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Genetic Syndromes Including Intellectual Disability and Different Cancer Types

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Keywords
Intellectual disability · Cancer · Genetic screening

Abstract
Background: The aim of this study was to determine genetic syndromes including both cancers and intellectual disabilities, specific cancer types accompanying intellectual disabilities. ***Summary:*** We obtained the data from the clinical synopsis of the syndromes available in the OMIM database (https://www.omim.org/). In the first step, we detected 794 syndromes using different terms of intellectual disabilities and cancers. In the second step, we investigated the clinical synopsis of each syndrome in detail. Of these, we included 99 syndromes in which both intellectual disability and any type of cancer were presented. In the third step, we collected following data of these 99 syndromes: OMIM number, gene and location, syndrome/protein, tumor/neoplasia, inheritance, growth, head/neck, respiratory, cardiovascular, abdomen, genitourinary, skeletal, skin/nails/hair, neurologic, endocrine features, immunology, prenatal manifestations, laboratory abnormalities, and other system findings. ***Key Messages:*** The most common cancer types among these 99 syndromes are listed in percentage. Since individuals with intellectual disabilities have difficulty expressing themselves and understanding the symptoms of the disease, the diagnosis of diseases in these people is late and their treatment becomes difficult. We suggest that genetic tests to be performed in intellectual disability are important for early diagnosis, follow-up, and treatment of accompanying cancers. We especially emphasize the importance of leukemia, brain tumors, and tumors of embryonal origin in individuals with intellectual disability.

Introduction

Intellectual disability (ID) is a neurodevelopmental disorder characterized by cognitive deficits and functional impairments that occur during development. The physical health of people with IDs has been identified as an area of ongoing concern and priority [1]. There is a poor understanding of cancer prevention among patients with IDs [2]. Patients suffering from ID face inequalities throughout the entire spectrum of cancer care, including barriers to participating in screening and detection of cancer at more advanced stages than in the general population [3]. Deaths causing from cancers among this population occur up to 20 years earlier than the general population [2]. It is important to recognize that at least some of these malignancies and deaths could be avoided through adequate prevention and earlier intervention [3].

Both environmental and genetic factors have been implicated in the development of cancer in ID.

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Unhealthy lifestyles (including bad dietary habits and lack of physical activity) and genetic causes (well-known syndromes such as Down syndrome, tuberous sclerosis) have been highlighted as factors associated with increased cancer risk in ID [4–8]. However, syndromes that may be associated with ID have not been sufficiently investigated in detail. Recognition of these syndromes is important for cancer screening and follow-up. The aim of this study was to determine especially which cancers accompany ID and present detailed clinical symptoms in these genetic syndromes.

Methods

Online Mendelian Inheritance in Man (OMIM®) is an online catalog of human genes and genetic disorders. We obtained the data from the clinical synopsis of the syndromes available in the OMIM database (https://www.omim.org/). Keywords used in the literature search were "intellectual disabilities", "developmental delay", "intellectual impairment", "mentally retarded", "cognitive impairment", "delayed psychomotor development", "psychomotor retardation", "intellectual disability", "mental retardation", "cognitive delay", "impaired intellectual development", "global developmental delay" and "cancer", "carcinoma", "tumor", "neoplasia".

We obtained the data from the clinical synopsis of the syndromes available in the OMIM database (https://www.omim.org/). In the first step, we detected 794 syndromes using different terms of IDs and cancers. In the second step, we investigated the clinical synopsis of each syndrome in detail. Of these, we included 99 syndromes in which both ID and any type of cancer were presented. In the third step, we collected following data of these 99 syndromes: OMIM number, gene and location, syndrome/protein, tumor/neoplasia, inheritance, growth, head/neck, respiratory, cardiovascular, abdomen, genitourinary, skeletal, skin/nails/hair, neurologic, endocrine features, immunology, prenatal manifestations, laboratory abnormalities, and other system findings. Incidence frequencies were calculated as percentages. The flowchart of the study is presented in Figure 1.

Results

The detailed information on chromosomal location, genes, inheritance, types of cancers, and neuropsychological features of 99 syndromes was presented in Table 1. Types of cancer and associated genes were presented in Table 2. The most common types of cancer were as follows: 18.2% leukemia, 16.83% kidney cancer (renal carcinoma, renal cell carcinoma, tumors of the kidney, Wilms tumor), 16.83% brain tumor (meningioma, vestibular schwannoma, malignant schwannoma, pilocytic astrocytoma, medulloblastoma, ependymoma, giant cell astrocytoma), 13.86% skin cancer (squamous cell carcinoma, basal cell carcinoma, malignant melanoma, other types of skin cancer), 11.88% genitourinary tumor (rhabdomyosarcoma, embryonal tumors, sacrococcygeal teratoma, presacral teratoma), 9.1% lymphoma.

The anthropometric features involved in these 99 syndromes were as follows: 41.41% (41/99) for small-short stature, 34.34% (34/99) for microcephaly, 34.34% (34/99) for growth retardation/failure to thrive/poor overall growth, 19.2 (19/99) for macrocephaly, 11.11% (11/99) for overgrowth/macrosomia (comparative percentages of the anthropometric features were presented in Fig. 2). Anthropometric findings and associated genes were presented in Table 3. Additional accompanying findings of head/neck; respiratory-cardiovascular system; abdomen-skeletal system; genitourinary system; endocrine, immunology, prenatal laboratory features; skin, nails, hair, and other features of 99 syndromes were presented in online supplementary Tables 1–6, respectively (for all online suppl. material, see https://doi.org/10.1159/000549291).

Discussion

The previous studies emphasized the increased frequency of cancer diagnosis in individuals with ID, delayed diagnosis, and access to treatment [2]. The goal of cancer screening is to detect and treat precursor lesions at an early stage [9]. The importance of cancer screenings should be explained to the caregivers of these people and their participation should be ensured [10].

However, genetic etiology was not sufficiently emphasized in these cancer screening studies. As ID and cancer are both heterogeneous diseases with various etiologies, further studies are warranted to better understand the underlying mechanisms. Emerging evidence has suggested a plausible link between ID and cancer through several potential mechanisms. One possibility is that genetic variations contribute to both ID and oncogenesis. In some well-known syndromes, such as Down syndrome, a specific association with cancers has been emphasized in studies. For example, an effective cancer screening program for Down syndrome has recommended regular follow-up by palpation for

