Review

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Colorectal Cancer in People with Intellectual Disabilities

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Keywords
Colon cancer · Rectal cancer · Cancer screening · Intellectual disability · Learning disability · Treatment difficulties

Abstract
People with intellectual disabilities (PWIDs) are now living longer; thus, the incidence of cancer within this population is increasing. Available data indicate an excess of digestive tract cancers in PWIDs, but colorectal cancer has rarely been specifically studied and has not been extensively reviewed. This is despite risk factors such as being overweight, obesity, and lack of exercise being more frequent in PWIDs. In this article, we examine the literature on the frequency, screening, and treatment of colorectal cancer in PWIDs by assessing 4 databases, Medline, EBSCO-CINHL, ASSIA, and PsychLIT, from 1970 to February 2017. Findings indicate that the frequency trends slightly higher than that found in the general population. Screening presents a unique opportunity to discover early colorectal cancer, but is underused in PWIDs compared to the general population. Furthermore, the clinical presentation is frequently masked, particularly by challenging behaviours, and colorectal cancer is therefore often diagnosed late, making treatment difficult due to the advanced stage of these tumours. To improve the care of PWIDs, we need more resources to support them and their caregivers, and to increase awareness of the risk factors and signs and symptoms of colorectal cancer.

Introduction

People with intellectual disabilities (PWIDs) are living longer and, as with the wider population, increasingly experiencing serious illnesses such as cardiovascular and respiratory diseases [1]. However, cancer is an age-related disease and has not received the same attention as other medical problems in this population [2, 3]. Evidence on overall cancer incidence in PWIDs is limited due to the lack of reliable epidemiological data [4] but is currently estimated to be as frequent as in the general population [5, 6]. On the other hand, the pattern of cancer in PWIDs differs from that of the general population and suggests a higher risk for gastrointestinal cancers [5, 7, 8]. Colorectal cancer is 1 of the 4 most common cancers in the world [9]. It accounts for nearly 10% of the global incidence of cancer worldwide and constitutes the third most com-

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mon cancer in men and the second most common in women [10], but its risk in PWIDs remains relatively unknown.

Recognized risk factors for colorectal cancer are being overweight and lack of physical activity, as well as consumption of red meat, processed meats, and alcoholic beverages [10]. PWIDs are more prone to being overweight with the prevalence of overweight and obesity estimated at around 30% [11, 12]. Being overweight is an important risk factor for colorectal cancer, with an estimated increased risk of 41% for a body mass index (BMI) >30 compared to a BMI <23 [13]. Additionally, PWIDs are less involved in physical activity; a recent Australian study indicated that 60.3% of 68 adult PWIDs did not reach the national physical activity guidelines [14]. Globally, PWIDs are low consumers of alcohol compared to the general population. However, they have a moderate risk of malnutrition, with 17.6% of them in the high-risk category [14]. Constipation, which is clearly more prevalent in children and adults with IDs [15], is currently not considered a risk factor for colon cancer. Globally, being overweight, obese, inactive, or having poor nutrition, are well-documented factors which increase the risk for colorectal cancer within PWIDs. These characteristics suggest a potential increased risk for colorectal cancer which might be prevented by modifying and reducing obesity and increasing physical exercise. However more research is needed to clarify these findings [12, 16].

When discovered early, particularly by screening [17], colorectal cancer has a lower mortality rate than many cancers. However, cancers in PWIDs are often discovered late [18]. Screening for colorectal cancer uses either a faecal occult blood test (FOBT) or, more recently, a faecal immunology test (FIT), both of which look for blood in the faeces, while colonoscopy or sigmoidoscopy allows a direct visualization of the colon. Colorectal cancer screening is only a recent national imperative: for example, beginning in France in 2009 and in the UK in 2010. In the UK, screening begins at 50 years of age in Scotland or 60 years of age in England, Wales, and Northern Ireland, with the FOBT/FIT test being done every 2 years until 74 years of age. In France the screening begins at 50 years until 74 years of age. To date, few data are available to assess the impact of such screening programmes.

Colonoscopy is the major method to confirm diagnosis and collect tumour samples in cases where colorectal cancer is suspected. The procedure allows a visual evaluation of the entire colon and rectum and permits the biopsy of anomalies. In some countries, and in particular the USA, colonoscopy is a first-line screening method, whereas in other countries, such as the UK and France, the primary test is an FOBT in the global population without familial history of colorectal cancer or adenomas. If blood is detected, a second FOBT may be required, and colonoscopy undertaken if the FOBT is positive. Once a diagnosis is made, colonoscopy is needed for detecting and removing lesions and to confirm the diagnosis. This entails administering a preparation, consisting of various agents: stimulants, enemas, osmotic agents, and polyethylene glycol-based solutions to clean the lower digestive tract. Furthermore, an individual must be able to consume at least 4 L of clear liquid. The quality of the colonoscopy is directly related to the quality of the preparation of the colon [19]. This article aims to review the literature on colorectal cancer to explore the evidence to inform health and social care practice.

Table 1. Main terms to identify material on learning disability

Intellectual disability(ies)
Developmental disability(ies)
Mental retardation
Learning disability(ies)
Mongolism/mongoloid
Learning difficulty(ies)
Mental handicap

Methods

An integrative review process was chosen, as they offer opportunities to critique and synthesise literature from a diverse range of primary research to enable the current state of knowledge to be examined, especially when there is limited literature [20–22]. The aim of the literature review was to explore the literature on colorectal cancer and bowel screening in PWID.

