Optical Coherence Tomography in Group 2A Idiopathic Juxtafoveolar Retinal Telangiectasis

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Objective: To describe the changes observed with optical coherence tomography in group 2A idiopathic juxtafoveolar retinal telangiectasis.

Methods: We retrospectively reviewed the medical records of 13 patients (25 eyes). All eyes underwent optical coherence tomography examination consisting of 6 radial scans, fundus color photography, and fluorescein angiography. We calculated retinal foveal and central foveal thicknesses from software mapping results. We compared the optical coherence tomography data with fundus photography and fluorescein angiography findings.

Results: Foveal cystoid spaces, very small or more prominent, were present in 20 of 25 eyes. Some degree of disruption of the inner segment/outer segment photoreceptor junction line was observed in 18 eyes as from stage 2 of idiopathic juxtafoveolar retinal telangiectasis, and intraretinal pigmentary proliferation was observed in 9. A foveal detachment without subretinal new vessels was also present in 2 eyes. Despite these abnormalities, central foveal thickness was below or within the range of reference values in all eyes; foveal thickness, in 23 of 25. In the more advanced cases, severe disruption of the inner segment/outer segment photoreceptor junction line and outer retinal atrophy were seen.

Conclusions: Early in the evolution of group 2A idiopathic juxtafoveolar retinal telangiectasis, the optical coherence tomography examination disclosed intraretinal cystoid spaces without foveal thickening and disruption of the inner segment/outer segment photoreceptor junction line. Foveal thinning was present in later stages.

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DIOPTIC JUXTAFOVEOLAR RETINAL TELANGIECTASIS (IJRT) is a rare cause of progressive bilateral visual loss, usually diagnosed in the fifth or sixth decade of life. Gass,1 who originally described this condition as IJRT in 1968, classified the disease into several types on the basis on ophthalmoscopic and angiographic findings.2 In 1993, Gass and Blodi3 revised this classification and defined 3 distinct groups or types. Group 2A is one of the most common subgroups in this classification and defined by occult, bilateral juxtafoveolar telangiectasis with minimal exudation, superficial retinal crystalline deposits, right-angle venules, and pigmentary changes characterized by the intraretinal migration of retinal pigment epithelium (RPE) cells. This group has been subdivided into 5 stages of development, according to the progression of the disease,1 with stage 5 characterized by the occurrence of subretinal neovascularization.

A few histopathological studies showed a slight retinal thickening in the temporofoveal area, combined with tiny microcystic cavities in the inner and outer plexiform layers and no distinct telangiectasis.4 The etiology and genetic factors of IJRT are unknown, and there is little information about its natural history. Laser photocoagulation is known to be ineffective for group 2A IJRT. More recently Yannuzzi et al5 proposed simplifying this classification and used the term perifoveal telangiectasia, which we will also use as equivalent of group 2A IJRT. We describe herein the optical coherence tomography (OCT) findings for 13 patients with group 2A IJRT and discuss its pathogenesis.

METHODS

We retrospectively reviewed the medical records of patients who underwent OCT ex-
amination for macular telangiectasis from 2003 to 2005 in 2 different settings. The files were retrieved in the databases of the OCT machines in both settings. Thirteen patients had group 2A IJRT when initially examined, according to the Gass classification, and were included in this study. No institutional review board approval was needed for this retrospective study.

For each eye, corrected visual acuity had been recorded on a Snellen chart. All eyes had undergone color and red-free fundus photography, fluorescein angiography (FA), and OCT examination with the Stratus OCT 3 (Carl Zeiss Meditec, Dublin, Calif). Six-millimeter mapping of the macula was available in all medical records, and in most cases additional scans had been performed to improve the assessment of their macular profile. All of the radial scans of each mapping were examined. One case was excluded because of a history of subretinal new vessels that had been treated with laser photocoagulation, leaving 25 eyes of 13 patients for the study.

