Contents lists available at Science-Gate

International Journal of Advanced and Applied Sciences

Journal homepage: https://www.science-gate.com/IJAAS.html

Enhancing teachers' awareness of the genetic aspects of rare syndromes in students with intellectual disabilities: An experimental study

Khaled Awad Elballah [1], Hussein Ahmed Shahat [2,*]

[1]Department of Special Education, College of Education, King Faisal University, Al-Ahsa, Saudi Arabia
[2]Department of Art Education, College of Education, King Faisal University, Al-Ahsa, Saudi Arabia

ARTICLE INFO

Article history:
Received 19 July 2026
Received in revised form
24 September 2026
Accepted 26 October 2026

Keywords:
Genetic awareness
Intellectual disabilities
Rare syndromes
Teacher training
Genetic counseling

ABSTRACT

The varying symptoms and manifestations of physical and mental development among students with intellectual disabilities (ID) are a concern that can lead to negative consequences, including poor academic performance, limited social and behavioral skills, and social health problems. This study employed an experimental approach to enhance genetic awareness among teachers of students with ID. A researcher-developed genetic awareness scale was used, along with a mentoring program comprising sessions aimed at increasing the cognitive and professional awareness of teachers in supporting interactions with students with rare syndromes (RSs) and ID in the school environment and understanding the implications of genetic abnormalities in each case. The researchers administered a questionnaire containing specific questions to 62 teachers working in inclusive public elementary schools and special centers within intellectual disability programs. The findings represent an initial step toward increasing awareness and knowledge of the genetic causes, types, and manifestations of RSs in the school context, thereby helping specialist teachers manage these cases effectively according to the capabilities and characteristics of each syndrome. The results showed significant differences between the pre- and post-intervention scores on the genetic awareness scale for RSs. Considering teachers' perspectives can contribute to the development and implementation of individualized programs that effectively support students with RSs in elementary school intellectual disability programs. A strategy for raising awareness of these syndromes through genetic counseling was also adopted.

© 2026 The Authors. Published by IASE. This is an open access article under the CC BY-NC-ND license (https://creativecommons.org/licenses/by-nc-nd/4.0/).

1. Introduction

The underlying cause of intellectual disability (ID) is mostly genetic in origin; the prevalence of ID is approximately 2–3% of the world's population (Jansen et al., 2023). Furthermore, estimates suggest that approximately 2000, or 10%, of all 20,000 human genes may be involved (Chiurazzi and Pirozzi, 2016; Vissers et al., 2016). Since the identification of the double helix in 1953 and the exact number of chromosomes in 1955, the diagnostic possibilities in ID have increased enormously (Jansen et al., 2023). ID can be caused by exogenous factors such as maternal alcohol abuse during pregnancy, infections, birth complications, and extreme malnutrition, but genetics is known to have an important role in its etiology (Vissers et al., 2016). ID is both clinically and genetically heterogeneous; clinical heterogeneity is reflected in the diversity of different ID disorders but also by the clinical variability in a specific disorder caused by mutations in the same gene (Sadleir et al., 2017). Although there is a five percent rate of various disabilities, there are no accurate statistics on the number of people with RSs, especially in the field of intellectual and developmental disabilities. Rare diseases are defined by the European Union as life-threatening or chronically debilitating diseases that are of such low prevalence (less than 5 per 10,000) that special combined efforts are needed to address them (Moliner and Waligora, 2017).

While Down Syndrome (DS) is associated with extra genetic material—instead of 46 chromosomes, a person with DS has a full or partial extra copy of a chromosome, delaying their development—it is not considered a rare syndrome, as one in every 554

* Corresponding Author.
Email Address: h.ali@kfu.edu.sa (H. A. Shahat)
https://doi.org/10.21833/ijaas.2026.10.017
Corresponding author's ORCID profile:
https://orcid.org/0000-0001-9650-6417
2313-626X/© 2026 The Authors. Published by IASE.
This is an open access article under the CC BY-NC-ND license (https://creativecommons.org/licenses/by-nc-nd/4.0/)

children in the Kingdom of Saudi Arabia is born with DS, and 20,000 Saudi children under the age of 17 have DS (Alabri, 2023). ID is defined as a defect in cognitive functioning and adaptive behavior that originates before the age of 18 years and has a worldwide prevalence of 2–3% (Pande et al., 2024). The genetic etiology of disorders of ID is highly heterogeneous, encompassing a wide spectrum of genetic variations, including structural variants (SVs), copy number variants (CNVs), small insertions/deletions, and single-nucleotide variants (SNVs), identified across more than 1000 genes (Kochinke et al., 2016).

Monogenic disorders contribute to 30–50% of cases of ID; around 20% are due to disease-causing large or small CNVs, and the cause of 50% of cases remains unknown. ID or delayed psychomotor development are by far the most common reasons for referral to genetic services, and most severe forms are caused by single genetic defects (Hu et al., 2019).

In Western societies, where consanguineous marriage between parents is rare, unlike in Eastern societies, most affected individuals are rare cases. Dominant de novo copy number variants (CNVs) and mutations in single genes account for ID in the majority of these individuals (Musante and Ropers, 2014). The development of the genome in the past two decades, especially in the field of diagnosis, has led to the discovery of a number of syndromes. Recent studies show that the prevalence of ID is significantly correlated with parental consanguinity, being 3-4 times higher (Hu et al., 2019). RSs such as Rubinstein-Taybi syndrome occur in 1:125,000 live births. While individual syndromes are rare, a significant number of individuals are affected by genetic syndromes associated with ID (Waite et al., 2014).

Therefore, in this research, we try to identify the level of awareness and knowledge of teachers and specialists about RSs in ID, in terms of the causes and number of genes responsible for their occurrence, as well as the differences in the external manifestations of each case. Then, we present a vision of how to deal positively on the behavioral and social levels and the individual educational programs that inclusive education schools can provide through a counseling program including 10 sessions. There is a significant gap in the literature concerning teachers' experiences and perceptions of RSs of special education specialists in Saudi Arabia. The author describes 20 RSs associated with ID. While some, such as Angelman Syndrome, Prader-Willi Syndrome, Williams Syndrome, and Klinefelter Syndrome, are relatively well-known, others, like Rubinstein-Taybi Syndrome, Laron Syndrome, Beckwith-Wiedemann Syndrome, Smith-Magenis Syndrome, and Patau Syndrome, are less commonly recognized.

The students with IDs include a variety of cognitive, emotional, behavioral, physical, and learning disabilities and can be classified based on general quotient and behavioral adjustment.

