Letter: Cataract development in Norwegian patients with congenital aniridia

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Original Publication:

Ulla Edén, Neil Lagali, Anette Dellby, Tor P. Utheim, Ruth Riise, Xiangjun Chen and Per Fagerholm, Letter: Cataract development in Norwegian patients with congenital aniridia, 2014, Acta Ophthalmologica, (92), 2, E165-E167.
http://dx.doi.org/10.1111/aos.12225

Acta Ophthalmologica

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Journal:	*Acta Ophthalmologica*
Manuscript ID:	ACTA-13-02-0195.R1
Wiley - Manuscript type:	Letter to Editor
Date Submitted by the Author:	07-May-2013
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Keywords:	aniridia, congenital aniridia, cataract development, congenital cataract

Cataract Development in Norwegian patients with Congenital Aniridia

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Editor,

Congenital Aniridia is caused by mutation of the PAX 6 gene, the so-called 'master gene' in ocular development. Although cataract has been reported in several aniridia cohorts (Nelson et al.1984; Sale et al.2002; Hingorani & Moore 2008; Abouzeid et al. 2009; Park et al. 2010; He et al. 2012), the timing and detailed phenotype of cataract in aniridia has not been well described. Here we report the onset of cataract, timing of cataract surgery and phenotypic features of cataract in a Norwegian aniridia cohort.

A cohort of 26 Norwegian patients (52 eyes) with congenital aniridia was examined on a single occasion, after obtaining written informed consent and ethical approval from the Regional Committee for Medical and Health Research Ethics, Oslo. Medical records were examined to detail cataract presence and surgical intervention. Digital slit lamp photographs of the lens and visual assessment were used to analyze the type and development of cataract, and aniridia associated keratopathy (AAK) was characterized according to our previously published grading scale (Edén et al. 2012).

Mean patient age was 29y (range: 4 – 63y). Only three eyes were phakic with clear lenses; the remaining eyes had either cataract or had been operated on for cataract. The youngest individual with cataract was four years old at examination, but congenital or early onset cataract was documented in medical records of 5 patients (6 eyes). Of 12 patients with non-operated cataract, 5 (9 eyes) had lens luxation upwards. 14 patients (27 eyes) had glaucoma. Those least affected presented with a discrete posterior polar cataract. In other cases a discrete sub-capsular opacification of varying density or opacification extending radially from the mid-periphery of the posterior capsule was found in addition to the polar cataract. A posterior sub-capsular mid-peripheral ring of opacification was observed, in some also combined with a more substantial polar cataract. The size and density of the opaque ring varied (Figure 1). Findings in other patients included: nuclear cataract (both turbid and yellow-brownish), one generalized subcapsular edema (mature cataract), and one with a dehydrated opaque lens (hypermature cataract) (Figure 1). In two patients, an anterior polar cataract was identified one of which had an additional posterior subcapsular opacification (Figure 1).

Of the 52 eyes examined, 25 had had surgical intervention (cataract, glaucoma or both). Nine patients (13 eyes) had cataract surgery only, 6 patients (8 eyes) both cataract and glaucoma surgery and 3 patients (4 eyes) glaucoma surgery only. Age at the time of cataract surgery is given in Figure 2. Secondary cataract was observed in 4 patients (6 eyes).Of the 25 eyes with surgical intervention, 8 eyes (30%) had AAK affecting visual acuity compared to 8/25 eyes (32%) in the group of eyes without intraocular surgery. No clear trend could be found towards an increased prevalence of AAK in operated eyes.

Cataract is common in aniridia, with over 90% prevalence in our cohort, similar to a Korean cohort with 60 eyes where 88% had cataract or were operated for cataract (Park et al. 2010). Cataract prevalence in aniridia in the literature varies from 50-85 % (Nelson et al.1984).

