Genet. Res., Camb. (1966), 8, *pp.* 377–378
Printed in Great Britain

SHORT PAPER

Identity of mutant genes 'Shrivelled' and cataracta congenita subcapsularis in the mouse

By A. C. VERRUSIO and F. C. FRASER

Human Genetics Sector, Department of Genetics, McGill University, Montreal, Canada

(*Received* 22 *July* 1966)

In 1962 Fraser and Schabtach described an autosomal mutant gene, with intermediate dominance, which caused degeneration of the fibres of the ocular lens, followed by cataract. It was first observed in the inbred A/Jax mouse strain, and is still maintained on this genetic background. The mutant, 'Shrivelled', resembled in many ways a mutant described as cataracta congenita subcapsularis (Cat) by Paget & Baumgartner-Gamauf (1961). As with 'Shrivelled', inheritance is dominant, homozygotes show earlier onset and more rapid progression of the degenerative processes than do heterozygotes, and subcapsular liquefaction of the lenticular material occurs without involving the lens nucleus. In addition, however, Paget and Baumgartner-Gamauf described the passage of large amounts of liquefied material through the lens capsule into the vitreous, intense swelling of the retina, capsular involvement, and persistence of the hyaloid artery.

Because of the similarity between the two mutants, Fraser and Schabtach suggested they might represent different alleles at the same locus, or independent occurrences of the same mutation. The question could not be settled at that time because it was not possible to arrange for crossing the two mutants. Recently, mice carrying the mutant cataracta congenita subcapsularis (Cat) were obtained and the gene was tested for allelism with 'Shrivelled' (Svl).

Reciprocal crosses were made between the two mutant homozygotes. All the F1 offspring showed early onset and rapid progression of the cataract, as in homozygotes. This strongly suggests that the two genes are allelic. Crosses of F1 offspring with unaffected animals produced 164 offspring (79♂, 85♀) all of which had the characteristics of heterozygous mutants. The lack of unaffected offspring is evidence that the genes are alleles or separated by less than 5 cross-over units ($p < 0{\cdot}05$). The phenotypic differences between the mutant in the A/Jax strain and in the Paget stock appear to result from differences in the genetic background. Mutant animals from the Paget stock were crossed to non-mutant A/Jax animals and three back-crosses were made to the A/Jax strain. As the genetic background became more like A/Jax, an increasing number of mutant animals showed the phenotype described for 'Shrivelled'.

It was concluded that Svl and Cat are recurrences of the same mutation. The 'Shrivelled' mutant gene should therefore be designated Cat^{Fr}.

SUMMARY

The cataract-producing mutant gene previously described as 'Shrivelled' appears to be a recurrence of the mutant described by Paget as cataracta congenita subcapsularis, and should henceforth be referred to as Cat^{Fr}.

We should like to thank Dr Oliver Paget and Dr A. Spiegel for providing us with a stock of 'Cat' mice. Financial support of NIH Training Grant 2G-837 is gratefully acknowledged.

REFERENCES

FRASER, F. C. & SCHABTACH, G. (1962). 'Shrivelled': a hereditary degeneration of the lens in the house mouse. *Genet. Res.* **3**, 383–387.

PAGET, O. E. & BAUMGARTNER-GAMAUF, M. (1961). Histologische Untersuchungen an einer dominant erblichen Form einer Cataract bei der Hausmaus. *Zool. Anz.* **166**, 55–69.