2. Literature review

2.1. Rare genetic syndrome in intellectual disabilities

Rare genetic disorders have a prevalence of less than 2,000. An estimated 6,000 to 7,000 rare diseases have been identified, including those with characteristics of ID, developmental disorders, or behavioral disorders. Some RSs are not related to their popularity in the scientific or general community; for example, Huntington's disease is more well-known than Williams syndrome (Roubertoux and de Vries, 2011). Table 1 describes 20 RSs of ID in terms of the name of the syndrome, the genetic cause, and its characteristics associated with ID.

In the following section, the researchers review some details about RSs in ID as follows. Cat meow syndrome is named for its distinctive cry, similar to a cat's meow, and its prevalence is estimated at 1:50,000 live births. The degree of ID ranges from severe to moderate (Griffith et al., 2011). It is caused by a congenital disease resulting from a split or missing arm of one pair of chromosome 5. This genetic abnormality affects the structure of the vocal cords and larynx, leading to a high-pitched, cat-like meow in the affected person's voice. In addition, it can cause mild to severe ID, learning difficulties, language and speech impairments, hearing problems, and respiratory issues. Angelman syndrome is associated with moderate to severe ID, with severe being the most common. Approximately 80% of affected individuals experience seizures (Clayton-Smith and Laan, 2003).

Frequent smiling and laughing, hyperactivity, and sleep disturbance are common behavioral features of the syndrome (Clayton-Smith and Laan, 2003). Happy Doll Syndrome is caused by a genetic defect in gene number 15 in the mother in 70-75% of cases and a mutation in the UBE3A gene located on chromosome segment q11-q13. Physical and behavioral features of the disease appear between the ages of three and six, such as frequent laughing and smiling, feeding problems, sleep disturbances, hyperactivity, and short attention span (Griffith et al., 2011; Pearson et al., 2019). Alagille syndrome is an embryopathy due to mutations in the gene JAG1. Neonatal cholestasis is a main feature (Turnpenny and Ellard, 2012). Several features such as ID, sagging, feeding problems, obesity, incomplete genitalia, short stature, and delayed growth characterize Prader-Willi syndrome. It is caused by a deletion on chromosome 15 inherited from the father and is designated as 15q11. q13. In 70% of cases, the deficiency in the paternal chromosome is present. 35% of cases come from maternal chromosome 15, without the presence of the paternal chromosome (Butler et al., 2019).

Turner's syndrome affects females at a rate of 1/5000 of female births and causes mental disability and delayed physical and sexual development. Apert syndrome is a rare congenital malformation caused

by a genetic mutation that results in abnormal growth of the bones of the skull and face; fusion of the fingers and toes; ID; bulging eyes; a small nose; low ears; visual and hearing problems; and defects in the heart, urinary, and reproductive systems. Edwards syndrome's cause is chromosome 18, resulting from an increase in the total number of chromosomes, where the number is 47 chromosomes in group 18, whether this increase is complete or partial. Cases of the first type of mutation, or the mosaic type, are less severe and symptomatic and therefore can live longer (Remolina et al., 2024).

Table 1: Rare syndromes, genetic causes, and characteristics related to intellectual disability

Genetic condition	Genetic cause	Characteristics related to intellectual disability
Cri-du-chat syndrome	Deletion of the short arm of chromosome 5 (5p deletion).	Developmental delay and intellectual disability of variable severity; speech and language impairment are common.
Angelman syndrome	Loss of function or expression of the maternally expressed UBE3A gene in the brain, commonly involving 15q11.2–q13.	Severe developmental delay, severe speech impairment, and usually severe intellectual disability.
Alagille syndrome	Pathogenic variants in JAG1; less commonly, NOTCH2.	Developmental and learning difficulties may occur, but intellectual disability is not a defining feature in every affected person.
Prader–Willi syndrome	Loss of expression of paternally inherited genes in the 15q11.2–q13 imprinted region.	Developmental delay and learning difficulties; intellectual disability ranges in severity.
Turner syndrome	Complete or partial monosomy X, commonly 45,X; other sex-chromosome patterns also occur.	Specific learning difficulties, particularly in visuospatial and mathematics domains, may occur; global intellectual disability is not typical.
Apert syndrome	Usually a pathogenic variant in FGFR2.	Developmental delay and learning difficulties may occur; cognitive outcomes vary.
Edwards syndrome	Trisomy 18, including full, mosaic, or partial forms.	Severe developmental impairment and profound intellectual disability are common among individuals who survive beyond infancy.
Klinefelter syndrome	Usually an additional X chromosome in a male karyotype (47,XXY).	Language-based learning difficulties and executive-function challenges may occur; intellectual disability is not typical.
DiGeorge syndrome (22q11.2 deletion syndrome)	Deletion in chromosome region 22q11.2.	Learning difficulties and variable cognitive impairment; intellectual disability occurs in a subset.
Cornelia de Lange syndrome	Pathogenic variants in genes including NIPBL, SMC1A, SMC3, RAD21, and HDAC8; NIPBL is the most commonly affected gene.	Developmental delay and intellectual disability of variable severity; speech and communication difficulties are common.
Kabuki syndrome	Pathogenic variants in KMT2D or KDM6A.	Developmental delay and intellectual disability of variable severity; learning and language difficulties may occur.
Patau syndrome	Trisomy 13, including full, mosaic, or partial forms; karyotypes may include 47,XX,+13 or 47,XY,+13.	Severe developmental impairment and profound intellectual disability are common among individuals who survive beyond infancy.
Rubinstein–Taybi syndrome	Pathogenic variants in CREBBP or EP300, or deletions involving these genes.	Developmental delay and intellectual disability, commonly ranging from mild to severe.
Coffin–Lowry syndrome	Pathogenic variants in RPS6KA3 on Xp22.2.	Intellectual disability is usually more severe in males; females may have variable, often milder, cognitive effects.
Fragile X syndrome	CGG-repeat expansion in FMR1 at Xq27.3, usually causing gene silencing.	Intellectual disability is variable and is generally more severe in males; females may have learning and cognitive difficulties.
Phenylketonuria	Usually biallelic pathogenic variants in PAH at 12q23.2; untreated disease causes elevated phenylalanine.	Untreated phenylketonuria can cause severe intellectual disability and neurological impairment; early dietary treatment can prevent much of this outcome.
Williams syndrome (Williams–Beuren syndrome)	Microdeletion at chromosome region 7q11.23, typically including ELN.	Developmental delay and a characteristic cognitive profile; intellectual disability is commonly mild to moderate.
Laron syndrome	Usually biallelic pathogenic variants in GHR causing growth hormone insensitivity.	Severe short stature is characteristic; intellectual disability is not a defining feature.
Beckwith–Wiedemann syndrome	Abnormal regulation of imprinted genes in chromosome region 11p15.5, including changes in methylation or copy number; some cases involve CDKN1C variants.	Overgrowth and tumor predisposition are characteristic; intellectual disability is not a typical defining feature.
Smith–Magenis syndrome	Usually a deletion of chromosome region 17p11.2 including RAI1, or a pathogenic variant in RAI1.	Developmental delay and intellectual disability of variable severity, with prominent behavioral and sleep-related features.