Table. Clinical Characteristics of the Population With Group 2A IJRT

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<tr>
<th>Patient No./ Sex/Age, y</th>
<th>Eye</th>
<th>IJRT Stage</th>
<th>VA</th>
<th>Foveal Cysts</th>
<th>IS/OS PR Junction Line Disruption</th>
<th>Foveal Detachment</th>
<th>Pigment Changes</th>
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<th>FT, µm‡</th>
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Abbreviations: CFT, central foveal thickness; FT, foveal thickness; IJRT, idiopathic juxtafoveolar retinal telangiectasis; IS/OS PR, inner segment/outer segment photoreceptor; L, left; R, right; VA, visual acuity; +, present.

†Indicates the mean value of retinal thickness at the intersection of the 6 radial scans of the mapping. The mean ± SD reference value (determined by manual measurement) was 170 ± 18 µm.

‡Indicates average of all the retinal thickness measurements in the central 1000-µm area of the mapping. The mean ± SD reference value was 212 ± 20 µm.

§Subretinal new vessels were seen.
||Patient 7 had 2 years of follow-up.
||Patient 11 had 20 months of follow-up.

Attention was paid to the appearance of the inner segment/outer segment photoreceptor (IS/OS PR) junction line. Although we had no control group, because the study was retrospective, we compared FT and CFT values with the range of reference values provided by Chan and Duker (mean ± SD FT, 212 ± 20 µm; mean ± SD CFT, 170 ± 18 µm).

At diagnosis, patients’ mean age was 58 years (range, 39-73 years). They included 5 men and 8 women. Telangiectasis was bilateral by definition, and all of the 26 eyes were involved. However, we excluded 1 eye from the analysis because subfoveal neovascularization was found at the first examination. The main characteristics of the included eyes are given in the Table.

On color and red-free photographs as well as FA, 5 of the 25 eyes met the Gass criteria for stage 2 of group 2A IJRT, ie, slight graying and loss of transparency of the parafoveal retina and very small telangiectatic capillaries (Figure 1). Eleven of the 25 eyes corresponded to stage 3 of group 2A IJRT, ie, eyes in which 1 or several slightly dilated blunted retinal venules are present in the parafoveal retina (Figures 2, 3, and 4). Nine eyes corresponded to stage 4 of group 2A IJRT, ie, they
exhibited some degree of juxtafoveal RPE hyperplasia (Figure 5 and Figure 6). In one of these cases, RPE hyperplasia extended onto the retinal surface, on the temporal side of the fovea (Figure 6). In addition, some tiny crystalline deposits were also present in the macula of 10 of the 25 eyes (Figure 2).

Fluorescein angiography confirmed the presence of mild capillary telangiectasis in all eyes. The telangiectasis was limited to the temporal half of the foveal area or less in 18 eyes and included all of the fovea in the other 7. On the late phase of the angiogram, diffuse leakage was observed in the area of telangiectasis, without definite cystoid spaces and, especially, no central cyst.

On OCT images, 1 or several abnormalities were noted on radial scans, especially on horizontal scans. They included intraretinal cystoid spaces (foveal cysts), disruption of the IS/OS PR junction line, foveal detachment, foveolar thinning, and RPE proliferation. These different findings were variously combined.

Foveal cysts were present in 20 of 25 eyes. In 9 eyes, the cystoid spaces were tiny and located in the inner layer of the foveola, at its center or slightly shifted temporally (Figures 1 and 2). In 11 eyes, the cystoid spaces were more prominent and tended to involve the entire foveolar thickness (Figure 3). Some degree of disruption of the IS/OS PR junction line was present in 18 eyes, including 2 of the 3 eyes with stage 2 disease, 8 of the 13 eyes with stage 3 disease, and 8 of the 9 eyes with stage 4 disease (Figures 1, 3, 4, 5, 6, 7, 8, and 9). Intraretinal pigmentary proliferation was observed on OCT images in 8 eyes. In 3 of these, it consisted of small intraretinal hyperreflective dots only. In 4 eyes, pigmentary proliferation formed a dome-shaped hyperreflective elevation progressing into the retinal tissue (Figure 5). In the last case, pigment proliferation formed a flat hyperreflective structure in the inner retinal layer, masking the features of the underlying tissue (Figure 7). In 2 eyes, a foveal detachment was present at least at 1 stage of the evolution (Figure 9).
These abnormalities were variously combined. Foveal cysts were the only abnormality in 6 eyes and were combined with disruption of the IS/OS PR junction line in 14 eyes, intraretinal pigmentary proliferation in 6, and foveal detachment in 2.

