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Cancer in people with intellectual disability: lower incidence, later-stage diagnosis – who counts? who cares?

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People with intellectual disability have a unique cancer profile, shaped in part by the heterogeneity of the underlying causes of intellectual disability.[1 2] Those with two of the most common genetic causes of intellectual disability are known to have different cancer risks in comparison to the general population. For example, people with Down syndrome have a higher risk of leukaemia in childhood but a lower risk for many solid tumours, while people with fragile X syndrome (FXS) have an unusually low cancer incidence,[3] with recent evidence also exploring links between fragile X messenger ribonucleoprotein, which is absent or mutated in FXS and glioblastoma development.[4] Additionally, the typically low socioeconomic position and the significant influence of social determinants of health in this population likely increase their exposure to known cancer risks (eg, occupational hazards, carcinogenic exposure and adverse lifestyle factors). Together, these influences may contribute, in part, to the cancer burden experienced by people with intellectual disability.

In this issue of *BMJ Oncology*, a population-based cohort study by Cuypers *et al* (in press)[5] provides important registry data from the Netherlands on cancer incidence in people with intellectual disability. Their findings show fewer incident cancer cases among individuals with intellectual disability than among those without (51.0 vs 16 104.1 per 10 000 person-years; adjusted OR 0.79 (0.76–0.81)), with cases peaking at younger ages compared to the general population. They also highlight significant variations, including the occurrence of cancers at more advanced stages, a particularly concerning finding. Elevated odds of cancers of unknown primary origin were observed, along with reduced odds of skin cancer. These are stark findings.

This editorial is coauthored by an academic researcher and a parent of a child with intellectual disability—someone who is also experiencing their own cancer journey. This brings a unique real-life perspective to considering the findings. In the coauthor's own personal situation during their cancer journey, they were able to verbalise their symptoms as they experienced changes in visual processing, facial recognition and disorientation. Even though they were able to communicate symptoms and had a strong sense of agency, they experienced long waitlists for scans and delays in screening and access to diagnostics despite the availability of universal healthcare. A similar situation exists for people with intellectual disability, whose deficits with cognitive and adaptive functioning present additional challenges as they are seen as 'invisible'[6] with diagnostic processes complicated by diagnostic overshadowing.[7]

Given these challenges, there is an obvious question when examining the emerging evidence, 'is there truly a lower incidence of cancer in this population?' or 'is it a reflection of the atypical life many people with intellectual disability lead and the systemic health inequalities and inequities that lead to cancer underdiagnosis more broadly?' It is potentially a heightened concern in those who are diagnosed at advanced stages[8] and in those with elevated odds of cancer of unknown primary, observed by Cuypers *et al* (in press).

To date, most studies of cancer incidence and outcomes in this population have been limited by bias and small scale.[9] The current study (in press) additionally highlights diagnostic challenges, including potential diagnostic delays and incomplete staging. For cancers of unknown primary origin, higher incidence was found among people living independently—a group potentially facing greater barriers accessing healthcare. This is a critically important finding for future research, along with the observation that

individuals living independently are at greater risk for digestive, respiratory and female genital cancers—potentially reflecting greater socioeconomic disadvantage.

The study calls for strategies and awareness initiatives across public health, long-term care and cancer programmes, stimulating the need to consider an additional question which the authors touch upon: '*how effective are cancer screening programmes for people with intellectual disability, especially given the earlier age of cancer onset observed here in this population?*' Increasingly, evidence explores participation, barriers and facilitators[10] rather than outcomes as key barriers, and this is where attention must be focused. These findings suggest a need for tailored screening programmes that are accessible, particularly for independent living adults with intellectual disability. Across Europe, there is growing recognition of the inadequacies in cancer care for this population.[11] The pressing need is to improve diagnostic and cancer care pathways for people with intellectual disability. This includes lowering the threshold for investigating potential cancer symptoms and enhancing health surveillance and ensuring that people with intellectual disability—both those known and unknown (eg, the 'invisible majority') to services—are included in national cancer control strategies, where they are currently overlooked.

To advance this field, the ability to combine and analyse multiple datasets is crucial. However, as is the case in much research involving people with intellectual disability, identifying intellectual disability at the individual level remains exceptionally difficult in large population studies. This undermines efforts to explore causal pathways to tumour development. From this perspective, the authors' report—*that despite lower overall cancer incidence, there is higher late-stage diagnosis with unique subtypes and sociodemographic patterns*—serves as both a wake-up call and a roadmap for future research and practice.

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