Cases that show symptoms such as small size at birth, ID, small mouth and lower jaw, ear defects, congenital heart defects, and undescended testicles. Klinefelter syndrome (KS) is caused by a random error that causes a male to be born with an extra sex chromosome. Symptoms include breast enlargement, lack of facial hair, small testicles, and inability to conceive. It occurs in about 1 in 8,000 males and occurs only in males. Chromosome 23 becomes triploid instead of diploid. An extra copy of the X chromosome in each cell (XXY) is the most common cause. Symptoms also include delayed walking and speech, delayed mental development in the child, and delayed onset of secondary symptoms in males (Gravholt et al., 2018). DiGeorge syndrome results from a hemizygous deletion on chromosome 22q11.2. The region carries Drosha homologues (DGCR8) that are crucial in miRNA biogenesis. DGCR8 itself has a direct effect on brain function and cognition, as shown by the observation that insertional inactivation of the gene induces learning impairment. Cornelia de Lange syndrome: The syndrome is estimated to affect 1 in 40,000 live births. Health problems, including gastrointestinal disorders, are a dominant feature (Luzzani et al., 2003).

Common behavioral features include anxiety, hypersensitivity, self-stimulation, self-injurious behavior, and compulsivity (Griffith et al., 2011). Kabuki Syndrome is a rare genetic disorder characterized by distinctive facial features, ID, long palpebral fissures with eversion of the lateral third of the lower eyelid, arched and wide eyebrows with a rare lateral third, postnatal growth failure, and

genitourinary anomalies (Lee et al., 2024). It is characterized by signs such as feeding problems, anemia, immunodeficiency, and seizures. Congenital heart and dental anomalies, hearing impairment, and they often show signs of happiness. They require intervention to enhance speech and education (Lehman et al., 2017). Patau syndrome (trisomy 13) is an old condition in terms of clinical diagnosis. The causative chromosome 13 was identified in 1960. It is considered one of the least common trisomies due to the high rate of miscarriage in this condition. The mortality rate is also very high, and severe ID is a constant feature in these cases. Its symptoms include cleft lip, cleft palate, ear deformity, microcephaly, and an increased number of fingers and toes (Satgé et al., 2017).

PS involves multiple congenital anomalies and significant ID, making care challenging for the family, primary care practitioners, and specialists. Rubinstein-Taybi Syndrome (RSTS): This syndrome shares similar physical characteristics of delayed mental and motor development, distinctive facial features, and large thumbs and index fingers on the hands and feet, which is considered a genetic mutation in most cases. This condition is characterized by short stature, curvature of the spine, a prominent nose, outward or inward slanting of the eyes, a distinctive appearance of the hands and feet, which is one of the main characteristics of this syndrome, and large thumbs on both hands and feet or large fingers on all fingers accompanied by strabismus or hearing impairment and congenital heart defects (Tekendo-Ngongang et al., 2020). Coffin-Lowry syndrome (CLS) is a rare genetic disorder characterized by ID; abnormalities of the head and face; large, soft hands with short, thin fingers; short stature; and various skeletal abnormalities. An abnormally prominent eyebrow, down-slanting eyelid folds, and large ears.

Fragile X Syndrome is the most common cause of ID caused by mutations affecting a single gene. The gene involved is FMR1 (Fragile X Mental Disabilities-1), located on the X chromosome at Xq27.3 (Roubertoux and de Vries, 2011). Phenylketonuria, which is caused by mutations of the phenylalanine hydroxylase gene (12q23.2-q24.1), results in severe cognitive impairment and behavioral disorders. A special diet to prevent the metabolic disorder from birth reduces the cognitive and behavioral symptoms and improves actual cognitive processes. Williams-Beuren Syndrome (WBS): One in 20,000 to 50,000 live births is due to a missing piece of chromosome 7 containing 15 or more genes. A mutation in the elastin gene is thought to account for about 95% of Williams cases. Symptoms include ID in 75% of cases, low birth weight, congenital heart defects, sensitivity to light and loud sounds, low thyroid hormone, abnormal tooth development, a distinctive facial shape (microgyny), and small head size at birth (Kruszka et al., 2018).

Laron Syndrome: This is a genetic disease that results from a defect in the growth hormone receptors. The deficiency of a substance secreted by the liver called insulin-like growth factor 1, which plays a role in stimulating cells to grow, causes people with this disease to be very short in stature and have infantile facial features even after adulthood, with small hands and feet (Janecka et al., 2016; Laron, 2015). Beckwith-Wiedemann Syndrome (BWS), a condition that usually occurs as a result of a genetic mutation through dominant inheritance, has symptoms that include premature birth, enlargement of the body and tongue, a red skin birthmark on the face, bulging eyes, congenital heart defects, seizures, and ID.

Smith-Magenis Syndrome (SMS) is a rare neurobehavioral disorder characterized by a recognizable pattern of physical, behavioral, and developmental features. It is caused by specific genetic changes in chromosomal region 17p11.2, which contains the RAI1 gene. Smith-Magenis syndrome is a rare disorder that occurs in 1 in 15,000 to 25,000 births. Common features include developmental delay and ID, poor gross and fine motor skills, delayed speech, a distinctive facial appearance, vision problems, middle ear abnormalities, chronic ear infections, hoarseness, decreased sensitivity to pain, and hypersensitivity (Smith and Gropman, 2021).