The review of the literature was not restricted by date and incorporated literature from 1970 to 2017. Careful consideration was given to search terms, especially as there is no universally accepted term to describe PWIDs. Consequently, intellectual disability in all its forms was used (see Table 1), alongside colorectal neoplasms or bowel cancer or bowel neoplasm or colon cancer or colorectal cancer, as these terms embraced the nomenclature for colorectal cancer. Screening was also added to the search terms since bowel screening was in place in some countries. Four databases (Medline, ASSIA, EBSCO-CINHL, and PsychLIT) were used for this review as these incorporated medical databases related to PWIDs [21, 22]. The terms were used in combination and were adjusted to suit the terminology of the database searched. To capture the "grey literature," and ensure total coverage of the data, searches of professional organizations (e.g., Foundation for People with Learning Disabilities), government bodies (e.g., Learning Disabilities Observatory, England), and special interest groups were performed

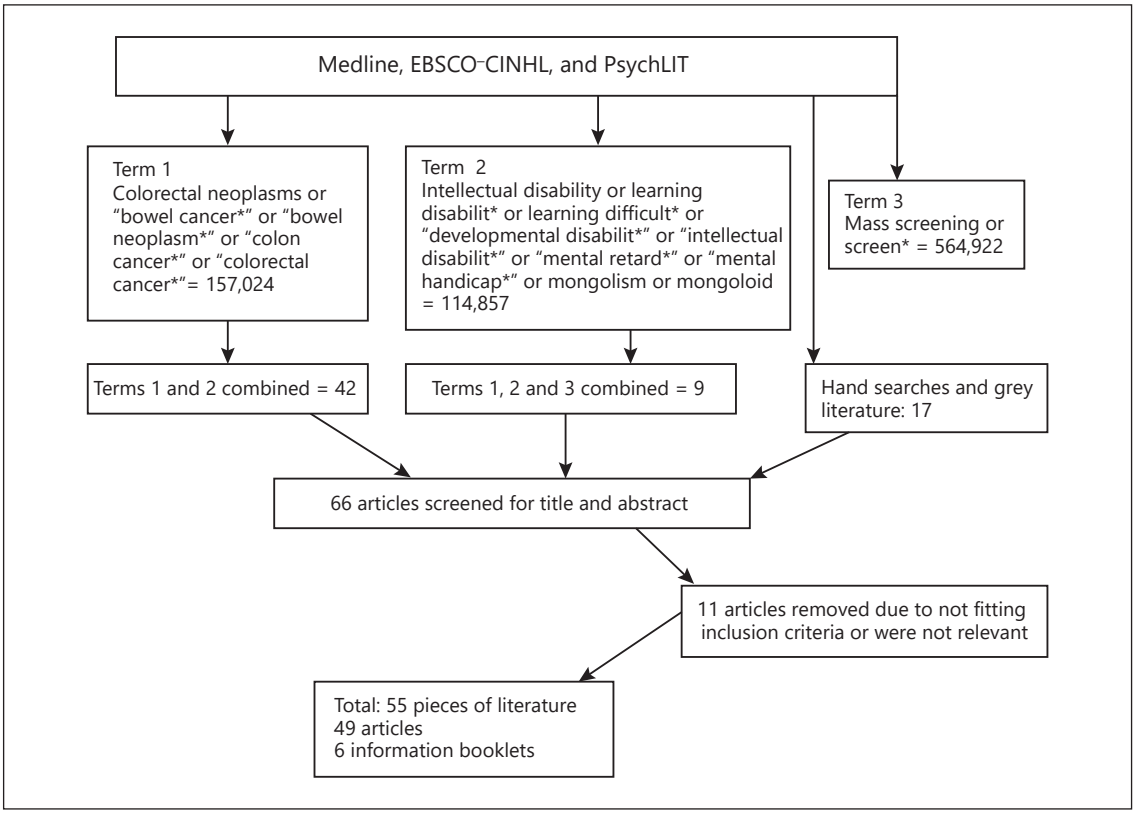

Fig. 1. The number of papers relating to PWIDs and colon screening and colon cancer.

[23]. Hand searches were also undertaken by reviewing the reference lists of the articles retrieved by the database searches. It should be noted that due to the limited literature available all types of studies and interventions were considered [22]. In addition, rather than review the empirical literature, the authors wanted to update current knowledge on colon cancer and bowel screening; hence, articles were included that had an educational slant as well as information booklets aimed at supporting the PWID and the caregiver. The number of articles retrieved by each database and search term is shown in Figure 1.

The inclusion criteria consisted of all studies being written in English and relating specifically to colon/bowel screening/cancer screening in PWID. It should be noted that 2 articles, 1 French and 1 Japanese, were included due to their relevance and availability of translations. Exclusion criteria extended to other cancers/cancer screening. Titles and abstracts were screened to ensure adherence to the inclusion criteria and eligibility to the final literature review. In total, this review contains 55 pieces of literature: 49 articles and 6 information booklets are reported. Table 2 indicates the main studies with the exception of case studies and information booklets, to avoid repetition of data extraction. The CASP [24] and Whittemore and Knafl [22] were drawn on for the data extraction criteria for Table 2; whilst the Joanna Briggs Institute Levels of Evidence [25] were used for quality appraisal, where 1 was the highest and 5 the lowest. The articles found relate to 4 areas: population and mortality studies, institutional population, genetic conditions, and clinical presentation and treatment in PWID. The focus of the review is on colon cancer and screening and treatment to raise the awareness of colorectal cancer in health of social care practitioners.

