

KAPITAŁ LUDZKI
NARODOWA STRATEGIA SPÓJNOŚCI

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Introduction to Human Genetics

Module 1 Mendelian genetics

Mendelian inheritance in humans

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Garrod, Archibald E. 1902: The Incidence of Alkaptonuria. A Study in Chemical Individuality. London, vol. 1, pp. 101-105.

THE INCIDENCE OF ALKAPTONURIA: A STUDY IN CHEMICAL INDIVIDUALITY

ARCHIBALD E. GARROD

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AS THE MORE RECENT views on alkaptonuria have tended to show that the essential feature of that condition is the excretion of homogentisinic acid, to the presence of which substance the special properties of alkapton urine, the darkening with alkalis and on exposure to air, the power of staining fibres deeply, and that of inducing uric acid, are all due. In every case which has been fully investigated since Wolcott and Baumann first isolated and described this acid its presence has been demonstrated and examination of the material from some of the earlier cases also has led to its detection. The second allied alkapton acid, uropigment, has hitherto only been found in the cases investigated by Kirk and in those in association with larger amounts of homogentisinic acid. By the kindness of Dr. B. Kirk I have recently been enabled to examine fresh specimens of the urines of his patients who have now reached adulthood and was able to satisfy myself that at the present time even they are no longer excreting uropigment. After as much of the homogentisinic acid as possible had been allowed to separate out in the test with the small amount of alkapton acid was converted into the

A Urine from a patient with alkaptonuria

1 The specimen on the left, which has been standing for fifteen minutes, shows some darkening at the surface, due to the oxidation of homogentisinic acid.

2 After two hours, the urine is entirely black.