Twenty RSs were reviewed, and the author found other types of RSs that are accompanied by different symptoms and cause ID, developmental and language delay, epilepsy, and physical problems, such as short stature, sensory problems, cardiovascular problems, and kidney problems. Fragile X Syndrome is the most common cause of ID caused by mutations affecting a single gene. The gene involved is FMR1 (Fragile X Mental Disabilities-1), located on the X chromosome at Xq27.3 (Roubertoux and de Vries, 2011). Phenylketonuria, which is caused by mutations of the phenylalanine hydroxylase gene (12q23.2-q24.1), results in severe cognitive impairment and behavioral disorders. A special diet to prevent the metabolic disorder from birth reduces the cognitive and behavioral symptoms and improves actual cognitive processes. Williams-Beuren Syndrome (WBS): One in 20,000 to 50,000 live births is due to a missing piece of chromosome 7 containing 15 or more genes. A mutation in the elastin gene is thought to account for about 95% of Williams cases. Symptoms include ID in 75% of cases, low birth weight, congenital heart defects, sensitivity to light and loud sounds, low thyroid hormone, abnormal tooth development, a distinctive facial shape (microgyny), and small head size at birth (Kruszka et al., 2018). Laron Syndrome: This is a genetic disease that results from a defect in the growth hormone receptors.

The deficiency of a substance secreted by the liver called insulin-like growth factor 1, which plays a role in stimulating cells to grow, causes people with this disease to be very short in stature and have infantile facial features even after adulthood, with small hands and feet (Janecka et al., 2016; Laron, 2015). Beckwith-Wiedemann Syndrome (BWS), a condition that usually occurs as a result of a genetic

mutation through dominant inheritance, has symptoms that include premature birth, enlargement of the body and tongue, a red skin birthmark on the face, bulging eyes, congenital heart defects, seizures, and ID. Smith-Magenis Syndrome (SMS) is a rare neurobehavioral disorder characterized by a recognizable pattern of physical, behavioral, and developmental features. It is caused by specific genetic changes in chromosomal region 17p11.2, which contains the RAI1 gene. Smith-Magenis syndrome is a rare disorder that occurs in 1 in 15,000 to 25,000 births. Common features include developmental delay and ID, poor gross and fine motor skills, delayed speech, a distinctive facial appearance, vision problems, middle ear abnormalities, chronic ear infections, hoarseness, decreased sensitivity to pain, and hypersensitivity (Smith and Gropman, 2021). Twenty RSs were reviewed, and the author found other types of RSs that are accompanied by different symptoms and cause ID, developmental and language delay, epilepsy, and physical problems, such as short stature, sensory problems, cardiovascular problems, and kidney problems.

2.2. Genetic counselors' roles in rare genetic syndromes in intellectual disabilities

The increasing application of diagnostic genetic testing in medicine leads to an increasing need for a "genomic workforce" to deliver care to the affected individuals and their families (Schaaf, 2021). This includes genetic tests, pre-test education, counseling and consenting, and interpretation of diagnostic findings. Improving the quality of teacher training and genetic counseling education for effectively providing genetic counseling (Kohzaki, 2014). Genetic counseling professionals can provide information to families and caregivers about rare mental retardation syndromes through counseling sessions or workshops. Genetic counseling requires a specialist in chromosome and gene testing. We need qualified professionals to provide information on chromosome and genetic testing. This work can be done by pediatricians or internal medicine specialists, medical researchers, or nurses who have been trained in genetic counseling. There is one genetic counselor for every 220,000 people in Japan and Europe, and in some countries, it may be one for every 3,700,000 (Kohzaki, 2014).

There is no accurate data about genetic counselors in many Arab countries. Genetic counselors describe a feeling of responsibility towards the family of their patients, wishing to ensure that they are informed and aware of the implications of a new genetic diagnosis (Hodgson and Gaff, 2013). Genetic counselors can support children with ID by facilitating therapies, special needs schooling, and extending benefits in daily life and family life. Understanding ID schooling needs helps them provide appropriate referrals and address educational concerns (Hodgson and Gaff, 2013; Shrivastava et al., 2016). The roles of genetic counselors are to help families adjust to genetic diagnoses and facilitate access to health care resources and social support. They provide psychosocial support to the patient and their family (Kohzaki, 2014). Once a child is diagnosed with ID, appropriate interventions such as rehabilitation should be implemented to prevent further deterioration of the condition (Shrivastava et al., 2016).

Genetic counselors work within a multidisciplinary team (Kohzaki, 2014). Genetic counseling primarily focuses on discussing genetic issues with families. However, when teachers and specialists have sufficient knowledge about ID syndromes, including their causes, manifestations, and developmental, cognitive, and educational complexities, they can contribute more effectively. Their understanding helps in determining appropriate ways to support each case individually, facilitating communication, and developing suitable intervention and training programs. This collaborative approach plays a crucial role in designing individualized educational plans. Without genetic counseling, teachers and specialists have a severe lack of awareness of the challenges facing students when making educational decisions (Hodgson and Gaff, 2013).

3. Methodology

A quasi-experimental design, as the research aimed to enhance genetic awareness among teachers of students with intellectual disabilities through a counseling program. Therefore, the approach was appropriate for this research. Accordingly, a two-group design was used, with the experiment being conducted on an experimental group of teachers, while the control group received no further intervention.

4. Data collection

This study used the Genetic Awareness Questionnaire for Teachers of Intellectual Disabilities (GAQ) (developed by the researchers) to examine the awareness of teachers and specialists regarding RSs in inclusive schools serving students with ID. Furthermore, a counseling program comprises four units, including twelve counseling sessions focusing on the causes of genetic diseases, including intellectual disability syndromes; methods for identifying children with RSs; communication strategies with children with RSs; and educational and behavioral services according to individualized education plans.

4.1. Data analysis

The current study employed a quantitative research methodology. Data were collected through an online questionnaire that included demographic and specific questions. The questionnaire aimed to

assess teachers' knowledge and awareness of RSs in intellectual disability, including their symptoms, genetic causes, and various communication and intervention strategies for each syndrome within the framework of Individualized Education Plans (IEPs). The questionnaire questions were designed to enable participants to express their perceptions and knowledge about RSs in inclusive elementary schools. This was complemented by a six-week counseling program, with two sessions per week. The research objectives were achieved using a design with two groups, experimental and control, to answer the research questions, using SPSS after collecting the data. A t-test was selected for independent samples to measure the difference between the average scores of the two groups and to measure the difference between the average scores of the experimental group in the pre- and post-tests and the post- and follow-up tests.

4.2. Participants

The participants consisted of 62 teachers and specialists, divided in to 31 experimental group and 31 control group, working in inclusive public primary schools in Al-Ahsa, Saudi Arabia. The participants taught children aged 8–12 years in inclusive programs. The demographic data are available for the participants in Table 2. Participants were not offered any compensation. This distribution included teachers with a wide range of experience, from early-career professionals to highly experienced teachers, with an educational background of bachelor's to master's degrees and no doctorates.