Results

Population and Mortality Studies

There are a number of studies exploring cancer in this population, which are divided into epidemiological studies and those of PWIDs living in institutions, with their family, or in the community. Two epidemiological and 2 mortality studies are available. The first, conducted in Finland on 2,173 PWIDs (1,090 men and 1,083 women) between 1967 and 1997, was a data linkage study, linking a register of PWIDs (which determined their level of ID) to a cancer registry. These individuals were then followed up. Findings for sex were not reported separately but a standardized incidence ratio (SIR) of 0.9 for colon and

Table 2. Summary of studies on colorectal cancer in PWIDs

Author	Quality	Country	Participants	Type of study	Aims	Findings
Bourgarel et al. [32] (2016)	3a	France	9,104 PWIDs	Scrutiny of records for colorectal cancer 2009–2010	Evaluate the frequency of colorectal cancer and participation in screening among PWIDs living in institutions	1,519 records identified 3 colorectal cancers; colorectal cancer screening for 24% of PWIDs aged 50–74 years compared to 34% in the general French population Colonoscopies due to positive screening test: 2.4%, similar to 2.7% in the general population
Bowler and Nash [59] (2015)	4b	UK	239 PWIDs	Intervention study: training pack and system to flag up non-responders	Increase uptake of bowel screening in PWIDs	193/239 took part, 117 undertook the test; increased uptake by 14%; 18 needed further investigation
Cooke [29] (1997)	3e	UK	5,301 in-patients, of which 213 PWIDs died	Review of hospital records from 1986 to 1995	To identify death rate from cancer in an institutionalised population of PWIDs	4 colon cancer deaths; cancer frequency is unclear but appeared to be increasing but due to changes within the institutional population was now falling; cancer remains less frequent than for the general population
Deroche et al. [56] (2017)	3b	USA	7,778 PWIDs and 35,036 non-IDs	Survey of hospital records between 2000 and 2009 linking South Carolina medicaid, state health plan, and payers of hospital billing	Aim to see if any of 3 disability groups (PWIDs, blindness, spinal cord injury) received colorectal cancer screening compared to those without a disability	PWIDs had lower compliance for changes: 34.32%, compared to 48.48% in other groups 61.88% of PWIDs never received screening compared with the other case groups; colonoscopy more prevalent, screening 29.53%
Diab and Johnston [55] (2004)	3b	USA	Disabled people: 41,106 (1998) and 59,939 (2000)	Analysis of data from the Behavioural Risk Factor Surveillance System 1998–2000 and nationwide telephone survey of 50 states, district of Columbia and Puerto Rico	Examine relationships between level of disability and receipt of preventive health services	Only 1 state reported on colorectal screening; women had the test less frequently, severely disabled people had lower uptake
Fischer et al. [19] (2012)	4b	USA	47 PWIDs, 31 male 16 female, and 40 people in the general population	Quality review of colonoscopies performed from 2002 to 2010 on PWIDs	Compare colonoscopy in PWIDs with examinations performed in a random group of 40 adults without IDs	Higher rate of colonoscopy preparation failure and failed examinations in PWIDs; preparation complications were also noted
Glover et al. [8] (2017)	3e	UK	All patients registered between 2010 and 2014; 664 PWID deaths, 371 male, 293 female	Review of cancer mortality records of PWIDs from 2010 to 2014	Identify the rates and patterns of mortality of people living in England and identified by their GP as having IDs, in relation to age, sex and causes of death and compared to the general population	Male deaths 55.9% and female deaths 44.1%; colorectal cancer deaths account for the largest subgroup of cancer deaths
Glover et al. [53] (2014)	3b	UK	Not known	Review of 94–101 Learning Disability Partnership Boards that provided data for bowel cancer screening for PWIDs and for the general population	Present information from the Joint Health and Social Care Self-Assessment Framework (JHSCSAF) on reported rates of cervical cancer, breast cancer, and bowel cancer screening for PWIDs in England in 2012/2013 compared to screening rates for the general population	Boards reported data on bowel cancer screening: 59%; lower up-take for PWIDs than in general: 28.1 vs. 40.5%; geographical coverage variations noted

Table 2 (continued)

Author	Quality	Country	Participants	Type of study	Aims	Findings
Hasle et al. [46] (1996)	3b	Denmark	597 females with TS	Women with TS linked to the Danish Cancer Registry compared to cancer rates for Danish women	Cancer incidence of a cohort of women with TS	Colon cancer was observed in 5 patients; suspicion of increased colon cancer risk
Iacono and Sutherland [57] (2006)	4b	Australia	2,540 adults with developmental disabilities	Survey of carers looking at health screening in the last 12 months in PWIDs	Explore participation of health screening activities in PWIDs	Response rate 26% (659) in PWIDs; higher participation in screening if registered with services
Jancar and Speller [64] (1994)	3e	UK	32 PWIDs compared with 32 without IDs	Retrospective review of hospital register of deaths from intestinal obstruction 1940–1989	Explore fatal intestinal obstruction in PWIDs	Among 1,354 deaths, intestinal obstruction accounted for 32 (18 female, 14 male); colon cancer: 2.2% – 6 times the national rate, 2 deaths from cancer were both female
Jancar [28] (1990)	3e	UK	302 PWIDs	Review of patient records from 1976 to 1985	Identify death rate from cancer in an institutionalised population of PWIDs	53 died from cancer; 4 women died of colon cancer; notes an increase of cancer
Jancar and Jancar [7] (1977)	3e	UK	1,125 PWIDs, 536 male and 589 female	Review of patient records from 1936 to 1975	Identify death from cancer in PWIDs	5 deaths from colon cancer (6%; 1 male, 4 female); 5 deaths from rectal cancer (6%; 4 male, 1 female) Increased incidence of cancers (17% vs. 13%) in PWIDs
Joel and Marcellino [58] (2016)	4b	UK	238 and 12 PWIDs	Review of screening records from the local Direct Enhanced Service (DES) register and community nurse intervention study	To improve bowel screening uptake in PWIDs in Bristol	FOB tests complete in 8% of PWIDs Intervention FOB on 12 PWIDs – 11 negative, 1 positive Identified barriers and gave education for carers and clients
Kiani et al. [26] (2010)	3c	UK	503 PWIDs with moderate and profound IDs	Review of records from Leicestershire ID data base from 1993 to 2006	Compare mortality from cancer in PWIDs with that found in the general population	17% of deaths during the 14-year study; 47 people (9%) had died from cancer; no significant difference in cancer-specific mortality in PWIDs compared with the general population
Lauer [27] (2016)	3b	USA	847 PWIDs	Reviewed mortality records of PWID service users of the Massachusetts Department of Developmental Services (DDS) from 2012 to 2013	Explore mortality in PWIDs	Cancer was the second leading cause of death, resulting in 13.4–13.7% of deaths each year; colon was one of the main causes of cancer deaths (18/847)
Marriott et al. [61] (2014)	5c	UK	Nil	Awareness paper/education	Explore 5 English screening programmes and suggest ways to improve uptake of cancer screening in PWIDs	Uptake lower for bowel screening in PWIDs – 25.8% compared to 39.4% of the general population
Marriott et al. [52] (2015)	5c	UK	Nil	Educational review of the role of screening liaison nurses	Describe the role of the screening liaison nurses for adult PWIDs employed by Peninsula Community Health	Limited evidence for bowel screening programme, bowel cancer screening programme, and low up-take
Osborn et al. [51] (2012)	3b	UK	6,566 PWIDs	Retrospective review of Health Improvement Network (THIN), a primary care research database	Determine whether rates of cancer screening differed in PWIDs compared to people without IDs in primary care	PWIDs less likely to have a record of screening for bowel cancer than the general population

