

# An Integrated Modelling Methodology for Estimating Global, Regional, and Country-Specific Incidence and Prevalence of Tay-Sachs Disease at the Subtype Level

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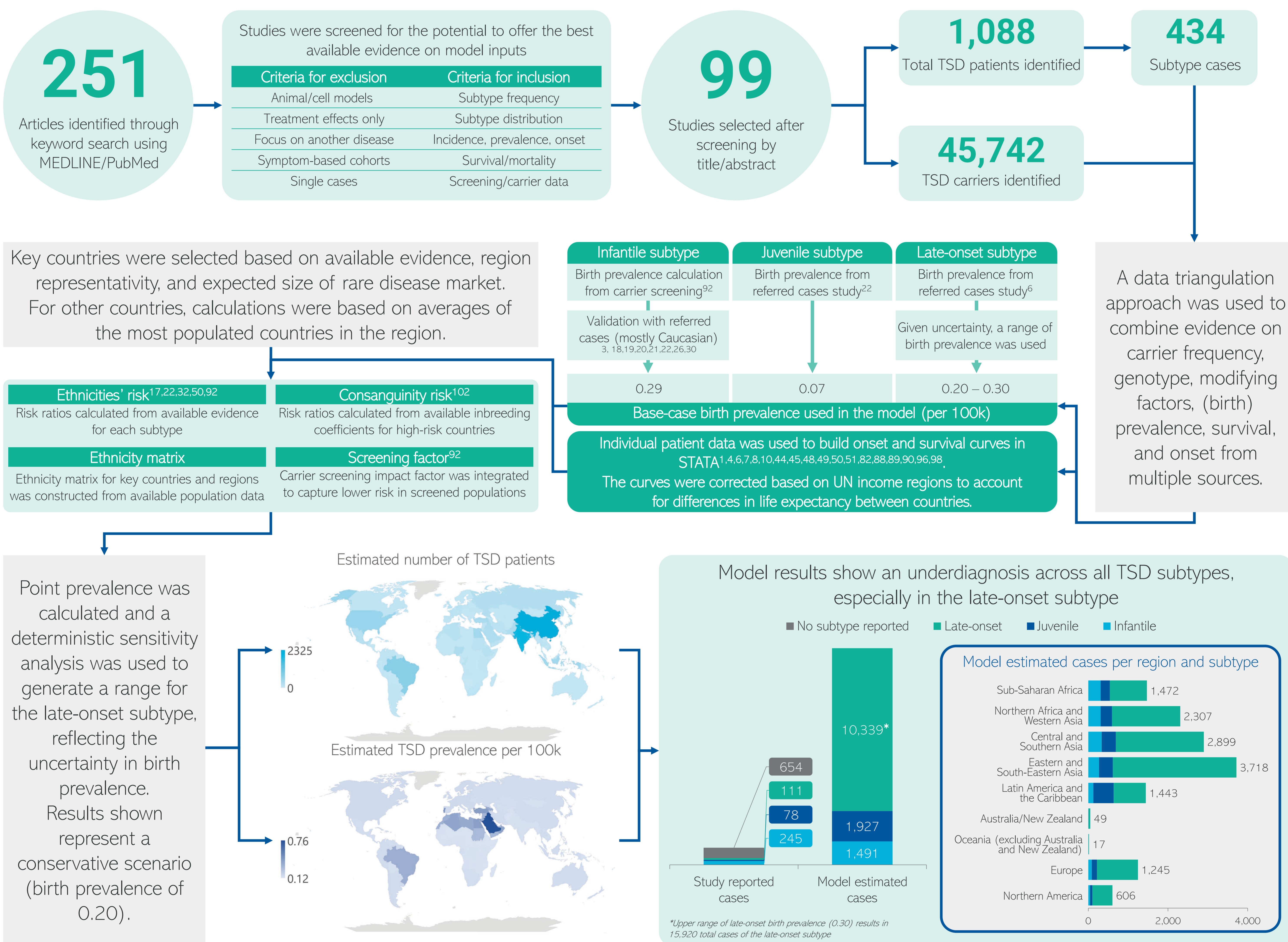
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Tay-Sachs disease (TSD) is a rare autosomal recessive disorder causing progressive neurodegeneration due to HEXA deficiency. Infantile, juvenile, and late-onset subtypes are described based on the age of symptom onset.

Despite genotype variability between certain ethnicities, there are well-established associations between common mutations and subtypes. Limited published evidence is

available on prevalence for large regions of the world, especially for the late-onset subtype, which may be underdiagnosed. This prevents a realistic estimate of the current disease burden. The objective of this study was to develop an epidemiological model of the prevalence and patient pool per TSD subtype at the global, regional, and country levels.



This is the first reported epidemiological model of TSD at the subtype and country level. By combining limited evidence, the estimates better capture the unmet need of the late-onset subtype. This method can be applied in other rare genetic diseases to quantify uncertainty and inform clinical program planning and health economic modelling.

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