Table 2: Participants' demographic and professional characteristics

Variable	Category	n	%
Study group	Experimental group	31	50.00
	Control group	31	50.00
	Total	62	100.00
Academic qualification	Bachelor's degree	39	62.90
	Diploma	16	25.81
	Master's degree	7	11.29
	Total	62	100.00
Years of teaching experience	Less than 5 years	18	29.03
	5–10 years	21	33.87
	11–20 years	15	24.19
	21–30 years	5	8.06
	More than 30 years	3	4.84
	Total	62	100.00

4.3. Research instruments

4.3.1. Genetic awareness questionnaire (GAQ)

It includes items to assess participants' awareness of RSs in individuals with ID. The questionnaire included a section to evaluate participants' demographic information, including gender, educational level, years of experience, and educational qualifications. This questionnaire has 22 items, including four dimensions: genetic information (6 items), gene transmission (5 items), identification of rare syndromes (7 items), and genetic testing (4 items). A four-point Likert scale was used to answer these items: Always = 4, Often = 3, Sometimes = 2, Rarely = 1. The highest score was 88, and the lowest was 22. The highest score indicates a high level of awareness of rare syndromes in intellectual disability, while the lowest score indicates a lack of awareness. The scale's internal consistency was calculated on a sample of 60 teachers. Correlation coefficients between the scale items and the total score ranged from 0.567 to 0.895. Its reliability was calculated using Cronbach's alpha and the split-half method with Spearman-Brown and Gutman equations. Cronbach's alpha was 0.847. The split-half reliability coefficient was 0.833 using the Spearman-Brown equation and 0.767 using the Gutman equation. This indicates that the scale has high validity and reliability.

4.3.2. Counseling program (CP)

The researchers developed a counseling program aimed at enhancing genetic awareness among teachers of children with ID. Developing this awareness among these teachers is crucial for enhancing their skills and abilities in teaching and guiding children with RSs. To verify the effectiveness of the guidance program, the researchers presented the initial version to a number of educational experts. The researchers divided the program's procedural objectives into three sections: (1) Cognitive objectives, which included enabling teachers to deduce the genetic causes behind each child's individual condition, to pay attention to new developments in the field of genetics, to be interested in topics related to genetic diseases, and to understand that scientific knowledge about genetic disorders contributes to effective communication and interaction with children with RSs and ID. (2) Skills development objectives, which included encouraging teachers to attend and participate in training courses in the field of genetics and genetic disorders. He chooses activities that develop his skills in this field, such as participating in the preparation of training or mentoring programs focused on genetic counseling or participating in a multidisciplinary diagnostic team for children with ID. (3) Emotional objectives: These include the teacher's sense of excitement upon learning about a new field, namely genetics; achieving success in it; being motivated to continue working in it; and pursuing further specialized studies in it through graduate programs. The mentoring program employed a variety of methods, including dialogue and discussion, modeling, cooperative learning, brainstorming, and reinforcement, grounded in constructivism as a theoretical foundation. It utilized models, videos, images, and engaging in discussion and dialogue. The training program was divided into 12 sessions, held twice a week, with each session lasting 40 minutes. It was further divided into three phases: the introductory phase, the training phase, and finally, the evaluation phase. Table 3 details these sessions in terms of their number, phase, objectives, and methods.

Table 3: Program stages, number of sessions, objectives, and techniques

Program stage	Number of sessions	Program objectives	Techniques used
Stage 1. Introduction	1	Establish rapport between the researchers and teachers; explain the program's aims and procedures.	Dialogue and discussion; brainstorming; reinforcement.
Stage 2. Training	10	Increase teachers' understanding of genes and hereditary factors; encourage awareness of developments in genetics relevant to intellectual disability; develop the ability to recognize rare syndromes; explain genetic causes; use photographs and videos to support syndrome recognition; identify appropriate communication approaches; and identify educational, behavioral, and language-support needs.	Brainstorming; reinforcement; modeling; presentation of models and photographs; cooperative learning; videos.
Stage 3. Evaluation	1	Evaluate teachers' achievement of the program's procedural objectives.	Reinforcement; feedback.

5. Results

Question 1: What is the post-test difference in mean GAQ scores between the experimental and control groups? An independent-samples t-test was used to answer this question. Table 4 indicates a statistically significant difference between the mean GAQ scores of the experimental group EG (M = 63.3) and those of the control group CG (M = 43.1). Regarding the mean scores, this difference favors the larger mean, that of the GAQ (t = 38.1, p < .001).

Question 2: What is the difference between the EG's mean GAQ scores in the pre-test and post-test? A paired-samples t-test was used to answer this question. Table 5 shows a difference between the mean GAQ scores of the EG in the post-test (M = 63.3) and the pre-test (M = 45.5). Regarding the mean scores, this difference favors the larger mean, which is the post-test mean (t = 48.6, p < 0.001), confirming that this difference is statistically significant between the mean scores of the EG in the pre-test and post-test, in favor of the post-test.

Table 4: T-test results comparing post-test GAQ scores between the experimental and control groups

Scale	Group	n	Mean	SD	t	df	p	Cohen's d
GAQ	Experimental	31	63.3	1.77	38.1	60	< 0.001	9.67
	Control	31	43.1	2.37				

Table 5: Paired-samples t-test results comparing pre-test and post-test GAQ scores in the experimental group

Scale	Test	n	Mean	Mean difference	SE of the difference	t	df	p	Cohen's d
GAQ	Pre-test	31	45.5	17.8	0.366	48.6	30	< 0.001	8.73
	Post-test	31	63.3						

Question 3: What is the difference between the EG's mean GAQ scores in the follow-up and post-test? A paired-samples t-test was used to answer this question. Table 6 indicates no difference between the mean GAQ scores of the EG in the post-test (M = 63.3) and follow-up (M = 62.9; t = 0.193, p = 0.062), indicating no statistically significant difference between the mean scores of the EG in the post-test and follow-up.

