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# Atypical sector pigmentary dystrophy

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**Abstract**

The authors describe a family with atypical sector shaped pigmentary dystrophy in two generations with transmission from father to son. A review of the literature is given.

## Introduction

In 1977 a patient (case II-4), frightened by the death of his sister (case II-2), asked for genetic counseling. A family examination was started. The findings were suggestive for an autosomal recessive inherited form of pigmentary dystrophy. An appointment for genetic counseling was made. At that moment the patient was no longer interested in genetics: it was a beautiful day . . . and his wife was pregnant. A child was born and this child (case III-8, proband) visited the ophthalmologist in 1985 because of diminished visual acuity. The boy presented with an atypical pigmentary dystrophy strongly resembling his father's fundus picture. Now the mother asked for genetic counseling. The family study was completed and genealogical work was done.

## Methods of examination

All the patients underwent routine ophthalmological examination. The parents and the sibs of the 1977-proband (case II-4) had extensive blood and urine chemistry testing. In family members with funduscopic retinal abnormalities electroretinography (ERG), electro-oculography (EOG), subjective dark-adaptation (DA) measurement with the Goldmann-Weekers adaptometer, color vision (CV) testing and perimetry were performed. The methods for ERG, EOG and CV testing were described elsewhere (38, 39, 40, 55).

## Case histories

Case III-8, HH, male, born 23071977, proband (Fig. 1). Examination 1985. VA OU 0.5 after correction sph. +2.00 cyl. −1.50 axis 180°. No complaints of nightblindness. No nystagmus. Alternating divergent squint. Clear ocular media. Fundus: normal optic discs, narrowed retinal arterioles in the lower nasal quadrant; granular aspect of the retinal pigment epithelium (RPE), sometimes like pepper and salt; in the equatorial region patches

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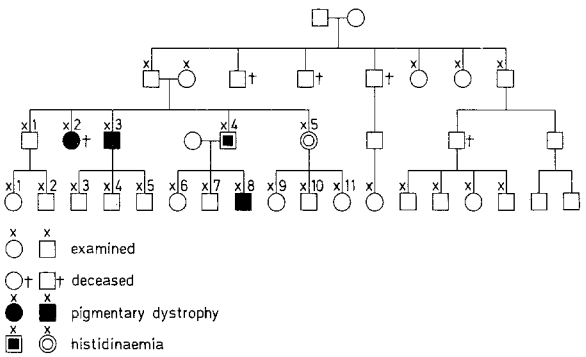


*Fig. 1.* Pedigree of a family with sector retinitis pigmentosa.

with pigmentation and choroidal atrophy; in the lower half of the retina a sharply delineated sector shaped atrophy (Fig. 2). ERG cone and rod function subnormal. EOG Lp/Dt ratio RE 1.38 and LE 1.47. CV: screening errors on AO-HRR, Ishihara, Tokyo Medical College test (TMC), and Standard Pseudoisochromatic Plates part 2 (SPP); New Color Test (NCT) box 4 normal; Lightness Discrimination Test (LDT) disturbed; Fm 100 Hue test red-green and blue-yellow axis, error score RE = 212 and LE = 260; anomaloscope examination not possible.

Case II-1, JH, male, born 17041937. Examination 1977. VA OU 0.8 after correction sph. −0.25 cyl. −0.5 axis 90°. No nystagmus. Normal ocular motility, normal IOP, clear ocular media. Fundus: normal optic discs, retinal vessels and macular area; some atrophic spots in the temporal superior periphery. Fluorescein angiography: normal. ERG cone function normal; rod function borderline. EOG Lp/Dt ratio RE 1.63 and LE 1.94. DA curve:borderline. CV:normal AO-HRR, NCT box 4, desaturated Panel D-15, FM 100 Hue and anomaloscope; many misreadings OU on Ishihara. Visual fields (Goldmann): OU depression of the lower fields. ECG normal. Serum kidney and liver functions and electrolytes and lipodogram normal; no histidinemia or -uria.