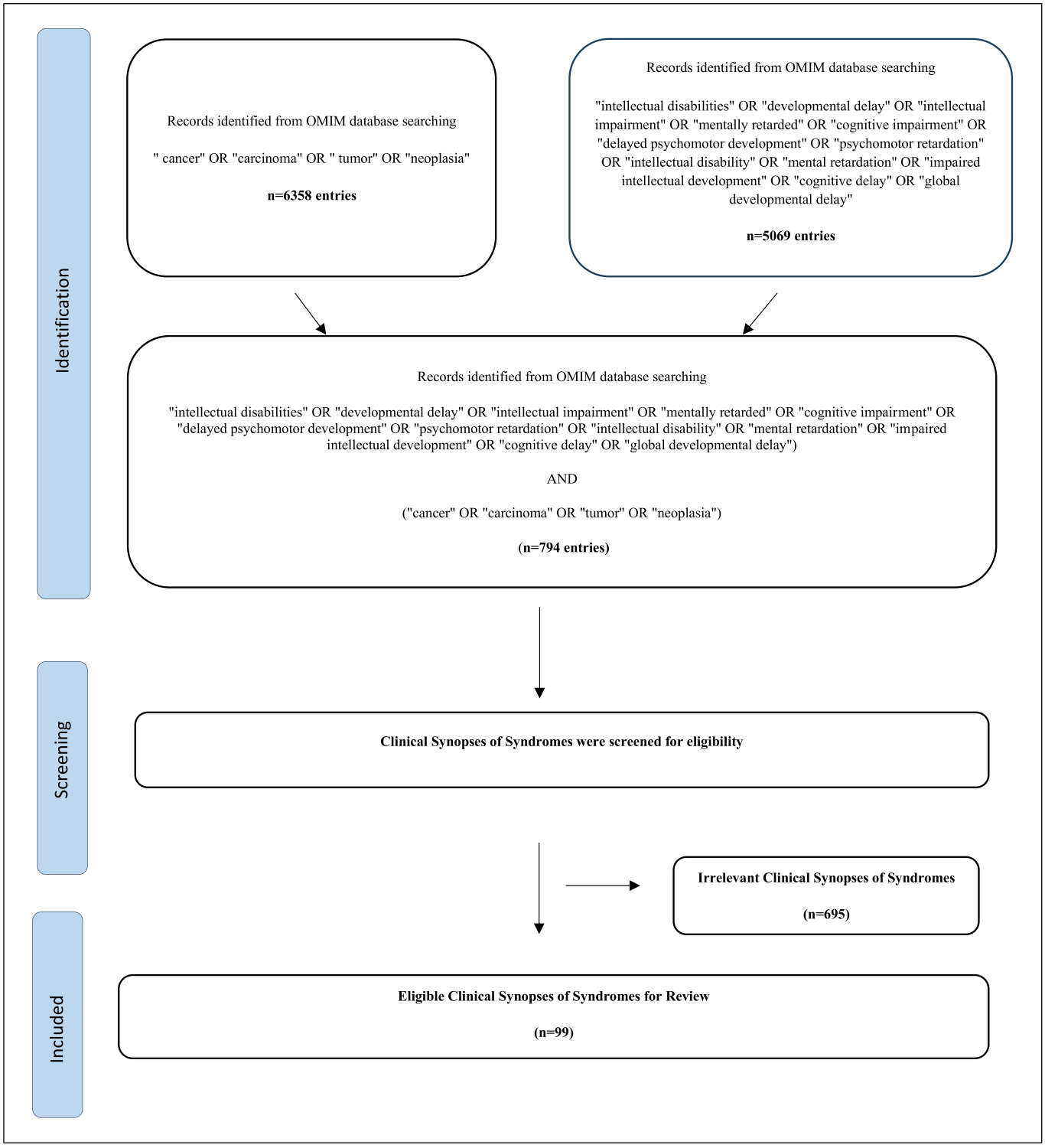

Fig. 1. Flowchart of screening procedure.

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Table 1. Genetic syndromes involving neoplasia and ID

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
194072	11p13	*WT1*	Wilms tumor, Airidia, genitourinary anomalies, and impaired intellectual development syndrome	Wilms tumor	AD, somatic mutation	Mental retardation
618272	14q32.13	*DICER1*	Global developmental delay, lung cysts, overgrowth, and Wilms tumor	Wilms tumor	Somatic mosaicism	Global developmental delay
158350	10q23.31	*PTEN*	Cowden syndrome 1	Breast cancer, ovarian carcinoma, cervical carcinoma, uterine adenocarcinoma, thyroid cancer, transitional cell carcinoma of the bladder, meningioma, mucosal neuromas	AD	Seizure, intention tremor, Lhermitte-Duclos disease, mental retardation, mild to moderate, psychomotor delay, mild to moderate, cerebellar gangliocytoma manifesting as seizure and tremor
218040	11p15.5	*HRAS*	Costello syndrome	Epithelioma, bladder carcinoma, rhabdomyosarcoma, vestibular schwannoma	AD	Delayed psychomotor development, sleep disorder, mental retardation, cerebral atrophy, ventriculomegaly, hydrocephalus, posterior fossa crowding, progressive, enlarged cerebellum, cerebellar tonsillar herniation, Chiari I malformation
162300	10q11.21	*RET*	Multiple endocrine neoplasia, type IIB	Ganglioneuroma, pheochromocytoma, medullary thyroid carcinoma, parathyroid disease rare	AD	Hypotonia, developmental delay
612469	11p13-p12	–	Wagro syndrome	Wilms tumor	AD	Developmental delay, impaired intellectual development, hyperphagia, pervasive developmental delay, anxiety disorder
617883	17q21.31	*BRCA 1*	Fanconi anemia, complementation group S	Increased susceptibility to cancer, breast cancer, ovarian cancer	AD	Developmental delay, ID, speech delay

Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
180849	16p13.3	*CREBBP*	Rubinstein-Taybi syndrome 1	Increased risk of tumor formation, especially of the head, increased risk of leukemia	AD	Mental retardation (average IQ 51), agenesis of corpus callosum, severe expressive speech delay, poor coordination, EEG abnormalities, seizures, hypotonia, hyperreflexia, good social contacts, short attention span, labile mood
620189	7p22.3	*MAD1L1*	Mosaic variegated aneuploidy syndrome 7 with inflammation and tumor predisposition	Wide spectrum of neoplasms and malignancies	AD	Delayed development (mild), normal intellect
601675	19q13.32	*ERCC2*	Trichothiodystrophy 1, photosensitive	Squamous cell carcinoma, basal cell carcinoma	AR	Mental retardation (IQ 45–58), poor gross and fine motor coordination, speech delay, diffuse signal hyperintensity of white matter seen on MRI
257300	15q15.1	*BUB1B*	Mosaic variegated aneuploidy syndrome 1	Propensity to tumor development, Wilms tumor, nephroblastoma, rhabdomyosarcoma, leukemia	AR	Developmental delay, profound, mental retardation, profound, hypotonia, generalized, seizures, generalized tonic-clonic, seizures, myoclonic, hypoplastic cerebrum, pachymacrogyria, cerebral oligogyria, hypodysplasia of the corpus callosum, agenesis of the corpus callosum, posterior fossa malformations, Dandy-Walker malformation, enlarged ventricles, hydrocephalus, cerebellar hypoplasia
109400	9q22.32	*PTCH1 PTCH2, SUFU*	Basal cell nevus syndrome 1	Ovarian carcinoma, basal cell carcinoma	AD	Mental retardation, calcification of the falx cerebri, medulloblastoma
312870	Xq26.2	*GPC3*	Simpson-Golabi-Behmel syndrome, type 1	Embryonal tumors, Wilms tumor	XLR	Development varies from normal to retarded, agenesis of corpus callosum, cerebellar vermis hypoplasia, hydrocephalus, hypotonia