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Table 2 (continued)

Author	Quality	Country	Participants	Type of study	Aims	Findings
Ouellette-Kuntz et al. [54] (2015)	3b	Canada	15,791 PWLDs, 791,792 without IDs	Review of health and social services datasets for Ontario residents aged 50–64 years with ID and the general population	Examine participation in colorectal cancer screening among PWIDs	FOB and participating in bowel screening were 32 and 46% lower in PWIDs; age, female, greater use of health care services, and registered with a GP were factors associated with bowel screening uptake in PWIDs
Patja et al. [5] (2001)	1a	Finland	1,090 men and 1,083 women	Linked a population cohort on ID register and cancer registries from 1967 to 1997	Assess risk of neoplasms in people with intellectual disabilities	Incidence of cancer similar to the general population; bowel cancer slightly lower
Read et al. [66] (2016)	5c	UK	6 PWIDs 1 advocate 2 specialist nurses	Discussion paper	Explore why PWIDs may be reluctant to access bowel cancer screening	Identified barriers and need for more education, better information and support
Read and Latham [76] (2009)	5c	UK	Nil	Clinical skills and review of current practice	Improve practice	Identified areas of ensuring good practice, e.g., communication, consent, planning
Satgé et al. [31] (2010)	3e	Israel	450 PWIDs	Review of deaths from 1991–1997 in residential centres	Explore deaths in PWIDs in residential settings	Colorectal cancer accounted for 10% of cancer deaths; similar trends to the general population
Satgé et al. [49] (2006)	5a	France	Nil	Review of the literature	Review of the literature on digestive track tumours in DS	Lower incidence; found 13 benign tumours and 127 cancers in 1 foetus, 8 children and 131 adults with DS; people with DS develop fewer digestive neoplasms compared to PWIDs
Sullivan et al. [6] (2004)	1a	Australia	9,409 PWIDs, 5,490 male, 3,919 female	Linked IDs database and cancer registry from 1982 to 2001	Determine the incidence of cancer in PWIDs	Colorectal cancer increased in males (1.39) and in females (3.10)
Sund et al. [50] (2009)	3e	Finland	302 individuals with FXS	Finnish registry of PWIDs from 1982–1986; follow-up on cancer incidence from Finnish Cancer Registry 1982–2005	Is there a reduced incidence of cancer in people with FXS?	11 reported cancers during the mean follow-up, but 1 digestive and 1 unspecified – hence unclear
Tuffrey-Wijne et al. [74] (2009)	4c	UK	13 PWIDs	Ethnographic study	To provide insight into the experiences and needs of PWIDs who have cancer	1 patient had rectal cancer; identified barriers, e.g., negotiating health care system, delayed cancer diagnosis, and a lack of treatment options; PWIDs did not understand their illness or the implications
Uno [33] (1996)	3b	Japan	134 PWID patients in institutions	Mass colonoscopy screenings study	Initiated to determine colorectal disease rates among mentally retarded subjects and whether they are at risk of developing colorectal cancer	Polyps were found in 24 patients; no cases of APS were detected; PWIDs were not at a high risk of developing either APS or colon cancer
Wilkinson et al. [62] (2007)	5a	USA	Nil	Literature review on screening tests	Explore screening recommendations for common preventable conditions using the US Preventative Service Task Force guidelines	Colon cancer was slightly more prevalent in PWIDs, rates of adenomatous polyps in institutionalized PWIDs approximated the general population; constipation commonly masks colon cancer symptoms; recommends following the guidelines for colon cancer screening

APS, adenomatous polyposis syndrome; DS, Down syndrome; FOB, faecal occult blood test; FXS, Fragile-X syndrome; Nil, no participants; PWIDs, people with intellectual disabilities; PWLD, people with learning disabilities; TS, Turner syndrome.

rectal cancer in both sexes was reported and international classifications of disease were used as the diagnostic criteria. Eight patients with colon cancer were observed while 9.3 were expected (SIR 95% CI 0.4–1.7), and 6 patients with rectal cancers were observed while 7 were expected (SIR 95% CI 0.3–1.9) [5]. The second study, conducted in Western Australia on 9,409 PWIDs between 1982 and 2001, was another data linkage study, linking an intellectual disability database to a cancer registry. There were 14 male patients with colon or rectal cancers when 9.38 were expected, indicating an increased SIR of 1.39 (95% CI 0.14–2.37). Nine cases were observed for women when 2.9 were expected, indicating an increased SIR of 3.10 (95% CI 1.42–5.88) [6]. Both authors noted the small sample size as a limitation.