Table 6: Paired-samples t-test results comparing post-test and follow-up GAQ scores in the experimental group

Scale	Test	n	Mean	Mean difference	SE of the difference	t	df	p
GAQ	Post-test	31	63.3	0.400	0.206	1.942	30	0.062
	Follow-up	31	62.9					

In summary, the results show that there are statistically significant differences between the pre- and post-application of the experimental group in the dimensions of the genetic awareness scale for RSs in people with ID. It shows that the diagnosis of RSs relies on clinical signs, including physical, developmental, and behavioral characteristics, and is confirmed through genetic testing. The physical characteristics of each syndrome can be learned through available information, images, course materials, scientific research, or online resources. Based on the survey results, participants acknowledged that their awareness and knowledge of RSs among individuals with ID were low before the implementation of the counseling program. This was attributed to a lack of sufficient information about the characteristics of each syndrome and the absence of an individualized plan that included details on diagnosis and genetic testing for children with genetic disorders. The results show the need to provide workshops, educational, and guidance courses on genetic counseling, whether for teachers or specialists. There is also a strong justification for the need for knowledge and awareness through training courses on appropriate methods of dealing with different students with RS. This means that knowledge about treatment strategies with these students is very low. Therefore, there was a strong need to implement a counseling program to increase awareness of RSs among the intellectually disabled.

6. Discussion

Previous studies (Chiurazzi and Pirozzi, 2016; Hodgson and Gaff, 2013) have shown that genetic awareness requires significant attention from educational institutions working with children with ID. This is due to the existence of some RSs that necessitate genetic testing to identify the gene responsible for the condition, as well as monitoring language, behavioral, and cognitive development. Undoubtedly, the presence of these RSs reflects linguistic, educational, and psychological difficulties for the children themselves, their parents, and

teachers. Therefore, programs that include activities and knowledge focusing on the names of RSs, methods of identifying them, the diagnostic process, understanding the services needed at various levels, and identifying programs that are beneficial in reducing linguistic, educational, cognitive, and behavioral deficits can be valuable for caregivers, whether teachers in inclusion programs or in specialized centers. We found statistically significant differences between the pre- and post-tests on the genetic awareness scale for teachers of RSs, favoring the post-test. This indicates increased genetic awareness among the experimental group. The ten-session program, which includes group discussions, brainstorming, modeling, visualization, reinforcement, and other activities, may be effective in enhancing genetic awareness by focusing on participants' genetic information, how genes are transmitted, identifying RSs based on physical characteristics, and understanding the services they may need.

In some cases, ID is linked to certain physical shapes or facial features that are easy for the diagnostician, parents, and teachers to recognize, such as Down syndrome and fetal alcohol syndrome, which include small eyes, a small head, a flat face, and a thin upper lip. Therefore, it is easy to recognize these cases early. Common features of Kabuki syndrome include distinctive facial features, intellectual disability, speech delay, hearing impairment, and seizures. The most common clinical characteristics of the genetic and phenotypic spectrum of KS include distinct facial features and ID (Lee et al., 2024). Certain syndromes can also be identified by their external or physical appearance, such as Williams syndrome, Prader-Willi syndrome, Turner syndrome, and Angelman syndrome. Edwards syndrome, also known as trisomy 18, has several clinical features, including facial and skull characteristics, and differences in the hands and feet, such as clenched fists, small thumbs, and clubfoot (Alshami et al., 2020). In general, some syndromes can be identified by their external features.

Greater awareness of RSs among healthcare professionals, teachers, and parents may allow an early diagnosis and, consequently, the multidisciplinary follow-up that children with this syndrome require. Early intervention is more likely to be effective and potentially change a child's developmental path. Small gains early in life could represent a significant difference in the children's future autonomy (Oliveira et al., 2024). Little is known about the difficulties parents and their children with genetic conditions that lead to ID face. These children often have multiple challenges resulting in their varying special schooling needs.

Mental disability is a heterogeneous category with great variation among them. Despite the establishment of criteria for diagnosis and acceptance into integration programs such as IQ and level of adaptive behavior, each student represents a special case in terms of IQ, academic achievement, level of communication, and behavior. Therefore, it is necessary to prepare an individual educational program. This represents a commitment by caregivers to meet the needs of the child, according to a multidisciplinary assessment within the framework of due process rights. RSs represent an additional burden in the case of ID, as they pose complications to the health, educational, and behavioral status of children. Each case requires knowing the strengths and needs and building an individual educational plan and training teachers through workshops on how to recognize and deal with each case. Each person with an RS needs special educational strategies, whether peer learning, cooperative learning, or individual learning. They also need individual educational plans in which the family, special education specialists, and specialists participate. The results of some previous studies indicated that Down syndrome cases differ from those with Brady-Willie, Angelman syndrome, and other syndromes, which require treatment strategies that are appropriate for each syndrome separately (Pearson et al., 2019; Waite et al., 2014). Based on the characteristics of students with RSs, the behavioral and social differences and variations are clear from one case to another, as well as the differences in physical and genetic characteristics. Therefore, training and guidance programs, whether for teachers or families, are necessary to increase awareness of how to deal effectively with these students in integration schools or at home. Among the factors that contributed to the improved genetic awareness of teachers of students with intellectual disabilities was their desire to learn scientific content about genes and the differences they cause in appearance, behavior, language, knowledge, and general health among students with rare syndromes. Also contributing was the program's content, which aimed to simplify genetic information in a way that avoided scientific or medical complexity, thus achieving its goal of increasing awareness and knowledge about these syndromes and the differences between them. This was accomplished through videos, images, discussions, and a program booklet containing images and information about 20 rare syndromes within the context of intellectual disability.

Among the factors that contributed to the program's success and its impact on the level of genetic awareness among the experimental group were identifying needs through a pre-test and pinpointing their strengths and weaknesses. Specific student goals were also set, linked to the characteristics of individuals with rare syndromes. The program content was enhanced with specific, practically applicable information for the classroom environment at the linguistic, behavioral, and social levels, utilizing interactive methods such as presentations, modeling, brainstorming, and discussions accompanied by images. The program content was based on the general characteristics of rare syndromes and their impact on educational, behavioral, and social aspects. For example, Prader-Willi syndrome is characterized by excessive

appetite and temper tantrums, thus requiring a controlled classroom environment and diverse activities (Butler et al., 2019). Angelman syndrome is characterized by laughter, hyperactivity, and language impairment, necessitating activities to release energy, focus attention, and develop language skills (Pearson et al., 2019; Turnpenny and Ellard, 2012). Williams syndrome involves a sociable and well-liked individual with repetitive movements and language impairment, requiring training in social and language skills and self-protection (Kruszka et al., 2018).