Case II-2, WH, female, born 12051939. Examination 1971. VA RE FC4m after correction sph. −2.00; LE HM 1m after correction sph. −1.00. Horizontal and rotatory nystagmus. Divergent squint LE. Nuclear cataract L>R. Liquefied vitreous. Fundus:atrophic optic discs, narrowed

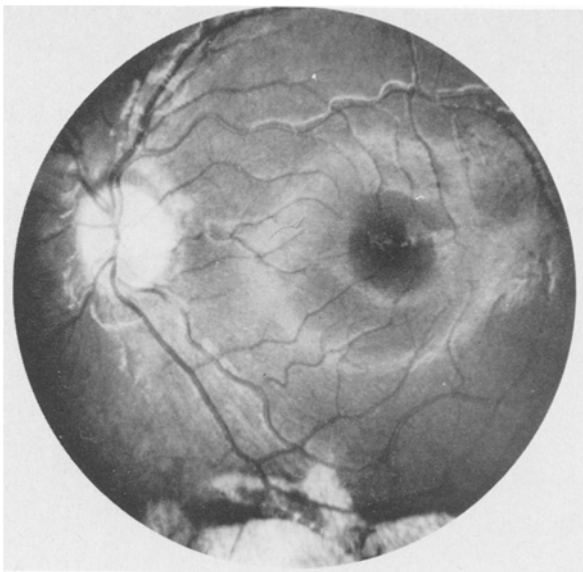

*Fig. 2.* Case III-8, posterior pole LE. Upper border of sharply delineated sector retinitis pigmentosa.

retinal arterioles; chorioretinal atrophy in the macular area, the equatorial region and especially the lower part of the retina. ERG cone and rod function almost absent. EOG and CV impossible. Examination 1977, after cataract extraction LE. VA RE FC 1m; LE HM 1m. Normal IOP. Fundusaspect unchanged. ECG normal. Serum kidney and liver functions, serum electrolytes and lipidogram normal. No histidinemia or -uria. She died in 1977 as a result of a traffic accident.

Case II-3, HH, male, born 11071940. Examination 1977. VA RE 0.3 after correction sph. −3.00 cyl. −0.5 axis 170°; LE 0.3 after correction sph. −3.50 cyl. −0.5 axis 120°. No complaints of night blindness until adultry. Horizontal nystagmus. No squint, normal ocular motility. Normal IOP. No cataract, normal aspect of the vitreous. Fundi: no pallor of the optic discs; narrowed retinal arterioles, especially in the lower part of the fundus; pigment dispersion macular area; peripheral pigmentations and atrophic patches, in the lower retina a sector shaped atrophy with heavily pigmented borders. Fluorescein angiography: pigment dispersion macular region; in the periphery, especially the lower retina, irregular pigment dispersion with pigment clumping and atrophy. ERG cone and rod