A mortality study conducted in the UK reviewed records from the Leicestershire Intellectual Disability Database between 1993 and 2006. It included patients with moderate to profound intellectual disabilities living mainly in institutions but also with their family or independently in the community [26]. From 2,995 adults, there were 47 cancer deaths, giving a standardized mortality ratio (SMR) for all cancers of 0.94. Of these, 4 were colorectal cancers. Another UK-based study examined cancer mortality records for PWIDs between April 2010 and March 2014 [8]. Of the 664 deaths (371 male, 293 female) 13% ($n = 87$) of mortalities were cancer related, with cancer of the digestive tract being the largest subgroup identified (32 of the 87 observed deaths). Sixteen of these deaths were related to colon and rectal cancer, while 6.8 were expected (SMR for men 2.68, SMR for women 1.85). However, the authors noted that recognition thresholds vary between countries and regions.

The final study reviewed mortality in PWIDs aged 18 years and older among service users of the Massachusetts Department of Developmental Services (DDS) over a 2-year period (2012–2013) [27]. The findings revealed that cancer was the second leading cause of death, resulting in 13.4–13.7% of deaths each year. Colon cancer was one of the main causes of cancer deaths ($n = 18/847$). This study, however, focused on those registered with services and may have missed those who have no contact with services. Although the above studies are seen as offering a high-quality evidence base, there are some caveats. Data linkage work is not able to offer total coverage of the population as there will always be missing or incorrect data. Not all data were reported by incidence by age whilst none of the studies were total population studies for the respective countries.

Institutional Population

More data are available for PWIDs living in institutions than for the general population of PWIDs. Data from 3 consecutive British studies by the Stoke Park group of hospitals used cause of death of their residents as documented in hospital records over a 65-year period (1930–1995). However, it should be noted that these studies did not use the international classification of diseases, did not report incidence by age, and standardised mortality measures were not employed. Results suggested an increased frequency of colon and rectal cancer in PWIDs compared to the general population. Among 61 PWIDs who died from cancer during the 40-year period from 1936 to 1975, 10 (12%) succumbed to colon ($n = 5$; 1 male, 4 female) and rectal ($n = 5$; 4 male, 1 female) malignancies. The authors emphasized that, compared to the general population, relatively more PWIDs die from these cancers (17 vs. 13%) [7]. Between 1976 and 1985, 53 PWIDs died from cancer, 31 of them (58.5%) from cancer of the gastrointestinal tract (4 women died of colon cancer) [28]. Finally, from 1986 to 1995, of 213 deaths, 7 of the 27 cancer deaths (25.9%) were related to tumours located in the colon ($n = 4$) or in the rectum ($n = 3$) [29]. Another study from the group estimated serum cholesterol levels from 496 PWIDs (258 female, 238 male). They found 23 colon cancers (12 male, 11 female) and that their serum cholesterol levels were significantly lower than those who died from other causes [30]. No other study has looked at this or found similar associations. The Stoke Park studies are useful, but have limitations; for example, the diagnostic criteria were not explicit and standardised mortality measures were not employed. An Israeli study reviewed 450 records of death in 53 residential centres between 1991 and 2005. The findings revealed 74 cancer deaths from people with mild, moderate, or severe IDs living in institutions. Colorectal cancers accounted for 9.4% of these cancer deaths [31]. A limitation was that those who died at home or in a foster family were not included in this study. A recent study in France used data from a large survey of 1,519 institutions, finding 3 cases of colorectal cancer among a total of 32 cancers, while 1 colorectal cancer was expected, suggesting a non-significant increase with an associated SIR of 3 (95% CI 0.6–8.76) [32].

A Japanese study found PWIDs not to be at more risk of adenomatous polyposis syndrome (APS) or colon cancer [33]. Colonoscopy screening was undertaken in an institution of 134 patients (age range 32–69 years). Although polyps were found in 24 patients (17.9%), and abnormal fixation, haemorrhoids or melanosis coli in 2 pa-

tients (1.5%), respectively, there were no cases of APS or colon cancer detected. In a review of the literature [34], a higher frequency of colon cancer in PWIDs living in institutions was observed compared to those who lived with their family or in the community. The authors indicated that patients with moderate to severe IDs tend to live in institutions and are more prone to colorectal cancer than patients with mild IDs, who often live in the community. However, the data presented in this review did not provide a precise answer on the degree of cancer risk according to the level of ID and were insufficient to evaluate differences between women and men. It should be noted that no particular histological type was described for the colon cancers reported in PWIDs within these studies, which is significant in order to determine the stage and type of cancers identified.

Genetic Conditions

There are particular genetic conditions which carry increased risk factors for these malignancies. Patients with sub-microscopic or a larger 5q deletion, which includes the *APC* gene, show features of familial adenomatous polyposis (FAP) syndrome. This is defined by the occurrence of more than 100 colon polyps and has a very high risk of colon cancer occurring earlier than in the general population. Some FAP patients also present mild intellectual disability and slight dysmorphism, and 7 such patients (2 male, 5 female) have been reported in the literature as case studies. As expected, some developed a carcinoma before the age of 50 years [35–39] or presented polyps with high-grade dysplasia, a clear indicator of a pre-neoplastic lesion [35, 40]. Other case studies have revealed at least 3 patients (2 male, 1 female) with a small deletion of a chromosome harbouring genes implicated in hereditary non-polyposis colorectal cancer (2p, 3p, 7p) who also had IDs and developed colon cancer, 2 of them early in life [41–43]. For example, a male patient with a mutation in the *PMS2* gene was diagnosed with a rectal carcinoma aged 14 years [44], whilst a man with mild to moderate ID due to Williams syndrome and Lynch syndrome (which increases the risk for colon neoplasia) developed a colorectal cancer aged 37 years [45]. In a Danish cohort of 597 women with Turner syndrome, those who presented with mild IDs were found to be at a higher risk of colon cancer compared to women in the general population [46]. The authors suggested that there was almost complete follow-up. Two colorectal carcinoma cases have been reported in a neonate and a young adult with trisomy 13 [47], and such tumours were described in a female patient with tuberous sclerosis [48]. A literature review on individuals with Down syndrome and digestive cancer identified 39 colon cancers, but overall people with Down syndrome develop fewer digestive neoplasms compared to PWIDs [49]. In a Finnish data linkage study from a cancer registry of PWIDs (1982–1986), 302 people with fragile-X syndrome were followed-up on cancer incidence until 2005 [50]. The findings were unclear as, although 11 cancers were reported, only 1 cancer was coded as digestive and another was unspecified, hence it was unclear whether these were related to colon cancer. Either way, the last 2 studies identified that colorectal cancers appear rarer compared to the general population and the occurrence remains less well established [49–50].

