The patient, a 4-year-old boy, was diagnosed as having Blau syndrome based on the manifestation of typical clinical features (ankle and wrist arthritis/tenosynovitis, diffuse eczematous rash, and uveitis), histologic evidence of noncaseating granulomas, and a heterozygous NOD2 mutation (p.R334W).
Ocular involvement was initially controlled by topical and oral corticosteroids, but over the years visual impairment progressed.
Other manifestations of Blau syndrome (arthritis and rash) subsided over time.
Bilateral panuveitis progressed after age 5 years, and was initially treated with methotrexate.
However, ocular inflammation persisted despite the addition of local steroid injections and repeated intravenous (IV) bolus methylprednisolone treatment; therefore, when the patient was age 10 years, infliximab (initially at 5 mg/kg increased to 10 mg/kg IV every 4 to 6 weeks) was initiated.
Although there was an initial improvement, 1 year after this treatment was started uveitis worsened, and at age 12 years infliximab was discontinued.
Adalimumab (24 mg/m2 every 2 weeks) was then initiated and the dosage of methotrexate (15 mg/m2/weekly) was increased.
However, ocular disease remained active.
Mycophenolate mofetil (750 mg/m2) and then abatacept (10 mg/kg/month IV) were sequentially administered, without significant improvement.
At age 16 years the patient still had granulomatous retinal lesions and anterior chamber inflammation, and macular edema developed, which led to retinal detachment.
In addition to the other steroid therapy, corticosteroid pulse therapy was necessary to control disease flares, with an average of 3 boluses/month for 6 consecutive months.
Because of the supposed autoinflammatory nature of Blau syndrome, we initiated a trial of IL-1 antibody administration (2 mg/kg/month of canakinumab).
During the 6 months that followed, no ocular flare occurred and no steroid pulse therapy was necessary.
Concomitant treatment with oral methotrexate and low-dose prednisone (0.2 mg/kg/day) remained unchanged.
Figure 1 shows fluorangiograms before treatment and after the first 6 injections.
The drug was well tolerated with no side effects, and findings on laboratory tests (performed monthly) were normal.