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Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
613254	12q15, 16p13.3	*TSC2*	Tuberous sclerosis 2	Myocardial rhabdomyoma, multiple bilateral renal angiomyolipoma, ependymoma, renal carcinoma, giant cell astrocytoma, chordoma, benign tumors of the eye, heart, and lungs	AD	Hamartomatous lesions of the brain, subependymal nodules, cortical tubers, infantile spasms, seizures, mental retardation, learning difficulties, intracranial calcification by X-ray or CT, attention-deficit disorder, hyperactivity, autism
305000	Xq28	*DKC1*	Dyskeratosis congenita, X-linked	Squamous cell carcinoma, acute myeloid leukemia, Hodgkin disease, pancreatic carcinoma	XLR	Delayed development, learning difficulties, mental retardation, cerebellar ataxia, cerebellar hypoplasia
117550	5q35.3	*NSD1*	Sotos syndrome	Wilms tumor	AD	Developmental delay, variable mental retardation, neonatal hypotonia, hyperreflexia, poor coordination, seizures, behavioral problems, expressive language delay, partial to complete agenesis of corpus callosum, persistent cavum septum pellucidum, large cisterna magna, ventriculomegaly, prominent trigone and occipital horns
210900	15q26.1	*RECQL3*	Bloom syndrome	Leukemia, lymphoma, adenocarcinoma, squamous cell carcinoma, hypersensitivity to chemotherapy	AR	Mild mental retardation in some, learning disability
617107	11q13.1	*FIBP*	Thauvin-Robinet-Faivre syndrome	Wilms tumor	AR	Delayed psychomotor development, mild, ID, mild, learning disabilities, language disabilities
190685	21q22.3	–	Down syndrome	Leukemia, acute megakaryocytic leukemia	Isolated cases	Mental retardation, Alzheimer disease, hypotonia, poor Moro reflex

Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
162200	17q11.2	*NF 1*	Neurofibromatosis, type İ	Optic glioma, meningioma, hypothalamic tumor, neurofibrosarcoma, rhabdomyosarcoma, duodenal carcinoid, somatostatinoma, parathyroid adenoma, pheochromocytoma, pilocytic astrocytoma, malignant peripheral nerve sheath tumors, tumors at multiple other sites including CNS	AD	Learning disabilities, impaired intellectual development, mild, aqueductal stenosis, hydrocephalus
304050	Xp22	–	Aicardi syndrome	Hepatoblastoma, benign teratoma, embryonal carcinoma, metastatic angiosarcoma	XLD	Mental retardation, profound, infantile spasms, seizures, hypotonia, Dandy-Walker malformation, Arnola-Chiari malformation, cavum septum pellucidum, choroid plexus cyst, intracranial cysts, delayed myelination, partial-total agenesis of corpus callosum, enlarged lateral and third ventricles, cortical heterotopias, subependymal heterotopias, pachygyria, hypoplastic cerebellar vermis, dysplasia of the cerebellar hemispheres, polymicrogyria, predominantly frontal and perisylvian, tectal enlargement, widening of the operculum, asymmetric brain development
267000	2q37.1	*DIS3L2*	Perlman syndrome	Bilateral renal hamartomas, nephroblastomatosis, Wilms tumor	AR	Corpus callosum agenesis, developmental delay
304150	Xq21.1	*ATP7A*	Occipital horn syndrome	Bladder carcinoma	XLR	Low-normal IQ

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Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
176920	14q32.33	*AKT1*	Proteus syndrome	Ovarian cystadenoma, parotid monomorphic adenoma	Somatic mutation	Brain malformations, spinal cord compression by tumor infiltration, mental retardation, moderate
235000	11p15.5	–	Hemihyperplasia, isolated	Increased risk for embryonal tumors, increased risk for Wilms tumor	AD	Mental retardation, mild to moderate
278700	9q22.33	–	Xeroderma pigmentosum, complementation group A	Early-onset skin cancer		Mental deterioration, low intelligence, microcephaly, sensorineural deafness, hyporeflexia, spasticity, ataxia, choreoathetosis
617598	5p15.33	*TRIP13*	Mosaic variegated aneuploidy syndrome 3	Wilms tumor	AR	Developmental delay, seizures
617052	5p13.2	*DNAJC21*	Bone marrow failure syndrome 3	Possible cancer predisposition	AR	Global developmental delay, hyperactivity
223370	–	–	Dubowitz syndrome	Aplastic anemia, acute lymphatic leukemia, lymphoma, neuroblastoma	AR	Mental retardation, moderate to severe, hyperactivity, hypotonia, speech delay, short attention span
274700	8q24.22	*TG*	Thyroid dyshormonogenesis 3	Thyroid cancer	AR	Mental retardation
163200	1p13.2, 11p15.5, 12p12.1	*KRAS, HRAS*	Schimmelpenning-Feuerstein-Mims syndrome	Basal cell carcinoma, syringocystadenoma papilliferum, central giant cell granuloma, trichoblastoma	Somatic mosaicism	Neurologic abnormalities in about 7%, mental retardation, seizures, hemimegalencephaly
101400	7p21.1, 10q26.13	*TWIST*	Saethre-Chotzen syndrome	Increased risk of breast cancer in women	AD	

Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
616541	5q14.2	*XRCC4*	Short stature, microcephaly, and endocrine dysfunction	Gastrointestinal stromal tumor of jejunum, thalamic glioma, low grade	AR	Developmental delay, speech delay, cognitive impairment, severe attention deficit, constructive apraxia, visuospatial memory deficit, dysarthria, dysmetria, dysdiadochokinesia, ataxia, progressive, pyramidal signs, diffuse, wide-based gait, atrophy of cerebellar vermis, mild, dilated cerebral ventricles, small caudate nuclei, simplified gyral pattern, sensory neuropathy
278760	16p13.12	*ERCC4*	Xeroderma pigmentosum, complementation group F	Skin cancer susceptibility	AR	Learning disabilities, mental retardation, ataxia, tremor, dementia, brain atrophy
610651	2q14.3	*ERCC3*	Xeroderma pigmentosum, complementation group B	Melanoma, basal cell carcinoma, squamous cell carcinoma, increased risk of malignancy	AR	Mental retardation, ataxia, enlarged cerebral ventricles, cerebellar atrophy, abnormal myelination, basal ganglia calcifications, hyperreflexia, decreased nerve conduction velocity
269150	18q12.3	*SETBP1*	Schinzel-Giedion midface retraction syndrome	Embryonal tumors, hepatoblastoma, sacrococcygeal teratoma	AD	Mental retardation, seizures, opisthotonus, spasticity, hypsarrhythmia, cerebral atrophy, ventriculomegaly, thin corpus callosum
163950	12q24.13	*PTPN11*	Noonan syndrome 1	Malignant schwannoma, multiple giant cell granulomas	AD	Articulation difficulties, mental retardation
268400	8q24.3	*RECQL4*	Rothmund-Thomson syndrome, type 2	Basal cell carcinoma, squamous cell carcinoma, osteogenic sarcoma	AR	Mental retardation in 5–13%
118450	20p12.2	*JAG1*	Alagille syndrome 1	Hepatocellular carcinoma, papillary thyroid carcinoma	AD	Mental retardation, mild, learning disability, absent deep tendon reflexes

Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
251260	8q21.3	*NBS1*	Nijmegen breakage syndrome	Lymphoma, glioma, medulloblastoma, rhabdomyosarcoma	AR	Normal IQ in infancy, then IQ drops with age, mental retardation by the age of 7 years, hyperactivity, neurodegeneration
260400	8q21.3	*SBDS*	Shwachman-Diamond syndrome 1	Myelodysplasia, acute myelogenous leukemia	AR	Learning disabilities, developmental delay, mild mental retardation decrease global brain volume, smaller age- and head size-adjusted areas of posterior fossa, vermis, corpus callosum, and pons than healthy controls, larger cerebrum-vermis ratio than the healthy controls
613563	11q23.3	*CBL*	Noonan Syndrome-like disorder with or without juvenile myelomonocytic leukemia	Increased susceptibility to juvenile myelomonocytic leukemia	AD	Delayed psychomotor development, mild, language delay
308100	Xp22.31	*STS*	İchthyosis, X-linked	A subset of patients have additional features, including mental retardation and hypogonadism associated with larger deletions at Xp22.3	XLR	
249400	1p13.2	*NRAS*	Melanosis, neurocutaneous	Susceptibility to malignant melanoma	Somatic mutation	Parenchymal neuromelanosis, Dandy-Walker malformation, delayed development, seizures, hydrocephalus, leptomeningeal melanocytosis, choroid plexus papilloma, meningioma, spinal cysts, arachnoid cysts, syringomyelia
619126	4q24	*TET2*	İmmunodeficiency 75 with lymphoproliferation	Lymphoma, B cell, EBV associated, lymphoma, T cell	AR	Developmental delay, moderate

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Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
602501	3q26.32	*PIK3CA*	Megalencephaly-capillary malformation-polymicrogyria syndrome	Increased risk of meningioma, increased risk of Wilms tumor, increased risk of leukemia	Somatic mutation	Developmental delay, mental retardation, hypotonia, seizures, MRI shows brain asymmetry, ventriculomegaly, hydrocephalus, large cerebellum, progressive, cerebellar tonsil herniation, crowding of the posterior fossa, cavum septum pellucidum, cavum vergae, polymicrogyria, cortical dysgenesis, thickened corpus callosum, thickened optic nerve sheath, dilated venous sinuses, white matter signal abnormalities in the deep white matter and periventricular regions
616553	16q22.1	*ACD*	Dyskeratosis congenita, autosomal dominant 6	Increased susceptibility to cancer	AD, AR	Delayed development, cerebellar hypoplasia
618162	11q23.3	*SIK3*	Spondyloepimetaphyseal dysplasia, Krakow type	Epstein-Barr virus-induced small-muscle cancer (in 1 patient)	AR	Developmental delay, seizures, Chiari malformation, hydrocephalus, ventricular enlargement, cerebral masses consistent with cavernous malformation, flattening of medulla
278730	19q13.32	*ERCC2*	Xeroderma pigmentosum, complementation group D	Early onset skin cancer (basal cell, squamous cell, and malignant melanoma)	AR	Mental deterioration, low intelligence, hyporeflexia, spasticity, ataxia, choreoathetosis
227646	3p25.3	*FANCD2*	Fanconi anemia, complementation group D2	Leukemia	AR	Mental retardation
105650	19q13.2	*RPS19*	Diamond-Blackfan anemia 1	Osteogenic sarcoma, myelodysplastic syndrome, colon cancer	AD	Mental retardation
606593	13q33.3	*LIG4*	Lig4 syndrome	T-cell acute lymphoblastic leukemia (in 1 patient)	AR	Developmental delay, global, delayed speech
224230	15q14	*NOP10*	Dyskeratosis congenita, autosomal recessive 1	Increased risk of malignancy (classic feature)	AR	Learning difficulties, mental retardation

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Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
615279	15q22.31	*MAP2K1*	Cardiofaciocutaneous syndrome 3	Hepatoblastoma	AD	Developmental delay, seizures, hypoplastic corpus callosum
176450	7q36.3	*MNX1*	Currarino syndrome	Presacral teratoma	AD	Anterior sacral meningocele, tethered cord, developmental delay
227650	16q24.3	*FANCA*	Fanconi anemia, complementation group A	Leukemia	AR	Mental retardation
600901	6p21.31	*FANCE*	Fanconi anemia, complementation group E	Leukemia	AR	Psychomotor retardation
610738	1q21.3	*HAX1*	Neutropenia, severe congenital, 3, autosomal recessive	Increased risk of myelodysplastic syndromes, increased risk of leukemia	AR	Psychomotor retardation (in some patients), seizures (in some patients)
191100	9q34.13	*TSC1*	Tuberous sclerosis 1	Myocardial rhabdomyoma, multiple bilateral renal angiomyolipoma, ependymoma, renal carcinoma, giant cell astrocytoma, chordoma, benign tumors of the eye, heart, and lungs	AD	Hamartomatous lesions of the brain, subependymal nodules, cortical tubers, infantile spasms, seizures, mental retardation (30%), learning difficulties, intracranial calcification by X-ray or CT, attention-deficit disorder, hyperactivity, autism
120470	18q21.2	*DCC*	Dcc netrin 1 receptor	Colorectal cancer, esophageal cancer, esophageal squamous cell carcinoma	AD, AR, somatic mutation	Delayed psychomotor development, ID, Mirror movements (in some patients), hyperreflexia (in some patients), agenesis of the corpus callosum, absent anterior commissure
116806	3p22.1	*CTNNB1*	Catenin, beta-1	Colorectal cancer, primary liver cancer, medulloblastoma, ovarian cancer, dysgerminoma, ovarian papillary adenocarcinoma, serous ovarian cystadenocarcinoma, breast cancer	AD, AR, somatic mutation	Delayed psychomotor development,ID, speech impairment, spastic diplegia, progressive, hypoplastic corpus callosum

Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
600993	18q21.2	*SMAD4,KRAS,TP53, MADH4, STK11*	Smad family member 4	Increased risk of gastrointestinal cancer, pancreatic ductal adenocarcinoma, colon cancer	AD, somatic mutation	Mental retardation, seizures (rare), cerebellar ataxia (1 patient), cerebellar atrophy, progressive (1 patient)
615879	2p23.3	*DNMT3A*	Tatton-Brown-Rahman syndrome	Increased risk of acute myeloid leukemia, particularly associated with R882 mutations, increased risk of hematopoietic malignancies	AD	ID, mild to moderate, seizures (less common), small optic chiasm, optic nerve hypoplasia, encephalomalacia, Chiari I malformation, hypotonia
260350	12p12.1, 12q13.13, 17p13.1, 18q21.2, 19p13.3	*NRAS, KRAS*	Ras-associated autoimmune leukoproliferative disorder	Breast carcinoma, bladder cancer, alveolar cell carcinoma, nonsmall cell lung cancer, adenocarcinoma of lung, pancreatic ductal adenocarcinoma, basal cell carcinoma	AD, somatic mutation, multifactorial	Mental retardation, developmental delay, mild to severe (in some patients), arachnoid cyst (in some patients), epilepsy, moderate learning difficulties
605131	16q23.1-q23.2	*WWOX*	Ww domain-containing oxidoreductase	Esophageal cancer, esophageal squamous cell carcinoma	AD, AR, somatic mutation	Epileptic encephalopathy, delayed psychomotor development, profound, seizures, refractory, spasticity, hyperreflexia, rigidity, hypokinesia, disorganized slow background activity seen on EEG, thin corpus callosum (in some patients), delayed myelination (in some patients), cerebral atrophy (in some patients)
164757	7q34	*EGFR, BRAF*	B-Raf protooncogene, serine/ threonine kinase	Alveolar cell carcinoma, nonsmall cell lung cancer, adenocarcinoma of lung, colorectal cancer, malignant melanoma	AD, somatic mutation	Developmental delay, mental retardation, seizures (in some patients), hypotonia, cognitive defects
607035	10q24.32	*SUFU*	Sufu negative regulator of Hedgehog signaling	Medulloblastoma, meningioma	AD, AR, somatic mutation	Developmental delay, ID, mild, ataxia, dysarthria, cerebellar abnormalities, molar tooth sign, polymicrogyria (in some patients)