The evidence here is mainly reliant on case studies, which are not themselves representative of the population. Despite this, they sensitise researchers towards some of the risks within this population.

Screening

Given that screening is a relatively recent development, there is a dearth of research. A survey of general practitioners in the UK revealed a very low participation rate of 5.92% among 6,566 PWIDs and produced an adjusted incidence rate ratio of 0.86 compared to the incidence in non-disabled adults. However, the study was conducted shortly after the commencement of screening, and the participation rate in the general population was also found to be low [51]. More recently, work in the UK collecting data through the Joint Health and Social Care Self-Assessment Framework (JHSCSAF), a mechanism within Public Health England of collecting data on PWIDs in England, suggested that, of all screening programmes in the UK, colorectal screening had the highest uptake in PWIDs, with an uptake of 41.6% in PWIDs, lower than the 50.4% uptake in the general population [52]. This is in contrast with other work in the UK published a year earlier and again obtained from the JHSCSAF, which suggested lower rates for bowel screening in PWIDs (28.1%) compared to the general population (40.5%) [53]. Canadian work comparing 15,791 PWIDs and 791,792 without IDs from administrative health and social services datasets for Ontario residents aged between 50 and 64 years has suggested lower colorectal cancer screening rates for PWIDs (32%) than in the general population. Of those PWIDs, 18.5% had undergone a FOBT compared to 26.4% of the non-disabled population between 2008 and 2009 [54]. It must again be remembered that the population in these studies only reflects those in contact with services or appearing on social service records. In one study [54] the authors acknowledged the limited availability and access to data.

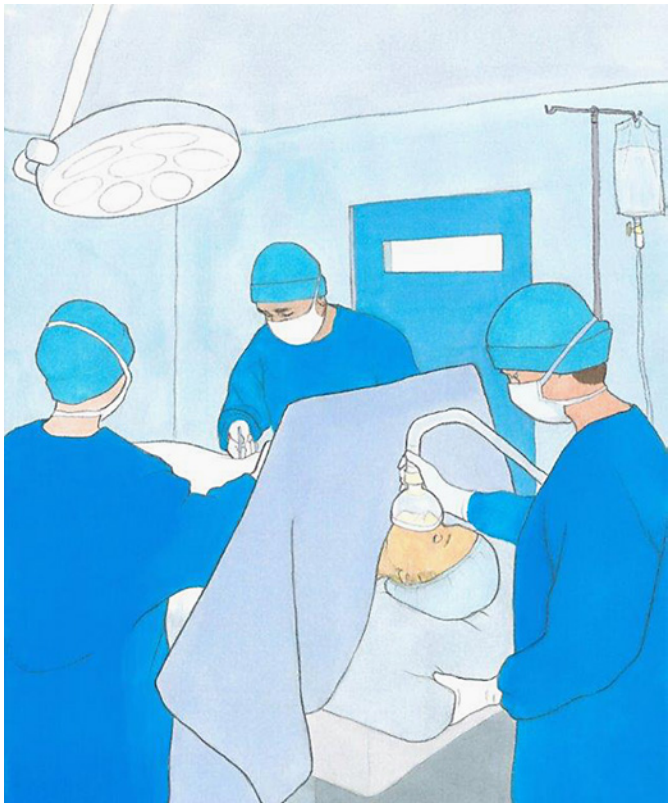
In France, a study of 653 PWIDs aged 50–74 years living in institutions found that the participation rate was 10% lower, with only 24% undergoing FOB testing compared to 34% in the general population [32]. In the USA, a review of participation in proctoscopy/sigmoidoscopy/colonoscopy and FOBT in 101,045 people with disabilities from 1998 and 2000 found it was lower in individuals with various disabilities compared to the general population [55]. The problem with this paper is that the types of disabilities were not documented separately and again full coverage may not have been attained. Another American study undertook a survey of hospital records between 2000 and 2009 and identified 7,778 PWIDs, 2,938 blind/low vision patients, 7,126 spinal cord injury patients, and 35,036 without IDs from their billing records. These indicated that only 34% of adults with IDs (vs. 48% of adults without IDs) aged 50–75 years adhered to colorectal cancer screening over the 10-year period [56]. These results must be reported with caution, firstly because all other disabilities were included in those without IDs. Furthermore, hospital billing records are not an accurate means of identifying compliance with screening due to administrative coding systems being subject to human error, and in this case ICD-9-CM codes for disabilities were not recorded if different from the underlying condition, whilst some participants may have been screened prior to the study and coded as non-compliant. In Australia, surveys were distributed to 2,540 PWIDs and their immediate support persons [57]. Of those returned, 51 were rejected due to incomplete data and 659 were analysed, giving a 26% response rate. Participation in FOBT was 7.5% for PWIDs aged 18–76 years, but data for people aged 51–74 years was not reported. The findings, however, have several weaknesses; for example, the non-responders may be a different population, surveys were only sent to those in contact with services, and there is always the possibility of recall bias or interpretation issues of the questions posed.