Central foveal thickness was within or below the range of reference values in all eyes, despite the presence of intraretinal cystoid spaces or foveal detachment (Table). In 16 of 25 eyes, it was less than 134 µm, ie, less than 2 SDs below the reference CFT. Foveal thickness was within or below the reference range in 24 of 25 eyes, and less than 2 SDs below the reference value of 172 µm in 11 of 25 eyes. Two eyes had FT greater than the reference values, one because of thick subfoveal pigmentary proliferation and the other because of a foveal detachment. In these 2 eyes, however, the retina itself had not thickened.

In 2 patients, it was possible to record the evolution of perifoveal telangiectasia during a period longer than 1 year. In patient 7, both eyes had stage 2 disease, and tiny foveolar cystoid spaces in the inner foveolar layer were detected at the initial examination. Two years later, the cystoid spaces were significantly enlarged and involved the outer retina (Figure 1). In patient 11 (Figures 8 and 9), the right fovea was already atrophic at the first examination, whereas the fovea was detached in the left eye. This eye was treated with photodynamic therapy on an empirical basis but without effect at 3 and 6 months, as previously reported. However, the detachment progressively resolved within the next 18 months and the fovea became atrophic, as in the right eye.

Idiopathic juxtafoveal retinal telangiectasis is a rare disease that was described by Gass in several publications from 1968 to 2005. In 1993, Gass and Blodi revised the classification and proposed to divide IJRT into 3 groups, among which group 2A corresponds to occult juxtafoveal telangiectasis, minimal exudation, superficial retinal crystalline deposits, and right-angle venules. Late in the course of the disease, foveal atrophy, intraretinal pigment plaques, and subretinal neovascularization develop. The disease affects both sexes and usually becomes symptomatic in the sixth decade of life. Gass and Blodi also proposed, on the basis of their observation of more than 100 cases, to divide the evolution of group 2A IJRT into 5 stages ranging from stage 1 (asymptomatic) to stage 5 (characterized by the proliferation of subretinal new vessels). The most common feature of all the stages is mild or moderate leakage in the fovea in the late phases of FA, on the temporal side or throughout the whole foveal area. Beginning in stage 2, slight graying and loss of transparency of the affected part of the fovea become visible on biomicroscopy, as well as telangiectatic capillaries. At stage 3, slightly dilated and blunted venules become visible. Gass and Blodi stressed that these dilated venules, which extend at a right angle deep into the parafoveal retina, are the sign of the development of capillary proliferation—from the outer capillary plexus, within the outer retinal layers, and even in the subretinal space—without any connection to the choroidal circulation. At
stage 4, foci of hyperplasia of the RPE begin to invade the retina. In some cases, these proliferations may even pass through the retina and proliferate on its surface.

The pathogenesis of IJRT is unknown. It is usually thought to be a primary disease of the macular capillaries, although the concomitant progressive atrophy of...
Figure 4. Patient 5 (both eyes) with stage 3 of group 2A IBRT. Visual acuities are 20/30 OD and 20/40 OS. A, Red-free photograph of the right eye (left) and fluorescein angiogram of both eyes (center and right) show dilated blunted venules and temporal telangiectasia. B, Horizontal 6-mm OCT scan and detail in the right eye (RE). Small inner foveal cysts are present in a nonthickened retina. There is also a small disruption of the IS/OS PR junction line (arrowhead). C, Horizontal 6-mm OCT scan and detail in the left eye (LE). The small inner cyst is combined with a large defect in the outer retina. The retinal tissue remaining at the foveal center is extremely thin (central foveal thickness, 100 µm). See the Figure 1 legend for abbreviations.