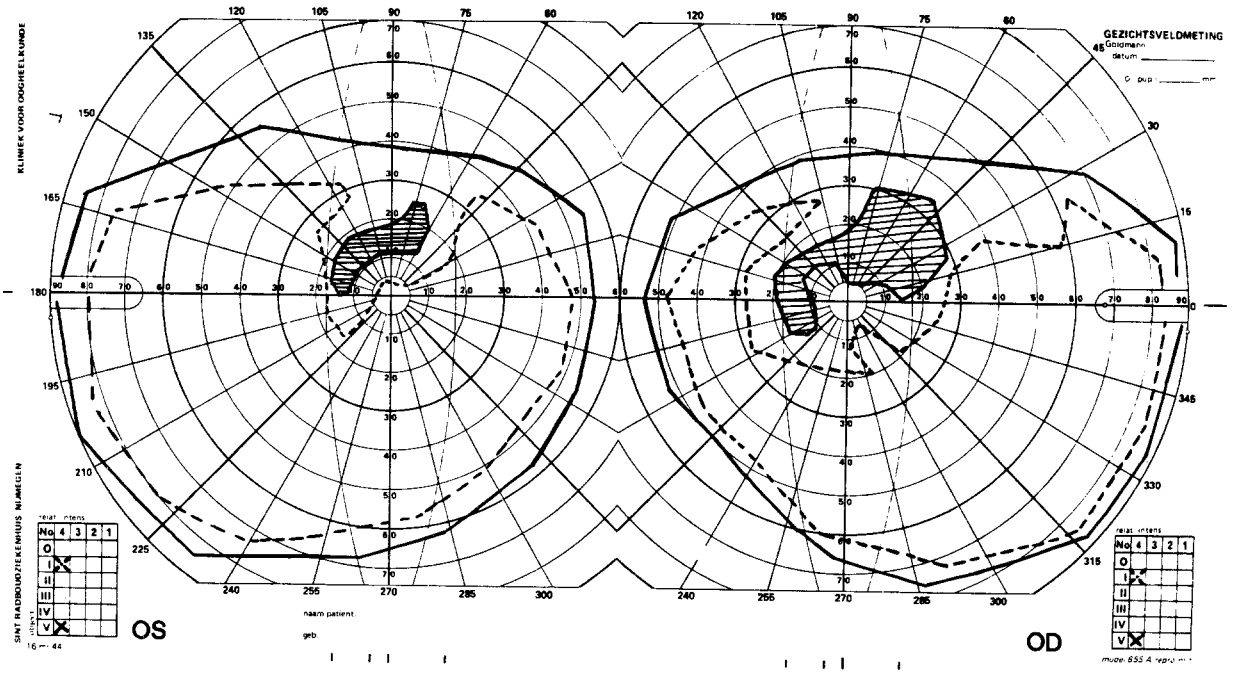


*Fig. 3.* Case II-3. Visual field defects corresponding with the sector retinitis pigmentosa.

function subnormal. EOG flat OU. DA curve borderline. CV: RE type III acquired blue-yellow defect (AO-HRR, NCT box 4 and 6, FM 100 Hue, Pickford-Nicholson anomaloscope); LE pseudoprotanomaly (Nagel II anomaloscope), otherwise normal. Visual field (Goldmann): enlarged blind spot OU; absolute scotoma upper part corresponding with the atrophic patches in the lower retina (Fig. 3). Serum kidney and liver functions, serum electrolytes and lipidogram normal. No histidinemia or -uria.

Case II-4, GH, male, born 18051942, father of the proband. Examination 1974. VA RE 0.4 after correction sph. −6.00 cyl. −0.50 axis 20°. LE 0.6 after correction sph. −4.00 cyl. −0.25 axis 75°. No complaints of night blindness until military service. Horizontal and rotatory nystagmus. No squint, normal ocular motility, normal IOP. Clear ocular media. Fundi: no pallor of the optic discs; pigment dispersion, patches with chorioretinal atrophy OU, sector shaped atrophy in the lower retina. Fluorescein angiography: especially in the lower part of the retina OU patches with atrophy of varying degree with irregular borders (Fig. 4 & 5). ERG cone and rod function subnormal. EOG Lp/Dt ratio RE 1.56 and LE 2.00. DA curve subnormal. CV: RE normal AO-HRR, Panel D-15; blue-yellow defect on FM 100 Hue; misreadings Ishihara and Farnsworth Tritan-plate; pseudoprotanomaly (Nagel anomaloscope). LE normal Panel D-15; red-green defect on AO-HRR, Tritan-plate and Ishihara; blue-yellow defect on FM 100 Hue; pseudoprotanomaly. Visual field (Goldmann): absolute scotoma OU in superior part, corresponding with the fundus aspect (Fig. 6). Examination 1977 and 1985. Clinical picture unchanged. Serum kidney and liver functions, serum electrolytes and lipidogram normal. HISTIDINEMIA and HISTIDINURIA. There were no other family members presenting with ocular abnormalities (Fig. 1). The grandparents of the proband had normal blood chemistry. Case II-5 had histidinemia and -uria. The mother of the proband, and also her mother, have diabetes without retinopathy.

Genealogical work was done (AP). In 6 generations of ancestors there was no indication for consanguinity between the mother and father of the proband. The mother stems from a twofold con-

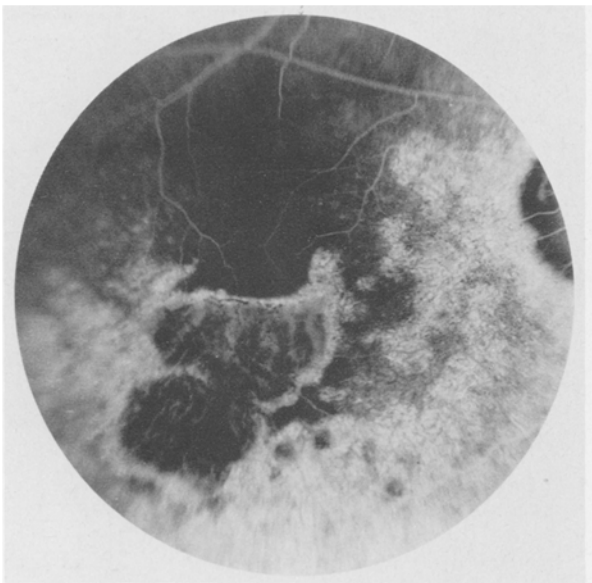

*Fig. 4.* Case II-4, posterior pole RE. Sector retinitis pigmentosa crossing the midline.