7. Conclusions

The study results showed that genetic awareness programs contribute to expanding the knowledge and skills of teachers working in inclusive schools with children with RSs and ID. Teachers' understanding may be limited to their own opinions and experiences regarding risk syndromes RSs in individuals with ID and appropriate interventions, given the scarcity of studies addressing genetic counseling programs. However, the findings of this study can help schools develop effective training programs to increase awareness of the genetic characteristics and common traits of RSs in individuals with ID. Based on these results, the importance of the role of teachers and schools in developing and adopting positive approaches to managing RSs in inclusive primary schools becomes clear. Implementing effective strategies for dealing with individuals with RDs requires providing a safe and supportive learning environment, both psychologically and socially. Schools should implement an integrated therapeutic approach involving geneticists, teachers, administrators, social support networks, and the children with RSs themselves to address the issue in a way that aligns with the school's values and its social and cultural context.

The necessity of conducting genetic testing for children to determine the relationship between physical manifestations and behavioral mystiques and the nature and type of the syndrome to determine intervention programs and educational needs. The problem for teachers and caregivers may be that the diagnostic process may depend on a single clinical, behavioral, or cognitive approach limited to intelligence quotient. It is necessary to rely on an integrated diagnosis and a differential diagnosis according to the diagnostic criteria specified for each syndrome. An assessment file that includes genetic or environmental causes of ID, case history, and extent of deterioration or development may help clarify each child's developmental trajectory. Then develop individual educational plans and programs according to the integrative diagnosis. Genetic testing is essential for children with intellectual disabilities, and parents and teachers should participate in genetic awareness programs through workshops and training courses. Genetic counselors can provide teachers with the knowledge and skills to work with individuals with rare syndromes. Formulating educational goals that are appropriate to the developmental and motor skills associated with each syndrome, modifying the classroom environment to meet the needs of each syndrome, and partnering with families to agree with teachers on educational and behavioral methods according to individual educational plans.

Future research should be conducted using a qualitative approach to examine parental genetic awareness within the Saudi cultural context and its impact on children's cognitive and behavioral development. Research could also explore the perspective of genetic counselors on parental acceptance of genetic testing for children with intellectual disabilities.

List of abbreviations

BWS	Beckwith-Wiedemann syndrome
CG	Control group
CLS	Coffin-Lowry syndrome
CNVs	Copy number variants
CP	Counseling program
df	Degrees of freedom
DGCR8	DiGeorge syndrome critical region gene 8
DS	Down syndrome
EG	Experimental group
FMR1	Fragile X mental disabilities-1
GAQ	Genetic awareness questionnaire
IDs	Intellectual disabilities
IEPs	Individualized education plans
IQ	Intelligence quotient
KS	Klinefelter syndrome
M	Mean
miRNA	MicroRNA
P	Probability value
PS	Patau syndrome
RSs	Rare syndromes
RSTS	Rubinstein-Taybi syndrome
SD	Standard deviation
SE	Standard error
SMS	Smith-Magenis syndrome
SNVs	Single-nucleotide variants
SPSS	Statistical Package for the Social Sciences
SVs	Structural variants
T	t-statistic
WBS	Williams-Beuren syndrome

Acknowledgment

The authors acknowledge the Deanship of Scientific Research at King Faisal University, Saudi Arabia, for financial support for the research, authorship, and publication of the Ambitious Researcher Grant (KFU264689).

Compliance with ethical standards

Ethical considerations

Ethical approval was obtained from the King Faisal University Ethics Committee (Approval No. KFU-REC-2025-JAN- ETHICS3024). Participation was voluntary, and informed consent was obtained from

all participants. Confidentiality and anonymity of participants were guaranteed.

Conflict of interest

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

References

Alabri W (2023). Maternal perspectives: The needs of Saudi families of children with Down's syndrome. Journal of Intellectual Disabilities, 27(1): 221–237. https://doi.org/10.1177/17446295211044406 **PMid:35144500**

Alshami A, Douedi S, Guida M, Ajam F, Desai D, Zales V, and Calderon DM (2020). Unusual longevity of Edwards syndrome: A case report. Genes, 11(12): 1466. https://doi.org/10.3390/genes11121466 **PMid:33297534 PMCid:PMC7762407**

Butler MG, Miller JL, and Forster JL (2019). Prader-Willi syndrome - clinical genetics, diagnosis and treatment approaches: An update. Current Pediatric Reviews, 15(4): 207–244. https://doi.org/10.2174/1573396315666190716120925 **PMid:31333129 PMCid:PMC7040524**

Chiurazzi P and Pirozzi F (2016). Advances in understanding – genetic basis of intellectual disability. F1000Research, 5(F1000 Faculty Rev): 599. https://doi.org/10.12688/f1000research.7134.1 **PMid:27127621 PMCid:PMC4830215**

Clayton-Smith J and Laan L (2003). Angelman syndrome: A review of the clinical and genetic aspects. Journal of Medical Genetics, 40(2): 87–95. https://doi.org/10.1136/jmg.40.2.87 **PMid:12566516 PMCid:PMC1735357**

Gravholt CH, Chang S, Wallentin M, Fedder J, Moore P, and Skakkebæk A (2018). Klinefelter syndrome: Integrating genetics, neuropsychology, and endocrinology. Endocrine Reviews, 39(4): 389–423. https://doi.org/10.1210/er.2017-00212 **PMid:29438472**

Griffith GM, Hastings RP, Nash S, Petalas M, Oliver C, Howlin P, Moss J, Petty J, and Tunnicliffe P (2011). "You have to sit and explain it all, and explain yourself." Mothers' experiences of support services for their offspring with a rare genetic intellectual disability syndrome. Journal of Genetic Counseling, 20(2): 165–177. https://doi.org/10.1007/s10897-010-9339-4 **PMid:21203808**

Hodgson J and Gaff C (2013). Enhancing family communication about genetics: Ethical and professional dilemmas. Journal of Genetic Counseling, 22(1): 16–21. https://doi.org/10.1007/s10897-012-9514-x **PMid:22833230**

Hu H, Kahrizi K, Musante L et al. (2019). Genetics of intellectual disability in consanguineous families. Molecular Psychiatry, 24: 1027–1039. https://doi.org/10.1038/s41380-017-0012-2 **PMid:29302074**

Janecka A, Kołodziej-Rzepa M, and Biesaga B (2016). Clinical and molecular features of Laron syndrome, a genetic disorder protecting from cancer. In Vivo, 30(4): 375–381.

