Retinitis Pigmentosa Associated With Fuchs’ Heterochromic Uveitis

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Objective: To investigate whether the combination of Fuchs’ heterochromic uveitis (FHU) and retinitis pigmentosa (RP) in the same patient is coincidental or represents a true association.

Methods: We have examined the frequency of FHU in 338 patients with RP and in 1984 patients who were seen in our primary care ophthalmic clinic because of reasons other than RP.

Results: Of 338 patients with RP, 4 (1.2%) had the typical findings of FHU. Three of them had Usher syndrome type II, and 1 had RP simplex. By contrast, only 1 patient in the control group had FHU (5%), and the difference in the frequency of FHU between the 2 groups was significant (P = .002, Fisher exact test).

Conclusions: Fuchs’ heterochromic uveitis is associated with RP. Since autoimmune phenomena have been previously described in patients with RP, it is conceivable that RP predisposes to the development of FHU.

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Fuchs’ heterochromic uveitis in those patients was typical, except that the 2 autosomal dominant RP cases were a father and son. Whether the phenomenon of both FHU and RP in the same patient is coincidental or represents a true association was a matter of debate. This question could not be decided, since all previous reports included case description alone. Our data from a controlled study clearly show, for the first time, that FHU was indeed associated with RP in our cohort. Furthermore, since 3 of our 32 patients with Usher syndrome type II had FHU, it seems that FHU is even more common in patients with Usher syndrome type II than in other patients with RP.

To explain the relatively high incidence of FHU in patients with RP, the pathogenesis of FHU and the inflammatory features of RP should be considered. Histological sections from eyes with FHU show inflammatory reaction that is predominated by lymphocyte and plasma-cell infiltration. In addition, several immune abnormalities have been described in FHU, among them the existence of autoantibodies to a corneal antigen, oligoclonal immunoglobulin G (IgG) in the anterior chamber, circulating immune complexes, and reduced activity of suppressor T-cell lymphocytes. Therefore, activation of both T-cell and B-cell lymphocytes seems to play a role in FHU. However, anterior chamber antigens that are the target of the immune response, other than the aforementioned corneal antigen, have not been identified so far.

Patients with RP and patients with uveitis show several common clinical and laboratory inflammatory features. Among these are vitreal cellular infiltrations, cataract, leakage from retinal blood vessels with retinal edema, and, rarely, retinal neovascularization. Both cell-mediated and humoral autoimmune responses to retinal antigens were found in patients with RP. For example, it was demonstrated that some patients with RP have circulating B cells reactive with retinal antigens; it was speculated that these cells represent immature plasma cells. It is unclear whether such autoimmune reactions

### Patient Data*

<table>
<thead>
<tr>
<th>Patient No./ Age, y/Sex</th>
<th>RP Type</th>
<th>Age at Diagnosis, y</th>
<th>Eye Affected by FHU</th>
<th>Complications</th>
<th>Treatment</th>
<th>Best-Corrected Visual Acuity</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/33/F</td>
<td>Usher type II</td>
<td>15</td>
<td>Right</td>
<td>PSC cataract</td>
<td>Cataract extraction</td>
<td>6/6</td>
</tr>
<tr>
<td>2/31/F</td>
<td>Usher type II</td>
<td>21</td>
<td>Left</td>
<td>PSC cataract</td>
<td>Cataract extraction</td>
<td>6/12</td>
</tr>
<tr>
<td>3/42/M</td>
<td>RP simplex</td>
<td>20</td>
<td>Right</td>
<td>HTN</td>
<td>0.5% Timolol</td>
<td>6/9</td>
</tr>
<tr>
<td>4/36/F</td>
<td>Usher type II</td>
<td>18</td>
<td>Left</td>
<td>PSC cataract</td>
<td>Cataract extraction</td>
<td>6/36</td>
</tr>
</tbody>
</table>

*RP indicates retinitis pigmentosa; FHU, Fuchs’ heterochromic uveitis; PSC, posterior subcapsular; HTN, ocular hypertension; and HM, hand movements.

Figure 1. Right (left) and left (right) iris of patient 2. Marked hypochromia and posterior subcapsular cataract are evident in the right eye.
are primary or secondary to the dystrophic process. However, it is interesting to note that inflammatory activity, when present in patients with RP, is similar to that in FHU in being low-grade and chronic. It is possible that autoimmunity against ocular antigens in patients with RP may lead to an increased risk for ocular inflammation. Although phenotypically similar to other patients with RP, it is possible that certain RP subtypes, such as Usher type II, are more susceptible to FHU because of a higher inflammatory tendency.

We speculate that the tendency for autoimmune reactions in patients with RP could be the cause for their increased susceptibility to develop FHU. Patients with RP may develop autoimmune reactions to anterior chamber antigens that are similar to retinal antigens, leading to the clinical manifestation of FHU. However, since most patients with RP do not suffer from FHU, and since FHU in patients with RP is unilateral, additional and yet unknown factors are possibly required to evoke FHU in patients with RP.

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REFERENCES