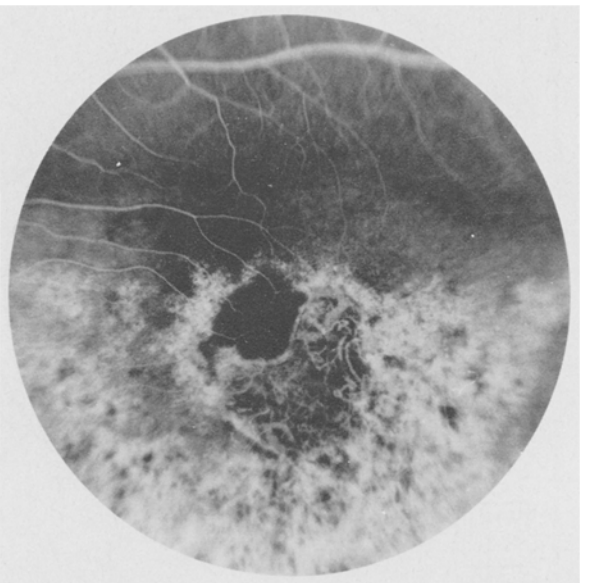

*Fig. 5.* Case II-4, posterior pole LE. Sector retinitis pigmentosa, pigment dispersion, atrophy choriocapillaris and RPE.

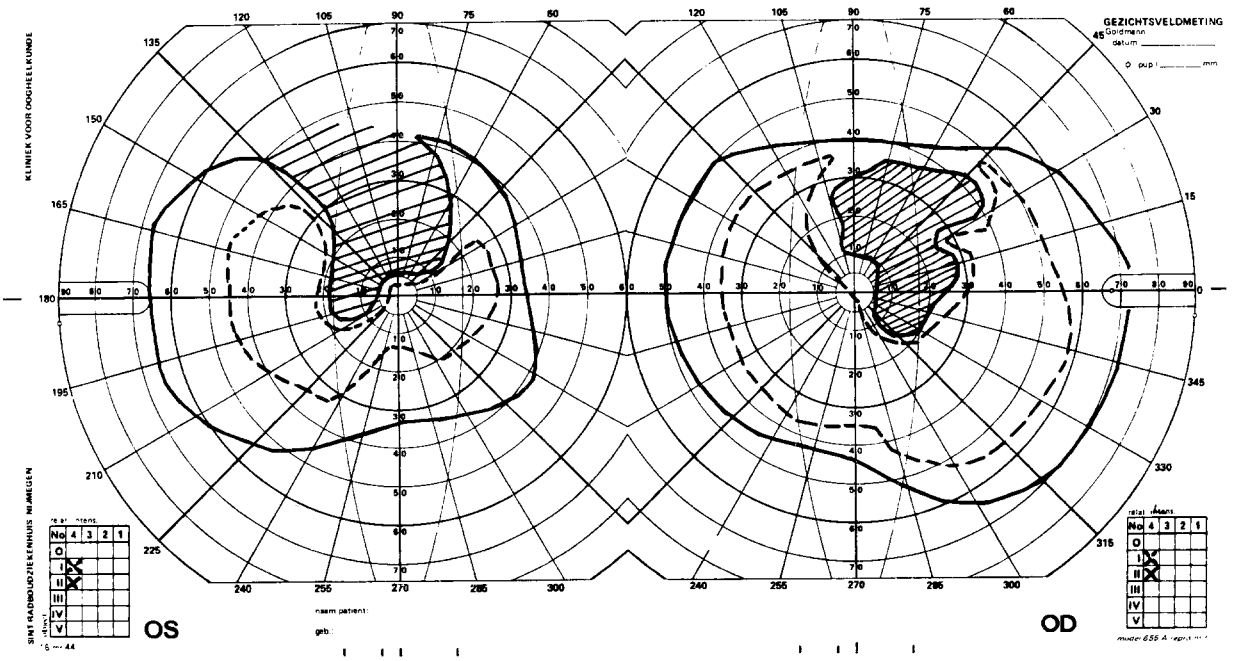


*Fig. 6.* Case II-4. Visual field defects correspond with fundus picture.

sanguineous marriage. There was also no indication for consanguinity between the paternal grandparents.