Work in terms of facilitating bowel screening is limited and comes mainly from the UK. One study reviewed records from a local Direct Enhanced Service register and identified 238 clients aged 60–74 years who were eligible for FOB screening, of whom 19 (8%) had participated in the screening [58]. Information was collected from 31 of 47 GP practices, with no indication of why all practices were not consulted. Another study reported within the paper used health promotional materials aimed at increasing the uptake of FOB screening. Community nurses within the region were asked to support 1 PWID to undertake the test and 12 further clients were supported with successful screening, with 1 test being positive. The studies were not clearly documented, making evaluation difficult. Another study increased uptake by 14% through the community learning disability team developing accessible screening letters, a training pack for PWIDs and carers, and a system to flag up non-responders [59]. Of the 239 people eligible for bowel screening, 36 declined to participate and 6 were too unwell. Bowel screening was discussed with 193, and 117 undertook the test (the remaining had either died, were ill, or had moved). Of 99 participants undertaking the test no further action was required, whilst 18 others required colonoscopy follow-up (no further results were reported). The team acknowledged they may not have identified all PWIDs and the education pack may not have suited all needs.

Within the literature there were a number of educational papers aimed at improving awareness about bowel cancer and screening. The first identified professionals as being crucial to active participation in bowel screening, as they were best placed to educate and encourage PWIDs [60], whilst the second [61] aimed at improving access to 5 screening services, 1 of which was bowel cancer. The team identified that there were no mechanisms to identify PWIDs, and advocated for PWIDs to be identified so that reasonable adjustments could be put in place. For example, they suggested that the GP practice should intervene 13 weeks after failure to respond to an invitation to attend bowel screening. Follow-up tests for positive FOBs identified that standard invitation letters were issued for a further screening appointment within 14 days. This meant there was limited time to enact reasonable adjustment. Another literature review on screening in PWIDs contained limited literature on cancer screening, although recommended following national guidelines for bowel screening [62].

Clinical Presentation and Treatment

Reports indicate that symptoms in PWIDs are similar to those observed in individuals without IDs: rectal bleeding [36, 43, 44, 48, 60, 62], diarrhoea [36, 48] and abdominal pain [30, 39, 48, 63], with other tumours being discovered early, resulting from investigations for anaemia [41]. One educational paper used 2 case studies to exemplify the difficulties that PWIDs can experience, such as the delay in reported rectal bleeding before medical investigations are undertaken [60]. Sometimes cancers are detected late, as observed in a study exploring deaths through abdominal obstruction. Of the 32 patients observed, 2 patients with mild to moderate IDs were diagnosed late from intestinal obstruction, and both died



Color version available online

Fig. 2. A picture from the booklet *Lucie est soignée pour un cancer*, showing surgery being performed on the patient.

from colorectal cancer [64], whilst another case study [63] found that 1 male patient with ID had died before a cancer diagnosis was made. The authors of this review found no systematic studies or evaluations unanimously indicating whether colorectal cancer was diagnosed late in PWIDs.

The use of colonoscopy to confirm diagnosis has been found to be difficult to perform in PWIDs [43]. One study found that the preparation for colonoscopy was adequate for only 51% of 40 patients with profound, severe, moderate, or mild IDs, compared to 97% of control patients without ID, whilst inadequate preparation meant that a caecal carcinoma was missed, and an advanced adenoma could not be removed [19]. This, however, was one study in one limited area and is not a reflection of all colonoscopy units. The dearth of literature precludes the authors determining if poor preparation for colonoscopy is a worldwide issue for PWID. However, poor preparation can necessitate a second colonoscopy [40] or even general anaesthesia [65]. There is limited research on how to prepare PWIDs for colonoscopy, other than the findings of the previous study and an educational paper about bowel cancer [66]. Suggestions were to make special arrangements and reasonable adjustments [19] in conjunction with the use of guides to explain colonoscopy to PWIDs [66].

The literature on the treatment of cancer is limited and where there have been delays in diagnosis the cancer will be at a more advanced stage [36, 59, 67]. Even if caught early, the PWIDs and those who support them have to make informed decisions. Within the review, a number of booklets were identified about how to explain cancer screening, diagnosis, and treatment in simple terms to PWIDs from a number of countries. These include: *Getting on with Cancer* [68], *Living with Cancer 3: Colorectal (Bowel) Cancer* [69] (Family Advice and Information Resource in Scotland, FAIR, no date; a CD was also available), *Having a Colonoscopy* [70], *An Easy Guide to Having a Colonoscopy* (NHS Cancer Screening Programme, no date), and *Keeping your Bowel Healthy* [71] in the UK; *Undersokelse og behandling av kreft* (one for men one for women [72]) in Norway, and in France, *Lucie est soignée pour un cancer,* which has a scenario based on a woman with colon cancer (Fig. 2) [73]. It is not the purpose of this review to evaluate the value of these publications; however, the authors believed it important to identify their existence. It should also be remembered that studies looking at cancer note the difficulties for treating this population due to barriers to treatment and follow-up procedures [61, 62, 74], and that patients may refuse treatment [62, 65]. This review only identified 1 British case study discussing aftercare, in this case dealing with stoma, in PWIDs [75]. The findings indicated that staff were poorly educated in terms of supporting PWIDs with stoma care, and this lack of knowledge and training was also apparent in home settings with the family and paid caregivers. These findings were reflected in another education article [76]. Due to the lack of evidence, it is unclear whether this reflects the situation worldwide given this work related to 1 individual. Similarly, there was 1 case study from Japan about chemotherapy which was successfully delivered at home [67].