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Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
300715	Xq21.1	*MAGT1*	Magnesium transporter 1	Increased susceptibility to EBV-associated B-cell lymphoproliferative disorders, Hodgkin lymphoma (in some patients)	XLR	Developmental delay, impaired intellectual development, cavum septum pellucidum, seizures (rare), leukoencephalopathy (rare), brain atrophy (rare)
611060	18q12.3	*SETBP1*	Set-binding protein 1	Embryonal tumors, hepatoblastoma, sacrococcygeal teratoma	AD	Impaired intellectual development, mild to moderate (IQ 30–76), delayed speech, absent speech, delayed motor development, seizures (uncommon), mental retardation, spasticity, hypsarrhythmia, cerebral atrophy, ventriculomegaly, thin corpus callosum
602700	22q13.2	*EP300*	E1a-binding protein, 300-Kd	Colorectal cancer	AD, somatic mutation	Developmental delay, ID, speech delay, mental retardation, mild to moderate, low-normal intelligence, autism spectrum disorder (in some patients), delayed psychomotor development, delayed gross motor development
139320	20q13.32	*GNAS*	Gnas complex locus; Gnas	Pituitary adenoma	AD, somatic mutation	Cognitive decline, cognitive deficits, mental retardation, hypocalcemic tetany, seizures, basal ganglion calcification, calcified choroid plexus
606241	14q32.13	*DICER1*	Dicer 1, ribonuclease III	Wilms tumor susceptibility, papillary thyroid carcinoma (rare), Sertoli-Leydig cell ovarian tumors may occur, rhabdomyosarcoma, medulloblastoma	AD, somatic mutation	Global developmental delay

Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
603089	3p21.1	*BAP1*	Brca1-associated protein 1	Uveal melanoma, mesothelioma, malignant, after asbestos exposure, cutaneous melanoma, meningioma, renal cell carcinoma, usually clear cell type	AD	Global developmental delay, mild, delayed walking, speech delay, language difficulty, impaired intellectual development, mild to borderline (in most patients), hypotonia, seizures, mild (in some patients)
187270	5p15.33	*TERT*	Telomerase reverse transcriptase	Cutaneous malignant melanoma, increased risk for cancer	AD, AR, somatic mutation	Learning difficulties (seen in recessive form), developmental delay (seen in recessive form), cerebellar hypoplasia (seen in recessive form)
136850	1q43	*FH*	Fumarate hydratase	Uterine leiomyosarcoma (less common), cutaneous leiomyosarcoma (less common), renal cell carcinoma, solitary papillary type 2 (about 20% of patients)	AD, AR	Mental retardation, profound, developmental delay, no language development, cerebral atrophy, seizures, status epilepticus, hypotonia, most patients do not achieve independent sitting or walking, ventricular enlargement, polymicrogyria, open operculum, choroid plexus cysts, decreased white matter volume, angulation of the frontal horns, small brainstem, agenesis of the corpus callosum
605100	17q23.2	*PPM1D*	Protein phosphatase, magnesium/manganese dependent, 1d	Breast carcinoma	AD, somatic mutation	Delayed psychomotor development, ID, learning difficulties, broad-based gait, language delay, high pain threshold, hypersensitivity to sound
190182	3p24.1	*TGFBR2*	Transforming growth factor-beta receptor, type II	Esophageal cancer, esophageal squamous cell carcinoma	AD, somatic mutation	Mental retardation (uncommon), developmental delay (uncommon), Chiari malformation (uncommon), hydrocephalus (uncommon)

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Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
164840	2p24.3	*MYCN*	Mycn proto-oncogene, Bhlh transcription factor	Increased risk of neuroblastoma	AD	Mental retardation, learning disability (90% patients), ventriculomegaly, developmental delay, motor delay, impaired intellectual development, megalencephaly, hypoplastic corpus callosum
190181	9q22.33	*TGFBR1*	Transforming growth factor-beta receptor, type I	Squamous epitheliomas, multiple	AD	Mental retardation (uncommon), developmental delay (uncommon), Chiari malformation (uncommon), hydrocephalus (uncommon)
167410	1p36.13	*PAX7*	Paired box gene 7	Alveolar rhabdomyosarcoma	AR, somatic mutation	Delayed motor development, impaired gait, variable, normal cognitive development, areflexia
608210	11p11.2	*EXT2*	Exostosin glycosyltransferase 2	Increased risk of chondrosarcoma	AD, AR	Delayed psychomotor development, impaired intellectual development, motor delay, variable, poor or absent speech, unsteady gait, seizures, nonspecific background slowing with focal spikes seen on EEG, brain hemorrhage (in 2 of 4 patients)
603816	16p13.3	*AXIN1*	Axis inhibitor 1	Primary liver cancer	AR, somatic mutation	Global developmental delay, brain atrophy (in 1 patient), pachygyria (in 1 patient), corpus callosum hypoplasia (in 1 patient)
609413	10q11.23	*ERCC6*	Ercc excision repair 6, chromatin remodeling factor	Alveolar cell carcinoma, nonsmall cell lung cancer, adenocarcinoma of lung	AD, AR, somatic mutation	Seizures (in 1 patient), impaired intellectual development, developmental regression, hypertonic lower extremities, diffuse leukodystrophy seen on MRI, cerebral atrophy, olivopontocerebellar atrophy, ventriculomegaly, mental retardation, profound, lack of motor development, lack of speech development

Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
601215	3q23	*ATR*	Atr serine/threonine kinase	Oropharyngeal cancer, nonmelanoma skin cancer (in some patients), breast cancer (rare), cervical cancer (rare)	AD, AR	Mental retardation, seizures, pachygyria, arachnoidal cysts, large basal ganglia, hypoplasia of the cerebellar vermis
602839	1p36.22	*PIC3CD*	Phosphatidylinositol 3-kinase, catalytic, delta	Increased susceptibility to B-cell lymphoma	AD, AR, digenic recessive	Myoclonic seizures, developmental delay, hypotonia, ataxia, tremor, dilated ventricles
156100	22q12.1	*MN1*	Mn1/Tel fusion gene, included	Meningioma	AD	Global developmental delay, delayed walking, impaired intellectual development, expressive language impairment, nonverbal, seizures (in some patients), brain malformations (in most patients), rhombencephalosynapsis, crowded posterior fossa, polymicrogyria, cerebellar hypoplasia, cerebellar dysplasia, thin corpus callosum, enlarged ventricles, hypoplastic olfactory bulbs, prominent posterior clinoid process
157660	9p13.3	*RMRP*	Mitochondrial Rna-processing endoribonuclease, Rna component of	Increased malignancy risk, especially lymphoma and skin neoplasm, Hodgkin lymphoma (in 1 patient), neuroendocrine carcinoma (in 1 patient)	AR	Cervical cord compression, impaired intellectual development, mild
606463	1q22	*GBA1*	Glucosidase, beta, acid	Increased risk for multiple myeloma, increased risk for monoclonal gammopathy	AD, AR, multifactorial	Subacute neurologic deterioration, seizures, delayed motor development, intellectual deterioration, speech abnormalities, ataxia, spastic paraparesis, myoclonus (subtype 3A), myoclonic seizures (subtype 3A), dementia (subtype 3A)