Patients in our cohort not operated for cataract showed a distribution of lens opacities that could be interpreted as a pattern of cataract development. A discrete posterior polar opacity seems to emerge first. The posterior location of polar opacities has been reported previously (Yamn et al.2011; Jin et al. 2012). The next phase is an additional subcapsular opacification in the midperiphery. These opacities then increase in density and size, radiate to the polar region and are always limited to the posterior subcapsular region. They eventually form a ring on the posterior capsule.

Acknowledgements
This study was supported by David and Beth Dahlins Foundation, Ögonfonden Sweden, Crown Princess Margareta's Foundation for the Visually Impaired, Carmen and Bertil Regnérs Foundation for Research in Eye Disease, the County Council of Östergötland, and Aniridia Norway.

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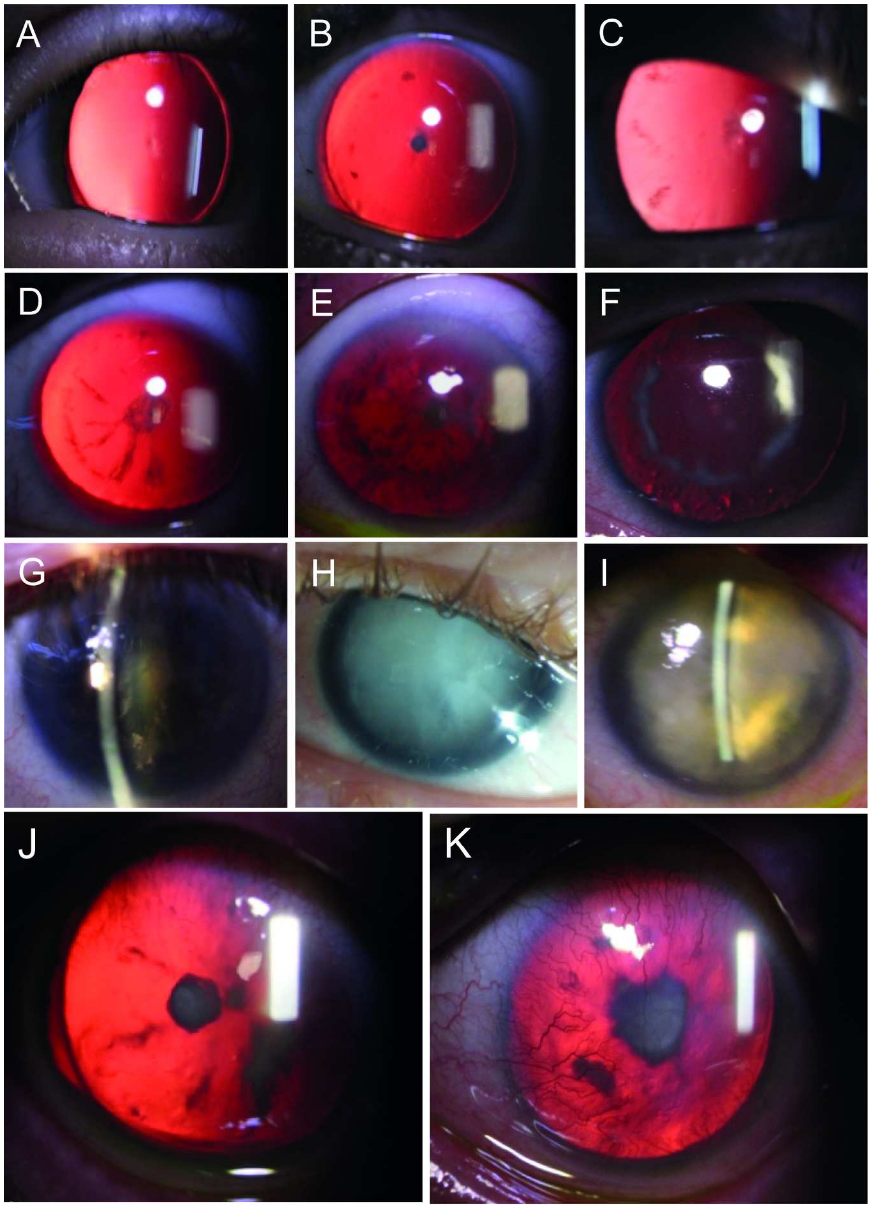
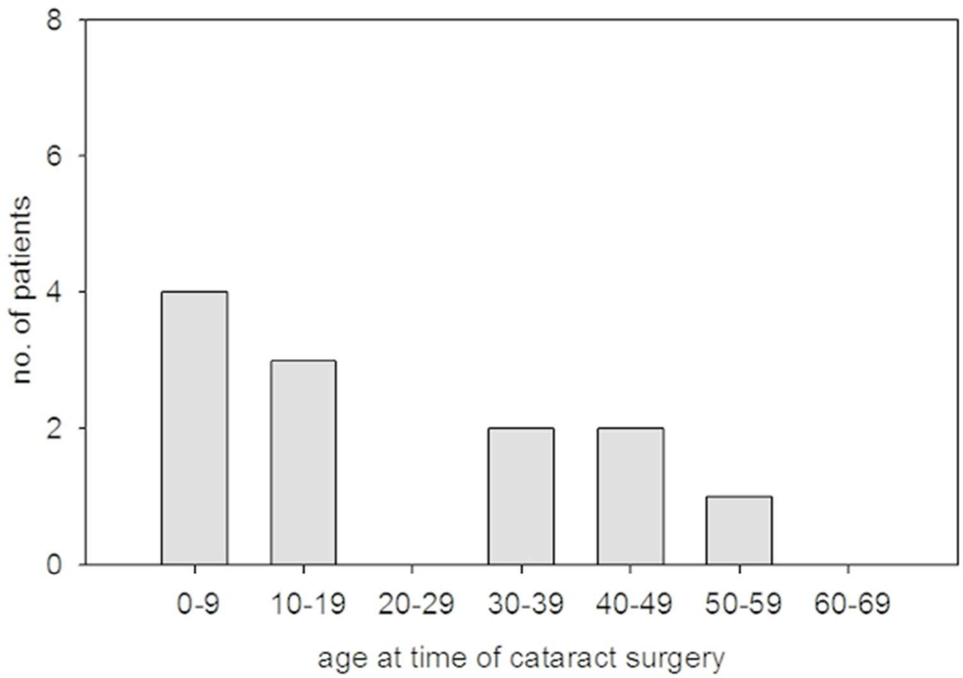
Figure Captions