Figure 5. Patient 6 (right eye) with stage 4 of group 2A IBRT. Visual acuity is 20/40. A, Color fundus photograph in the right eye shows intraretinal pigmentary proliferation on the temporal side of the fovea in the area of telangiectasia. B, The horizontal 6-mm OCT scan (top; H in part A) shows an inner foveal cyst, and the disruption of the IS/OS PR junction line (arrowheads). This oblique 6-mm scan (bottom; O in part A) passes through the pigmentary proliferation and shows a hyperreflective bulge invading the retina. Some smaller intraretinal pigmentary migrations (arrow) are also seen. Central foveal thickness is 123 µm. See the Figure 1 legend for abbreviations.
the neural and glial tissue has not been satisfactorily explained. New attempts were made recently to rename this disease as parafoveal telangiectasis or macular telangiectasis. The merit of the modified classification by Yannuzzi et al is to simplify the Gass model. Accordingly, the group 2A IJRT is called type 2 or perifoveal telangiectasia, and its 5 stages are reduced to nonproliferative and proliferative stages. However, in the absence of a consensus of opinion we have kept the term used by Gass.

A few histological data are available. In a postmortem study, Green et al found only very small cystic changes in the plexiform layers of the retina. No telangiectatic capillaries were seen, but the capillary wall had thickened and the pericytes had degenerated. Gass reviewed the histopathological sections of the eye reported by Green et al and found evidence of retinal capillary invasion of the retinal receptor layer. In another histopathological report, Eliassi-Rad and Green found telangiectatic vessels, subretinal neovascularization, and intraretinal pigment migration along the telangiectatic vessels. In a recent histopathological study of a specimen of submacular vascular proliferation surgically removed in a case of IJRT, Davidorf et al showed that retinal capillaries had proliferated under the retina on an intact RPE layer. Although their interpretation of the pathogenesis of IJRT was contested by Gass, the extension of the retinal capillaries throughout the outer retina, with little or no fibrosis, is one of the characteristics of the advanced stages of IJRT.

The other aspect of the disease pointed out by Gass is the progressive retinal atrophy in the area of telangiectasia. In this connection, he noted that “atrophy of the foveolar retina develops in the absence of typical cystoid edema.” He suggested that the atrophy could be due to the degeneration of Muller cells and consequently of the PR cells. Loss of the PR cells would then allow RPE cells to migrate onto the overlying retina. This aspect of the disease has been less discussed than the significance of the telangiectasis itself.

The merit of OCT is to provide information about the retinal structure and thickness in IJRT, which until now was mainly studied by means of FA. In the present study, OCT images showed intraretinal cystoid spaces or cysts at the early stage of the disease, as reported by Yannuzzi et al and Paunescu et al. However, we found that cysts were not usually associated with foveal thickening. The OCT images also disclosed that the disruption of the IS/OS PR junction line occurred relatively early in the course of the disease and revealed some unexpected features such as foveal detachments not due to subretinal proliferation of new vessels.

Figure 6. Patient 9 (right eye) with stage 3 of group 2A IJRT. Visual acuity is 20/25. A and B, Red-free photograph (A) and fluorescein angiogram (B) show macular telangiectasis with dilated venules and fluorescein leakage. C, Horizontal 6-mm OCT scan (left) and detail (right) show foveal atrophy with disruption of the IS/OS PR junction line (arrowhead), disappearance of the outer nuclear layer in the temporal part of the fovea (star), and a large outer retinal defect in the foveal center (arrow). See the Figure 1 legend for abbreviations.
In this series of cases, retinal FT was never greater than the reference values, except for 2 eyes in which FT was artifactually increased by the presence of significant foveal detachment or subretinal pigment proliferation. In 17 of 25 eyes, FT was even less than the reference values. Central foveal thickness was also less than the reference values in 21 of 25 eyes, and within the range of reference values in the other 4. Keeping in mind that variously large cystoid spaces were present in 20 of 25 eyes, this means that in these cases there was some degree of retinal atrophy.

Disruption of the IS/OS PR junction line was also common, at least on the temporal side of the fovea, but it also extended to the whole fovea in some advanced cases. The IS/OS PR junction was first recognized on ultrahigh-resolution OCT images as a hyperreflective line 20 to 40 µm above the PR line. This line is in fact usually visible on Stratus OCT scans. We are aware that it represents only an optical signal of the presence of the PR, probably because of the particular structure of the IS/OS junction, and that it does not represent the PR cells themselves. Disruption of this line does not necessarily mean loss of the PR cells. For instance, in this series we observed foci of IS/OS disruption in eyes with stages 2 and 3 disease, which retain good vision. The PR signal might be impaired by a change in the spatial orientation of the PR outer segments or by any other change in their structure without cell death, as in the invasion of the outer retina by retinal capillaries shown in some cases by ultrahigh-resolution OCT. In other cases, the absence of an IS/OS PR junction signal was combined with the disappearance of the hyporeflective band of the outer nuclear layer and strongly indicates PR atrophy (Figures 6, 7, 8, and 9).