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Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
154950	14q23.3	*MAX*	Max protein	Adrenal medullary tumor	AD	Developmental delay, prominent perivascular spaces in the basal ganglia/pericaudate region, prominent perivascular spaces adjacent to trigone of lateral ventricle
602667	8q21.3	*NBN*	Nibrin	Lymphoma, glioma, medulloblastoma, rhabdomyosarcoma	AR	Normal IQ in infancy, then IQ drops with age, mental retardation by the age of 7 years, hyperactivity, neurodegeneration
606478	7q31.33	*POT1*	Protection of telomeres 1	Neoplasms, benign and malignant, clonal hematopoietic disorders, lymphoma, leukemia, melanoma, glioma, epithelial tumors, mesenchymal tumors	AD, AR	Global developmental delay, neurologic regression, inability to walk, absent speech, intracranial calcifications, leukoencephalopathy
188830	17q24.2	*PRKAR1A*	Prkar1a/Rara fusion gene, included	Myxoid subcutaneous tumors, primary adrenocortical nodular hyperplasia, testicular Sertoli cell tumor, calcified, pituitary adenoma, mammary ductal fibroadenoma, schwannoma, psammomatous melanotic schwannomas, thyroid carcinoma, pheochromocytoma	AD	Mental retardation (IQ 24–85) (variable), hydrocephalus, cognitive decline
617244	15q15.1	*RAD51*	Fanconi anemia, complementation group R	Breast carcinoma	AD	Hydrocephalus, delayed early milestones, ID, learning disabilities

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Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
185470	1p36.13	*SDHB*	Succinate dehydrogenase complex, subunit B, iron sulfur protein	Paragangliomas, multiple tumors in 28% of patients, paragangliomas, head and neck (31%), chemodectomas, carotid body tumors, glomus jugular tumors, pheochromocytomas, adrenal (28%), pheochromocytomas, extraadrenal (48%), malignancy (34%), gastrointestinal stromal tumors (less common), renal cell carcinoma (less common), neuroblastoma (less common)	AD, AR	Developmental regression, cognitive impairment, encephalopathy, seizures (in 1 patient), ataxia, truncal hypotonia, appendicular hypertonia, elevated lactate seen on magnetic resonance spectroscopy, elevated succinate seen on magnetic resonance spectroscopy, abnormalities of the hemispheric white matter with sparing of subcortical U fibers seen on MRI, abnormalities of the thalamus seen on MRI, abnormalities of the corpus callosum seen on MRI, abnormalities of the spinal cord seen on MRI, headache (with pheochromocytoma), cranial nerve palsies can arise with head and neck paragangliomas, anxiety (with pheochromocytoma)
602690	11q23.1	*SDHD*	Succinate dehydrogenase complex, subunit D, integral membrane protein	Paragangliomas, multiple tumors in 74% of patients, paragangliomas, head and neck (79%), chemodectomas, carotid body tumors (most common location), glomus jugular tumors, vagal nerve tumors (glomus vagale), tympanic nerve tumors (glomus tympanicum), pheochromocytoma, adrenal (53%), pheochromocytoma, extraadrenal (21%)	AD, AR	Developmental regression, loss of motor skills, impaired intellectual development, encephalopathy, ataxia, dystonia, seizures, myoclonus, headache (with pheochromocytoma), cranial nerve palsies can arise with head and neck paragangliomas
603552	6q24.2	*STX11*	Hemophagocytic lymphohistiocytosis, familial, 4	Increased risk of myelodysplastic syndrome or leukemia (in some patients)	AR	Delayed psychomotor development (in some patients), seizures (in some patients), hypotonia (in some patients)

Table 1 (continued)

OMIM No.	Location	*Gene*	Syndrome	Tumor/neoplasia	Inheritance	Neuropsychiatric findings
608537	3p25.3	*VHL*	Von Hippel-Lindau tumor suppressor	Adrenal medullary tumor, pheochromocytoma, hemangioblastoma, pancreatic cancer, paraganglioma, adenocarcinoma of ampulla of Vater	AD, AR	Cerebral hemorrhage, headache, cerebral vascular events, cerebral hemorrhage, delayed development (in some patients), cerebellar hemangioblastoma, spinal cord hemangioblastoma
604319	14q12	*TINF2*	Terf1-interacting nuclear factor 2	Increased risk of malignancy	AD	Speech delay, learning difficulties, intracranial calcifications, cerebellar hypoplasia, cerebellar ataxia, central nervous system, psychomotor retardation, ataxia, cerebellar hypoplasia, cerebral calcifications, hypertonia, progressive neurologic deterioration
611170	7q21.2	*SAMD9L*	Sterile alpha motif domain-containing protein 9 like	Increased risk of AML, acute myelogenous leukemia (in some patients)	AD	Cerebellar ataxia, gait instability, dysarthria, dysmetria, yperreflexia, extensor plantar responses, ankle clonus, cerebellar atrophy, cerebellar degeneration, white matter abnormalities (in some patients), spinocerebellar ataxia, unsteady gait, hyperreflexia, gait ataxia, dysarthria, dysmetria, dysdiadochokinesis, extensor plantar responses, pyramidal signs, cerebellar atrophy seen on brain imaging, diffuse cerebral demyelination, distal sensory impairment (in some patients), decreased nerve conduction velocities (in some patients), axonal sensory polyneuropathy (in some patients), ADHD

Table 2. Types of neoplasm and associated genes

	Genes
Malignant neoplasm of kidney	*WT1, TSC1, TSC2, BUB1B, DIS3L2, FH, VHL, DICER1, GPC3, FIBP, TRIP13, PICK3CA,*
Malignant neoplasm of breast	*PTEN, BRCA1, TWIST, CTNNB1, NRAS, KRAS, PPM1D, ATR, RAD51*
Malignant neoplasm of ovary	*PTCH1, PTCH2, SUFU, PTEN, BRCA1, CTNNB1, DICER1*
Malignant neoplasm of cervix	*PTEN, ATR*
Malignant neoplasm of uterine	*PTEN, FH*
Malignant neoplasm of thyroid	*PTEN, RET, TG, JAG1, PRKAR1A*
Malignant neoplasm of bladder	*PTEN, HRAS, ATP7A, NRAS, KRAS*
Leukemia	*CREBBP, BUB1B, DKC1, RECQL3, SBDS, CBL, PIK3CA, FANCD2, LIG4, FANCA, FANCE, HAX1, DNMT3A, STX11, SAMD9L*
Lymphoma	*RECQL3, NBS1, TET2, MAGT1, PIC3CD, RMRP, NBN, POT1*
Pancreatic cancer	*DKC1, SMAD4, KRAS, TP53, MADH4, STK11, NRAS, KRAS, VHL*
Genitourinary tumor	*HRAS, GPC3, SETBP1, MNX1*
Adrenal tumor	*MAX, VHL*
Brain tumor	*PTEN, HRAS, NF1, NBS1, PIK3CA, TSC1, SUFU, DICER1, BAP1, MN1, NIBRIN; NBN, PTPN11*
Skin cancer	*ERCC2, ERCC3, ERCC4, PTCH1, PTCH2, SUFU, KRAS, HRAS, RECQL4, NRAS, DKC1, RECQL3, EGFR, BRAF, TERT, ATR*
Head and neck tumor	*TSC1, TSC2, NF 1, TG, SDHB, SDHD*
Lung cancer	*NRAS, KRAS, EGFR, BRAF, DICER1, ERCC6, BAP1*
Liver cancer	*CTNNB1, AXIN1, JAG1, MAP2K1, SETBP1*
Esophageal cancer	*DCC, TGFBR2*
Colorectal cancer	*EP300, EGFR, BRAF, SMAD4, KRAS, TP53, MADH4, STK11, CTNNB1, DCC, RPS19*