**Discussion**

If the 1977-proband had not canceled the genetic counseling we would have concluded in 1977 to an autosomal recessive pattern of inheritance for sector pigmentary dystrophy and histidinemia, the two traits being dissociated. Because of the transmission from father to son we have, 8 years later, to face either pseudodominance or an autosomal dominant heredity. There was no indication for consanguinity in 6 generations.

Case II-1 has a borderline ERG rod-function, a borderline DA curve, a subnormal RE EOG and a depression of the visual fields. This depression however affects the lower part of the fields, while the other affected members have scotomata in the upper part. It therefore is, in our opinion, not justified to diagnose case II-1 as a case with minor expression of the gene. The 2 children of this patient have no fundus abnormalities.

If we accept an autosomal dominant heredity we also have to accept a reduced penetrance because the paternal grandparents have no fundus abnormalities. Reduced penetrance is not uncommon in pigmentary dystrophy (7, 9).

The localization of the disease is predominantly in the lower part of the fundus, with corresponding scotomata in the upper part of the visual fields (Fig. 3, 6). The disease therefore has to be classified as a form of sector retinitis pigmentosa (SRP) or sectoral pigmentary dystrophy that was described for the first time by Bietti in 1937 and later by many authors (29, 31, 33, 46). SRP is a bilateral and symmetrical disease (78% of the cases; 3, 5, 19, 21) involving the lower (48%) or uniquely the inferior nasal quadrant. The aspect of the optic disc is normal in 68%. The retinal vessels are normal (25%) or narrowed (75%), especially in the inferior quadrants. The visual field defects usually are in agreement with the fundus aspect (11, 25), but may give the impression of bitemporal hemianopsia (15).

Visual acuity is more than 0.5 in 85% of the cases. The subjective DA curve is normal to monophasic with disappearance of the scotopic segment, even if the ERG is normal or near-normal (16, 17). The EOG is more disturbed than the ERG (17, 51); the EOG may be subnormal when the ERG is normal (10, 52). The ERG is normal, subnormal or abolished (1, 11, 27, 28, 36, 42, 58). In some cases there is a rod-cone, in other cases a cone-rod type ERG disturbance (18). The ERG implicit times (20) and the ERG culmination times (6) are normal. Sometimes the ERG decrease is more pronounced than expected on the basis of the fundusaspect (23, 42). Complaints of nightblindness were noticed in adults but not in younger individuals (25). Our findings are in agreement. CV is normal or blue-yellow defective (11) with pseudoprotanomaly (present study). Fluorescein angiography learns that the RPE is disturbed in a larger area than visible by funduscopy. In the impaired quadrants the vessels are narrowed and the flow delayed. There is atrophy of the RPE, the choriocapillaris and the choroidal vessels (2, 50).

From follow-up studies we know that SRP, although a dystrophy, is a disease with a relatively favourable course. There seems to be no correlation between the results of electrophysiology and other symptoms of visual function (18). In infants with barely visible fundus alterations the ERG b-wave is already decreased (25). VA and CV remain stable, while EOG, DA and visual field deteriorate (24). Tazawa *et al.* (50) report a similar loss of visual function, independent of age, in 3 members of a family.

Iuxtapapillar (56), unilateral (35, 41, 48) and an asymmetrical case (44, cited by Bisantis) are described. Another atypical form of SRP has fundus abnormalities in other than the inferior quadrants (25, 54; present study). In such cases all components of the ERG are decreased; the EOG is depressed. Occlusion of the posterior ciliary artery may mimic atypical SRP, but then the EOG may be normal (54). SRP is reported in combination with chronic disc edema (13), juvenile nephronophthisis (20), congenital achromatopsia (22, 53), neurofibromatosis (32), vitiligo (35), chronic angle-closure glaucoma (37), Morgagni syndrome (45, cited by Bisantis) and atypical Laurence-Moon-Bardet-

Biedl syndrome (49, cited by Bisantis).

SRP has a familial occurrence (4, 14, 30, 34, 37). An autosomal recessive inheritance (11, 20) as well as an autosomal dominant inheritance pattern (8, 12, 25, 26, 43, 47, 50) are reported. Two sisters, heterozygotes for X-linked recessive retinitis pigmentosa, also presented with SRP (10).

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