Structuring and Analysing Oral and Dental Data from the GenIDA Database in Rare Diseases with Intellectual Disability

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Résumé

Rare genetic disorders associated with intellectual disability, with or without autism spectrum disorders or epilepsy, may present with oral and dental abnormalities that have a significant impact on patients' health and quality of life. However, these manifestations remain insufficiently described due to the rarity of these conditions. Oral and dental features nonetheless represent clinically relevant signs that may contribute to diagnosis, follow-up and patient management, particularly in connection with rare oral and dental diseases monitored by the O-Rares network https://www.o-rares.com/. The international GenIDA database is a participatory registry that collects clinical information through online questionnaires completed by families of patients with rare neurodevelopmental disorders. This approach enables the creation of international and longitudinal cohorts and improves the characterization of the clinical spectrum and natural history of these diseases. However, oral and dental data within GenIDA are often scattered throughout the questionnaire, organized as interdependent questions and frequently described using non-medical language, sometimes in multiple languages. This organization makes direct data exploitation complex and limits their value for clinical research. The objective of this project is to structure oral and dental data extracted from GenIDA in order to make them more readable, usable and reusable. A reproducible methodology is proposed to identify relevant information, group responses related to the same clinical issue and organize the data in a coherent manner. Koolen-de Vries syndrome is used as a case study, as it is sufficiently represented in the GenIDA database, with 257 patients included. It allows the development and testing of a robust methodology intended to be subsequently applied to the analysis of oral and dental data reported by families in other rare conditions.

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Mots-Clés: Base de données, GenIDA, Maladies rares, Anomalies bucco, dentaires, Structuration des données.