testicular cancers [10]. Also, the increased risk of leukemia was especially associated with the diagnosis of Down syndrome. Our results also show that the diagnosis of leukemia accompanies ID in other genetic syndromes in addition to Down syndrome. There are other well-known syndromes such as tuberous sclerosis, neurofibromatosis, and WAGR syndrome, which can often be the cause of both cancer and ID [6–8].

In this study, we aimed to identify syndromes that can cause both cancer and ID performing a detailed screening based on the system findings in the OMIM database clinical synopsis. In our study, we determined 99 syndromes including both IDs and any types of cancer using OMIM database. We listed the most common types of cancer. The most common types of cancer detected in our study were (1) leukemia, (2) kidney cancer, (3) brain tumor, (4) skin cancer, (5) genitourinary tumor.

Liu et al. [11] investigated the prevalence of all cancers and specific cancer types in ID using a population-based cohort study in Sweden. Authors found an increased risk of any cancer, as well as of several specific cancer types for patients with ID. The types of cancer that were statistically significantly more common in patients with ID were gastrointestinal tract cancers, uterus, kidney, central nervous system, acute lymphoid leukemia, and acute myeloid leukemia.

Sullivan et al. [12] studied on the information from the Disability Services Commission of Western Australia and State Cancer Registry. The incidence of all cancers in people with ID was not significantly different from the general population. However, males with ID presented a significant increase in the diagnosis of leukemia, brain and stomach cancers, and a decreased risk of prostate cancer. Females with ID presented a significant increase in the diagnosis of leukemia, corpus uteri, and colorectal cancers.

The results of these population-based studies with large samples were consistent with our study. Similarly, leukemia and brain cancers were found to be part of the 5 highest reported cancers. In the population study that found higher rates of brain tumors, the answer to the question of whether ID developed due to brain tumors was

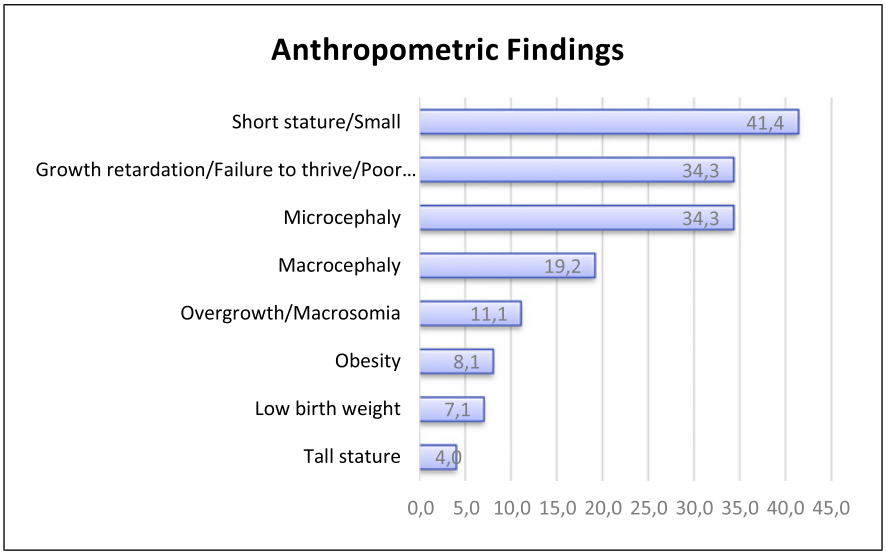

Fig. 2. Percentages of anthropometric findings.

Table 3. Involved anthropometric findings and associated genes

	Genes
Obesity	*CREBBP, WT1, PTEN, XRCC4, SMAD4, KRAS, TP53, MADH4, STK11, GNAS, PRKAR1A*
Overgrowth/macrosomia	*FIBP, DIS3L2, SUFU, DICER1, FIBP, HRAS, AKT1, PIK3CA, EGFR, BRAF*
Tall stature	*GPC3, FIBP, DNMT3A, SUFU*
Macrocephaly	*DICER1, PTEN, HRAS, EXT2, GPC3, MYCN, MAX, AXIN1, NSD1, FIBP, NF1, FH, AKT1, PIK3CA, NRAS, KRAS, EGFR, BRAF, SUFU, BAP1,*
Microcephaly	*BRCA1, CREBBP, ERCC2, SMAD4, EXT2, TINF2, ATR, KRAS, NBN, SDHD, WWOX, MYCN, TERT, TP53, MADH4, ERCC6, STK11, BUB1B, DKC1, FANCA, FANCE, CTNNB1, NOP10, LIG4, RPS19, FANCD2, ACD, NBS1, RECQL3, TRIP13, DNAJC21, XRCC4, ERCC4, ERCC3*
Growth retardation/failure to thrive/poor overall growth	*CREBBP, ERCC2, BUB1B, DKC1, GBA1, TINF2, PRKAR1A, RAD51, POT1, LIG4, NBN, ERCC6, ATR, RECQL3, MYCN, TRIP13, DNAJC21, KRAS, HRAS, NBS1, ACD, RPS19, HRAS, RET, BRCA1, DNAJC21, ERCC6, SETBP1, VHL, GBA1, TERT, FH, PTPN11, JAG1, SBDS, TET2, RPS19, LIG4, MAP2K1, EGFR, BRAF,BRCA 1, MAD1L1, EGFR, BRAF, BAP1, PAX7*
Low birth weight	*ERCC2, BUB1B, SBDS, FANCD2, FANCA, FANCE, ERCC6*
Short stature/small	*HRAS, BRCA1, CREBBP, ERCC2, ERCC3, ERCC4, ERCC6, PAX7, FANCA, FANCE, ATR, FANCD2, VHL, PRKAR1A, NBN, TINF2, RMRP, AXIN1, BUB1B, EXT2, PPM1D, TERT, GNAS, EGFR, BRAF, NRAS, SMAD4, TP53, MADH4, STK11, MAP2K1, SBDS, SIK3, RPS19, NBS1, CBL, DKC1, PTPN11, RECQL4, TRIP13, DNAJC21, KRAS, TWIST, XRCC4*

investigated [11] because ID may be a result of CNS cancers. However, the authors re-analyzed the data excluding brain tumors that developed within 5 years after the diagnosis of ID. Even in this case, the increased risk of brain cancer was detected. These results indicate that there are common genetic etiological factors between brain cancers and ID.

