hurler syndrome, mucopolysaccharidoses type IH	#607014	One of several mucopolysaccharidoses with clinical features including coarse facies and mental retardation Eye findings: corneal clouding There is also an intermediate phenotype of the Hurler and Scheie syndromes, OMIM #607015
Cornelia de Lange	Dutch pediatrician	Cornelia de Lange syndrome Note: although many authors have continued to give de Lange eponymous credit due to the detailed nature of her original descriptions, some authors prefer Brachmann-de Lange syndrome, a reference to the original 1916 report	#122470 Cornelia de Lange syndrome 1; #300590 Cornelia de Lange syndrome 2; #610759 Cornelia de Lange syndrome 3; #614701 Cornelia de Lange syndrome 4; #300882 Cornelia de Lange syndrome 5	Widely variable multisystem syndrome with characteristic facial dysmorphism together with prenatal and postnatal growth retardation and mental retardation Eye findings: synophrys, long lashes, hypertrichosis of the brows, ptosis, epiphora, nasolacrimal duct obstruction, blepharitis, myopia
Anne-Marie de Barsy	Belgian neurologist	de Barsy syndrome	#219150 autosomal recessive cutis laxa, type IIIA (de Barsy syndrome A); #614438 autosomal recessive cutis laxa, type IIIB (de Barsy syndrome B)	Autosomal recessive disorder with progeria-like appearance, cutis laxa, and distinctive facial features Eye findings: corneal opacification, cataracts, blue sclerae, myopia, strabismus
Margaret B. Horan	Australian pediatrician	Nance-Horan syndrome, cataracts-oto-dental syndrome, X-linked cataract-dental syndrome, X-linked congenital cataracts and microcornea	#302350	X-linked disorder characterized by severe congenital cataracts, microcornea, and numerous dental, craniofacial, and other abnormalities
Aagot Christie Loken	Norwegian neuropathologist	Senior-Loken syndrome, renal-retinal syndrome, juvenile nephronophthisis with Leber amaurosis, renal dysplasia and retinal aplasia	#266900 Senior-Loken syndrome 1; %606995 Senior-Loken syndrome 3; #606996 Senior-Loken syndrome 4; #609254 Senior-Loken syndrome 5; #610189 Senior-Loken syndrome 6; #613615 Senior-Loken syndrome 7; #616307 Senior-Loken syndrome 8	Autosomal recessive disease with renal failure in childhood or adolescence (nephronophthisis) and Leber congenital amaurosis
Jacqueline Anne Noonan	American pediatric cardiologist	Noonan syndrome, male Turner syndrome, female pseudo-Turner syndrome	#163950 Noonan syndrome 1; 605275 Noonan syndrome 2 (autosomal recessive); #609942 Noonan syndrome 3; #610733 Noonan syndrome 4; #611553 Noonan syndrome 5; #613224 Noonan syndrome 6; #613706 Noonan syndrome 7; #615355 Noonan syndrome 8; #616559 Noonan syndrome 9; #616564 Noonan syndrome 10; several additional Noonan-like and hybrid conditions have been described	Autosomal dominant disorder with short stature, facial dysmorphism, and a wide spectrum of congenital heart defects; a variety of eyelid, optic nerve, and fundus findings are described
Pirkko Santavuori	Finnish pediatric neurologist	Santavuori-Haltia syndrome, Santavuori syndrome, neuronal ceroid lipofuscinosis 1	#256730	One of many neuronal ceroid lipofuscinoses; characterized by vision loss, regression of motor development, loss of speech, and ataxia

To our knowledge, this is the first report of syndromes with prominent eye manifestations named after female physicians. Eponyms embed history and tradition into our shared medical language, and identifying eponymous women has historical relevance and celebrates their achievements.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

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