Discussion

Despite the limitations of identifying populations, mortality data, and institutionalized studies, the findings reported suggest that PWIDs have at least a similar risk of colorectal cancer as the general population [6, 8, 27–31,

34]. There is a suggestion that a higher incidence may be associated with genetic deletions and family history, especially those with sub-microscopic or a larger 5q deletion due to their higher propensity for FAP than in the general population [35–39], as well as other genetic deletions which the research suggest harbour genes implicated in colorectal cancer [42–50]. Despite this, there remains indication of a slightly lower risk [5]. Robust conclusions are difficult to make due to the studies not reporting the histology of the colon cancers reported in PWIDs, and without this it is impossible to determine the stage and type of cancers identified. There is a suggestion that women with IDs are at higher risk than men [6, 28, 30], but there is a more mixed picture from the case studies. Interestingly, aside from the case studies, colon cancer was found in children with PWIDs [47, 49]. Overall, findings indicate vigilance about colorectal cancer is needed when assessing the health or changes in behaviour in PWIDs. The introduction of bowel screening and encouragement to undertake screening by health and social care professionals working with PWIDs should help this [60, 62, 63, 76]. More studies are important that report the histological type of colon cancer so that firmer conclusions can be drawn.

The picture that emerges from the limited range of studies regarding bowel screening is that the uptake of bowel cancer screening is lower for PWIDs and reflects the pattern of uptake in the other cancer screening programmes. This is concerning given the epidemiological data and institutional research reported above which suggests a comparable risk of colon cancer as in the general population [6, 8, 27–31, 32, 34]. Unlike other cancer screening programmes, bowel screening it is less invasive and offers a unique way to detect cancer early in patients who have difficulties accessing health care. Despite this, there are recognized barriers to cancer screening for PWIDs: difficulties in obtaining informed consent, lack of accessible information, inadequate support to complete the test, reduced accessibility to screening procedures, and negative attitudes and limited awareness about PWIDs among health professionals [59, 77–81]. One of the biggest difficulties is that PWIDs often find it difficult to communicate their pain and unease, and often react with unusual and sometimes challenging behaviour [82], such as quietness or overactivity. Many caregivers, however, may not be aware of the cancer risk of adults with IDs, meaning significant symptoms may be neglected and consequently long delays can occur between the onset of symptoms and diagnosis [83]. For this reason, caregivers need to be vigilant about complaints and/or changes in behaviour, to avoid the diagnosis being made too late [84, 85].

Unfortunately, lack of early identification is often attributed to poor knowledge about cancer and cancer screening in health and social care staff [58, 78, 81, 83]. For example, 1 study [81] surveyed 324 social care staff, reporting that 89% had received little training in cancer care, whilst 83% noted their knowledge of the signs, symptoms, and risks of cancer was limited. This was supported by previous work suggesting limited knowledge and training of health staff, including GPs, nurses, and radiographers [61, 62, 86–88]. As bowel cancer is seen as a condition that can be identified earlier, improvements at local and national levels are needed in order to provide information and follow-up of PWIDs [76, 78]. Work facilitating bowel screening is emerging mainly from the UK, and focuses on local projects to increase awareness and education about bowel screening and advocating greater contact with the learning (intellectual) disability care teams [52, 58, 59, 61]. The introduction of bowel screening has also seen a number of resources for caregivers and nurses who want to enable PWIDs to prepare for screening, a possibly difficult diagnosis, and the therapeutic journey.

If further investigation was required, the literature identified that preparation was difficult for those needing colonoscopies due to the nature of the agents and compliance with the regime to clean the bowel. For this reason, many PWIDs were found to have poor-quality colonoscopies, which increased the risk of cancers being missed [19, 43]. This is compounded by research identifying that oncology nurses and doctors often lack the necessary training to communicate effectively with PWIDs, and therefore do not meet the patient's needs and rely on the caregivers [74, 89, 90]. These and other studies indicate the importance of increasing the training and knowledge of physicians and nurses to improve understanding and confidence in communication when working with PWIDs [86–90].

Research on treatment options for colon cancer in PWIDs was limited, but reiterated the barriers and lack of training in health and social care professionals. This is concerning as the wider literature has identified poorer results for abdominal surgery in people with cognitive impairment due to delayed diagnoses, operative technical errors, and increased risk of postoperative complications [64, 91, 92] and limited successes [41]. PWIDs have increased risks from anaesthesia, there are known intubation risks in people with Down syndrome [93], and the choice of anaesthesia agents [94]. The wider literature

also identifies issues with outpatient treatment in that, if chemotherapy is offered, it may be difficult to provide to PWIDs as an outpatient [67] and will most certainly need to be adapted due to increased condition-specific side-effects to some antineoplastic agents [95]. Issues also occur after discharge in terms of communication difficulties between the general practitioner, caregivers, and the oncological team [80]. Although colon and rectal cancer are treatable without major difficulties, it remains imperative to obtain an early diagnosis. For this to happen, greater uptake of bowel screening needs to be achieved, along with better awareness about the signs and symptoms of bowel cancer in health and social care staff and training for staff in the oncology units.

Conclusion

As discussed above, literature reporting epidemiologic and mortality evidence on the incidence of bowel cancer in PWIDs is scant. Despite this, the available evidence suggests that colorectal cancer frequency in PWIDs is at least equivalent to that found in the general population. It is possible that incidence is masked by difficulties in diagnosis due to communication problems and the limited training of health and social care staff for identifying the signs and symptoms of this disease within this population. This can lead to tumours being discovered late or not at all. If tumours are diagnosed at an advanced stage, treatment options are limited, and the associated aggressive interventions may impair compliance. It is therefore important to consider preventative measures, such as bowel screening. PWIDs have a number of risk factors: a high incidence of obesity, limited access to exercise, and poorer dietary intake of fibre. Many of these risks could be reduced by caregivers being vigilant in terms of bowel habits and implementing interventions to increase fibre intake and exercise in daily activities. It is essential to increase participation in screening, especially as it represents one of the least invasive options for care. For this to succeed, accessible information must be made available for both PWIDs and those who support them. This can only be achieved with greater investment and the development of professional education for paid staff and family members, as well as increased awareness of the needs of PWIDs within oncology teams.

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