Archibald Edward Garrod Encyclopedia Article

Archibald Edward Garrod

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1857-1936

British physician who introduced the concept of "inborn errors of metabolism" with his report on alcaptonuria in 1902. The urine of babies with alcaptonuria darkens when exposed to air because it contains homogentisic acid. Individuals with alcaptonuria lack the enzyme homogentisic acid oxidase, which is needed for the complete breakdown of tyrosine. Garrod's analysis of the family trees of affected individuals suggested that alcaptonuria was inherited as a Mendelian recessive gene. Similar patterns were found for other human "chemical abnormalities," including albinism, cystinuria, and pentosuria.