It is not yet known whether telangiectatic changes in the parafoveal capillaries are the cause of the atrophic process, or whether neuronal or glial dysfunction may cause these changes.

A foveal detachment was disclosed herein on OCT images in 2 of 25 eyes and was not associated with prominent cystic spaces. Subretinal new vessels were not suspected on FA or on OCT images. The significance of this detachment is unclear.

The OCT images also illustrated RPE changes in 9 of 25 eyes, which included some small intraretinal hyperreflective foci or intraretinal pigment bulging (Figure 5) and even preretinal pigment proliferation in 2 eyes (Figure 7). This intraretinal RPE proliferation has been explained by the loss of PR cells, which allows the RPE cells to migrate into the overlying retina, especially along the venules. In the present series, all of the eyes exhibiting RPE proliferation and migration indeed had disruption of the IS/OS PR junction and some degree of retinal atrophy.
Figure 8. Patient 11 (right eye) with stage 3 of group 2A IJRT. Visual acuity is 20/63. A, At the initial examination, a color fundus photograph and fluorescein angiograms (top) show capillary telangiectasis occupying all of the foveal area. B, A horizontal 6-mm OCT scan (bottom) shows the disappearance of the outer nuclear layer (star) and of the IS/OS PR junction line at the foveal center (between the 2 arrowheads). C, Twenty months later, the OCT scan shows more atrophic changes in the foveal center. Central foveal thickness decreased from 136 to 97 µm. See the Figure 1 legend for abbreviations.

Figure 9. Patient 11 (left eye) with stage 3 of group 2A IJRT. Visual acuity is 20/63. A, At the initial examination, a color fundus photograph and fluorescein angiograms (top) show capillary telangiectasis occupying all of the foveal area. B, A horizontal 6-mm OCT scan (bottom) shows that the fovea is detached (arrows). This eye underwent photodynamic therapy, which had no effect at 6 months. C, Twenty months later, the OCT scan shows the disappearance of the outer nuclear layer and of the IS/OS PR junction line at the foveal center (between the two arrowheads), as in the right eye. Central foveal thickness is only 74 µm. See the Figure 1 legend for abbreviations.
retinal atrophy or microcystic changes. The 2 cases in which RPE proliferation reached the retinal surface in a tumorlike formation constituted a condition that is rare but has already been detected on OCT images. Unlike congenital simple hamartomas of the RPE, these hyperplasia were flat and the fellow eye also had some degree of intraretinal RPE proliferation with well-characterized JFRT.

We recognize the limitations of this study. First, it was retrospective; second, the number of patients studied is probably too small to cover all aspects of the disease; and third, we were able to follow its evolution in only 2 patients during too short a period of time. A longer follow-up, as initiated by the prospective MacTel study (http://www.mactelresearch.com), would be necessary to better understand the natural history of macular telangiectasia. Moreover, although the accuracy of the images obtained by Stratus OCT is excellent, we realize that images obtained by ultrahigh-resolution OCT would perhaps oblige us to modify some of our interpretations. Finally, OCT images do not show the vascular component of JFRT. However, we have always compared OCT data with those provided by FA and fundus examination when interpreting OCT images. Future progress in the knowledge of the vascular component of this disease should include other imaging technologies such as confocal FA or adaptive optics. Functional testing such as multifocal electroretinography and microperimetry should also be useful to evaluate the functionality of the PR cells in the affected area. Nevertheless, from this series of 25 eyes, we can conclude that microcystoid changes in the fovea without retinal thickening and often with retinal thinning is a common feature in JFRT. Focal disruption of the IS/OS PR junction is also frequent combined with microcystoid spaces. In some cases, there is even a patent atrophy of the foveal outer retina. Pigmentary proliferation and migration are often associated with outer retinal atrophy. The primary cause of the disease remains unknown and might reside in the capillaries themselves or in the surrounding neuronal and glial tissue.

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REFERENCES