The prevalence of gastrointestinal system tumors, which we found at much lower rates in our study, ranked first in population studies [12]. However, it should be noted that we only include ID and cancer types within genetic syndromes. Environmental factors have been emphasized as the cause of gastrointestinal tumors associated with ID [12]. These are particularly increased risk of *H. pylori* infection among institutionalized patients [4, 13] obesity [14] and insulin resistance [15].

People with IDs may experience delays in the diagnosis of a neoplastic disorder due to communication

difficulties. To identify individuals with IDs at an increased risk of developing neoplasms, early and hence diagnostic genetic investigations should be applied during routine developmental checks. If a genetic syndrome is identified, we can offer potential appropriate screening procedures for neoplasms. Once the diagnosis is made, having clinical knowledge of increased risk for specific neoplasms should be factored into the regular clinical follow-up of these patients. It is important to follow up the presence of neoplasm regarding early associated symptoms. In future studies, providing cancer screening guidelines for vulnerable populations with disabilities is essential. Cancer screening practices in the 21st century will adapt health care to increased risk, genetic factors, and patient preferences [9]. We especially emphasize the importance of leukemia, brain tumors, and tumors of embryonal origin in individuals with ID.

There are some limitations for this study. It should be kept in mind that we should consider that for some syndromes the increased risk of a particular neoplasm type is much higher than others. The rates of the increase in cancer development may vary from one genetic syndrome to others. Therefore, the risk of developing neoplasm between syndromes is not equal. We need further studies investigating rates of cancer related to specific genetic syndromes that we described in our study to provide appropriate guidance for families. There are not enough studies to determine whether high-risk candidates should be screened. Future studies should focus on identifying those who will and will not benefit most from cancer screening [16]. Another limitation of our study is that the clinical information of the genetic syndromes was screened in the OMIM database and there are differences in description of the symptoms among the syndromes. We included these original descriptions adapted from OMIM database in our study as they are in order not to distort the original. However, to deal with this limitation, we performed our analysis as grouping them in a standardized manner.

Conflict of Interest Statement

The authors declare that they have no conflict of interest.

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Author Contributions

Gül Ünsel-Bolat: constructing the idea of research, planning methodology to reach the conclusions, organizing, supervising the course of progress and taking the responsibility of the research, taking responsibility in the writing of the whole of the study, and reviewing the article before submission scientifically besides spelling and grammar. Esra Dutar: constructing the idea of research, planning methodology to reach the conclusions, organizing, supervising the course of progress and taking the responsibility of the research, taking responsibility in necessary literature review for the study, and taking responsibility in the writing of the whole of the study. Hilmi Bolat: constructing the idea of research, planning methodology to reach the conclusions, organizing, supervising the course of progress and taking the responsibility of the research, and reviewing the article before submission scientifically besides spelling and grammar.

References

1 Gil N, Cox A, Whitaker KL, Kerrison RS. Cancer risk-factor and symptom awareness among adults with intellectual disabilities, paid and unpaid carers, and healthcare practitioners: a scoping review. J Intellect Disabil Res. 2024;68(3):193–211. https://doi.org/10.1111/jir.13110

2 COST action CA21123 cancer – understanding prevention in intellectual disabilities (CUPID), Brussels, 27 May 2022, COST 037/22.

3 Banda A, Naaldenberg J, Timen A, van Eeghen A, Leusink G, Cuypers M. Cancer risks related to intellectual disabilities: a systematic review. Cancer Med. 2024;13(9):e7210. https://doi.org/10.1002/cam4.7210

4 Wallace RA, Webb PM, Schluter PJ. Environmental, medical, behavioural and disability factors associ- ated with Helicobacter pylori infection in adults with intellectual disability. J Intellect Disabil Res. 2002;46(Pt 1):51–60. https://doi.org/10.1046/j.1365-2788.2002.00359.x

5 Caton S, Chadwick D, Chapman M, Turnbull S, Mitchell D, Stansfield J. Healthy lifestyles for adults with intellectual disability: knowledge, barriers, and facilitators. J Intellect Dev Disabil. 2012;37(3):248–59. https://doi.org/10.3109/13668250.2012.703645

6 Robison LL, Nesbit ME Jr, Sather HN, Level C, Shahidi N, Kennedy A, et al. Down syndrome and acute leukemia in children: a 10-year retrospective survey from Childrens Cancer Study Group. J Pediatr. 1984;105(2):235–42. https://doi.org/10.1016/s0022-3476(84)80119-5

7 Phelan MC, Stevenson RE, Collins JL, Trent HE 3rd. Fragile X syndrome and neoplasia. Am J MedGenet. 1988;30(1–2):77–82. https://doi.org/10.1002/ajmg.1320300106

8 Al-Saleem T, Wessner LL, Scheithauer BW, Patterson K, Roach ES, Dreyer SJ, et al. Malignant tumors of the kidney, brain, and soft tissues in children and young adults with the tuberous sclerosis complex. Cancer. 1998;83(10):2208–16.

9 Loud J, Murphy J. Cancer screening and early detection in the 21st century, HHS Public Access. Semin Oncol Nurs. 2017;33(2):121–8. https://doi.org/10.1016/j.soncn.2017.02.002

10 Rethoré M-O, Rouëssé J, Satgé D. Cancer screening in adults with down syndrome, a proposal. Eur J Med Genet. 2020;63(4):103783. https://doi.org/10.1016/j.ejmg.2019.103783

11 Liu Q, Adami HO, Reichenberg A, Kolevzon A, Fang FID, Sandin S. Cancer risk in individuals with intellectual disability in Sweden: a population-based cohort study. PLoS Med. 2021;18(10):e1003840. https://doi.org/10.1371/journal.pmed.1003840
12 Sullivan SG, Hussain R, Threlfall T, Bittles AH. The incidence of cancer in people with intellectual disabilities. Cancer Causes Control. 2004;15(10):1021–5. https://doi.org/10.1007/s10552-004-1256-0
13 Duff M, Scheepers M, Cooper M, Hoghton M, Baddeley P. Helicobacter pylori: has the killer escaped from the institution? A possible cause of increased stomach cancer in a population with intellectual disability. J Intellect Disabil Res. 2001;45(Pt 3):219–25. https://doi.org/10.1046/j.1365-2788.2001.00302.x
14 Turner S, Moss S. The health needs of adults with learning disabilities and the health of the nation strategy. J Intellect Disabil Res. 1996;40(5):438–50. https://doi.org/10.1111/j.1365-2788.1996.tb00650.x
15 Higginbotham S, Zhang ZF, Lee IM, Cook NR, Giovannucci E, Buring JE, et al. Dietary glycemic load and risk of colorectal cancer in the Women's Health Study. J Natl Cancer Inst. 2004;96(3):229–33. https://doi.org/10.1093/jnci/djh020
16 Yourman LC, Lee SJ, Schonberg MA, Widera EW, Smith AK. Prognostic indices for older adults: a systematic review. JAMA. 2012;307(2):182–92. https://doi.org/10.1001/jama.2011.1966