Figure 1. Developmental patterns of cataract in aniridia patients. (A) Early-stage discrete posterior subcapsular cataract. (B-E) The posterior subcapsular cataract is denser. A posterior subcapsular opacification develops on the capsule in the midperiphery as tiny flecks (B), or short, radially-oriented opacities (C). (D-F) Radial opacities extend towards to posterior pole and a mid-peripheral ring of opacification of varying density is present. Opacification always remained subcapsular. Late lens changes included yellow and turbid nuclear cataract (G), mature cataract (H) and hypermature cataract (I). (J) Anterior polar cataract in combination with a posterior polar cataract in one patient. (K) One patient presented with anterior polar cataract only.

Figure 2. Distribution of age at the time of cataract surgery in the first eye of the twelve patients operated for cataract. Over half of the patients underwent cataract surgery during childhood.



Developmental patterns of cataract in aniridia patients. (A) Early-stage discrete posterior subcapsular cataract. (B-E) The posterior subcapsular cataract is denser. A posterior subcapsular opacification develops on the capsule in the midperiphery as tiny flecks (B), or short, radially-oriented opacities (C). (D-F) Radial opacities extend towards to posterior pole and a mid-peripheral ring of opacification of varying density is present. Opacification always remained subcapsular. Late lens changes included yellow and turbid nuclear cataract (G), mature cataract (H) and hypermature cataract (I). (J) Anterior polar cataract in combination with a posterior polar cataract in one patient. (K) One patient presented with anterior polar cataract only.
163x227mm (300 x 300 DPI)



Distribution of age at the time of cataract surgery in the first eye of the twelve patients operated for cataract. Over half of the patients underwent cataract surgery during childhood.
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