Knobloch Syndrome in a Large Brazilian Consanguineous Family: Confirmation of Autosomal Recessive Inheritance

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Knobloch syndrome is a rare genetic disorder characterized by high myopia, vitreoretinal degeneration with retinal detachment and occipital cephalocele. The inheritance has been described as autosomal recessive (AR) but in addition to the original report with 5 affected patients [Knobloch and Layer, 1971] only one other family with 2 affected sibs has been described [Czeizel et al., 1992]. We have studied a large consanguineous kindred in wich there are 12 patients with severe ocular alterations associated with a congenital occipital encephalocele, compatible with the diagnosis of Knobloch syndrome. CT scan and MRI performed in one of the patients, allowed a better understanding of the cranial and ocular alterations in this syndrome. The pattern of occurrence in this highly inbred family clearly confirms autosomal recessive inheritance of Knobloch syndrome.

Disease Relapse in Patients with Ocular Manifestations of Wegener Granulomatosis

OPHTHALMOLOGY 1995, 102: 154-160

William J. Power, MD, Alejandro Rodrigues, MD, Renato A. Neves, MD, Leon Lane, MD, C. Stephen Foster, MD

PURPOSE: To evaluate possible factors associated with disease relapse in patients with ocular manifestations of Wegener granulomatosis.

METHODS: Eight patients with ocular manifestations of Wegener granulomatosis were longitudinally followed for a mean period of 34 months. Serial antineutrophil cytoplasmic antibody (ANCA) levels were determined on all patients.

RESULTS: All eight patients had either scleritis alone or scleritis combined with peripheral ulcerative keratitis. Clinical disease remission was achieved in all patients using immunosuppressive chemotherapy. Five patients had subsequent relapse after treatment withdrawal. The serum ANCA level had failed to revert to normal during remission in four of the five patients who had relapses. The ANCA levels for all three patients who remained in remission without therapy converted to normal. There was no significant difference in the initial serun ANCA level ($P=0.35$) or the mean cumulative cyclophosphamide dose ($P=0.13$) between those who had a relapse and those who did not.

CONCLUSION: Failure of ANCA titers to revert to normal levels may be associated with the potential for relapse in the patients with ocular manisfestations of Wegener granulomatosis.

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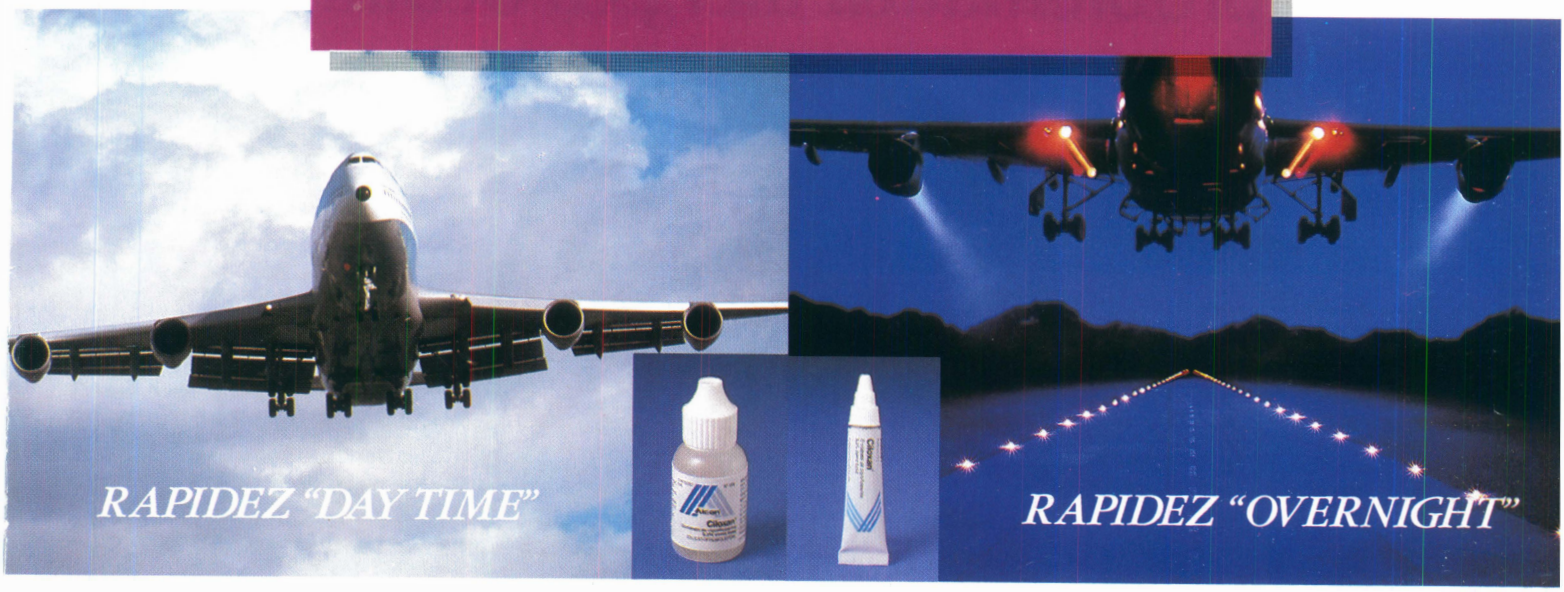

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