Jansen S, Vissers LELM, and de Vries BBA (2023). The genetics of intellectual disability. Brain Sciences, 13(2): 231. https://doi.org/10.3390/brainsci13020231 **PMid:36831774 PMCid:PMC9953898**

Kochinke K, Zweier C, Nijhof B, Fenckova M, Cizek P, Honti F, Keerthikumar S, Oortveld MAW, Kleefstra T, Kramer JM, Webber C, Huynen MA, and Schenck A (2016). Systematic phenomics analysis deconvolutes genes mutated in intellectual disability into biologically coherent modules. The American Journal of Human Genetics, 98(1): 149–164. https://doi.org/10.1016/j.ajhg.2015.11.024 **PMid:26748517 PMCid:PMC4716705**

Kohzaki H (2014). Problems and their solutions in genetic counseling education in Japan. Frontiers in Public Health, 2: 100. https://doi.org/10.3389/fpubh.2014.00100

Kruszka P, Porras AR, de Souza DH et al. (2018). Williams–Beuren syndrome in diverse populations. American Journal of Medical Genetics Part A, 176(5): 1128–1136. https://doi.org/10.1002/ajmg.a.38672 **PMid:29681090 PMCid:PMC6007881**

Laron Z (2015). Lessons from 50 years of study of Laron syndrome. Endocrine Practice, 21(12): 1395–1402. https://doi.org/10.4158/EP15939.RA **PMid:26401581**

Lee CL, Chuang CK, Chen MR, Lin JL, Chiu HC, Chang YH, Tu YR, Lo YT, Lin HY, and Lin SP (2024). Genetic and phenotypic spectrum of KMT2D variants in Taiwanese case series of Kabuki syndrome. Diagnostics, 14(16): 1815. https://doi.org/10.3390/diagnostics14161815 **PMid:39202303 PMCid:PMC11353766**

Lehman N, Mazery AC, Visier A et al. (2017). Molecular, clinical and neuropsychological study in 31 patients with Kabuki syndrome and KMT2D mutations. Clinical Genetics, 92(3): 298–305. https://doi.org/10.1111/cge.13010 **PMid:28295206**

Luzzani S, Macchini F, Valadè A, Milani D, and Selicorni A (2003). Gastroesophageal reflux and Cornelia de Lange syndrome: Typical and atypical symptoms. American Journal of Medical Genetics Part A, 119A(3): 283–287. https://doi.org/10.1002/ajmg.a.20191 **PMid:12784293**

Moliner AM and Waligora J (2017). The European Union Policy in the Field of Rare Diseases. In: Posada de la Paz M, Taruscio D, and Groft S (Eds.), Rare diseases epidemiology: Update and overview: 561–587. Springer, Cham, Switzerland. https://doi.org/10.1007/978-3-319-67144-4_30 **PMid:29214592**

Musante L and Ropers HH (2014). Genetics of recessive cognitive disorders. Trends in Genetics, 30(1): 32–39. https://doi.org/10.1016/j.tig.2013.09.008 **PMid:24176302**

Oliveira Í, Fernandes A, Pereira M, Rodrigues M, Silva N, and Mendonça C (2024). A rare cause of intellectual disability. Cureus, 16(1): e51451. https://doi.org/10.7759/cureus.51451

Pande S, Majethia P, Nair K et al. (2024). De novo variants underlying monogenic syndromes with intellectual disability in a neurodevelopmental cohort from India. European Journal of Human Genetics, 32: 1291–1298. https://doi.org/10.1038/s41431-023-01513-7 **PMid:38114583 PMCid:PMC7616498**

Pearson E, Wilde L, Heald M, Royston R, and Oliver C (2019). Communication in Angelman syndrome: A scoping review. Developmental Medicine & Child Neurology, 61(11): 1266–1274. https://doi.org/10.1111/dmcn.14257 **PMid:31074506**

Remolina P, Mendoza Barrera AC, Santana Heredia GM, Polanco Ramírez VA, Rodriguez De La Cruz D, and Perez MI (2024). Navigating the challenges of Edwards syndrome: A compelling case study. Clinical Images and Case Reports Journal, 6(6): 400.

Roubertoux PL and de Vries PJ (2011). From molecules to behavior: Lessons from the study of rare genetic disorders. Behavior Genetics, 41: 341–348. https://doi.org/10.1007/s10519-011-9469-y **PMid:21541644**

Sadleir LG, Mountier EI, Gill D et al. (2017). Not all SCN1A epileptic encephalopathies are Dravet syndrome: Early profound Thr226Met phenotype. Neurology, 89(10): 1035–1042.

https://doi.org/10.1212/WNL.0000000000004331
PMid:28794249 PMCid:PMC5589790

Satgé D, Nishi M, Sirvent N, Vekemans M, Chenard MP, and Barnes A (2017). A tumor profile in Patau syndrome (trisomy 13). American Journal of Medical Genetics Part A, 173(8): 2088–2096.
https://doi.org/10.1002/ajmg.a.38294 **PMid:28544599**

Schaaf C (2021). Genetic counseling and the role of genetic counselors in the United States. Medizinische Genetik, 33(1): 29–34.
https://doi.org/10.1515/medgen-2021-2054
PMid:38836204 PMCid:PMC11006247

Shrivastava SR, Shrivastava PS, and Ramasamy J (2016). Addressing the adverse consequences of disability on health status in rural settings. Journal of Neurosciences in Rural Practice, 7(2): 319–321.
https://doi.org/10.4103/0976-3147.178668
PMid:27114676 PMCid:PMC4821953

Smith ACM and Gropman AL (2021). Smith–Magenis syndrome. In: Carey JC, Battaglia A, Viskochil D, and Cassidy SB (Eds.), Cassidy and Allanson's management of genetic syndromes: 863–893. 4th Edition, Wiley-Blackwell, Hoboken, USA.
https://doi.org/10.1002/9781119432692.ch54

Tekendo-Ngongang C, Owosela B, Fleischer N et al. (2020). Rubinstein–Taybi syndrome in diverse populations. American Journal of Medical Genetics Part A, 182(12): 2939–2950.
https://doi.org/10.1002/ajmg.a.61888 **PMid:32985117**

Turnpenny PD and Ellard S (2012). Alagille syndrome: Pathogenesis, diagnosis and management. European Journal of Human Genetics, 20: 251–257.
https://doi.org/10.1038/ejhg.2011.181
PMid:21934706 PMCid:PMC3283172

Vissers LELM, Gilissen C, and Veltman JA (2016). Genetic studies in intellectual disability and related disorders. Nature Reviews Genetics, 17: 9–18.
https://doi.org/10.1038/nrg3999 **PMid:26503795**

Waite J, Heald M, Wilde L, Woodcock K, Welham A, Adams D, and Oliver C (2014). The importance of understanding the behavioural phenotypes of genetic syndromes associated with intellectual disability. Paediatrics and Child Health, 24(10): 468–472. https://doi.org/10.1016/j.paed.2014.05.002