Pediatr Pol 2026; 101 (1): 57-67
DOI: https://doi.org/10.5114/polp.2026.160532
Submitted: 23.02.2025, Accepted: 15.10.2025, Published: 27.03.2026

PEDIATRIA Polska
PISMO POLSKIEGO TOWARZYSTWA PEDIATRYCZNEGO
Polish Journal of PAEDIATRICS
OFFICIAL JOURNAL OF POLISH SOCIETY OF PAEDIATRICS

REVIEW PAPER

A brief guide to the genetics of autism spectrum disorders

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ABSTRACT

Autism spectrum disorders (ASD) constitute a group of etiologically and clinically heterogeneous neurodevelopmental disorders. Generally speaking, all ASD may be divided into non-syndromic and syndromic types. The former, also known as "pure" or "essential" autism, may have or not identifiable genetic background but there are no other features that constitute a recognizable syndrome. The latter, also referred to as "autism +" occurs with specific phenotypes where autistic features are present among other symptoms. All patients diagnosed with ASD should be offered further genetic counseling involving phenotypical and genotypical diagnosis, pedigree analysis and prognosis of future development. Autism spectrum disorders patients need broader, multidisciplinary care and risk assesment for family members. The aim of this article is to provide a succinct guide on how genetics affects all aspects of ASD care and how to utilize genetics as a general practitioner, pediatrician, or other medical professional working with patients affected by autism spectrum disorder.

KEY WORDS:
autism, ASD, genetics.

INTRODUCTION

Autism spectrum disorders (ASD) are a group of heterogeneous neurodevelopmental disorders of variable etiology and diverse phenotypes. Clinical manifestations involve disturbances in communication and social interactions as well as repeatable and restricted behaviors. First signs appear in early childhood and can significantly affect patients' everyday functioning [1]. The prevalence of ASD is estimated to exist in 1% of child global population, however it varies among reports. Centers for Disease Control and Prevention announced that 1 in 44 people worldwide suffer from ASD. World Health Organization, on the other hand, estimates the frequency of ASD at 1 in 160 people [2, 3]. Boys are diagnosed four times as frequently as girls, and the overall number of affected individuals has increased over the years [4, 5].

Based on etiology, ASD can be divided into two categories: syndromic and non-syndromic. The latter may be also referred to as "essential" or "pure". This distinction is crucial to determine appropriate diagnostics and clinical management pathways, including further referrals to genetic specialists.

Non-syndromic ASD does not involve any additional physical or clinical features that would indicate any specific genetic syndrome. In these cases, no unequivocal genetic cause can be identified; thus, the etiology remains largely unknown. The prevailing hypothesis supports a multifactorial model, in which genetic predispositions interact with environmental factors (Figure 1) [11].

Strong evidence for genetic contribution to non-syndromic ASD emerges from twin studies. By the end of the 20th century, a high rate of ASD heritability was observed. Estimated at approximately 90%, ASD is one of the most heritable neurodevelopmental disorders [12]. Moreover, the recurrence risk of ASD in families with one affected child is significantly higher compared to the general population [13].

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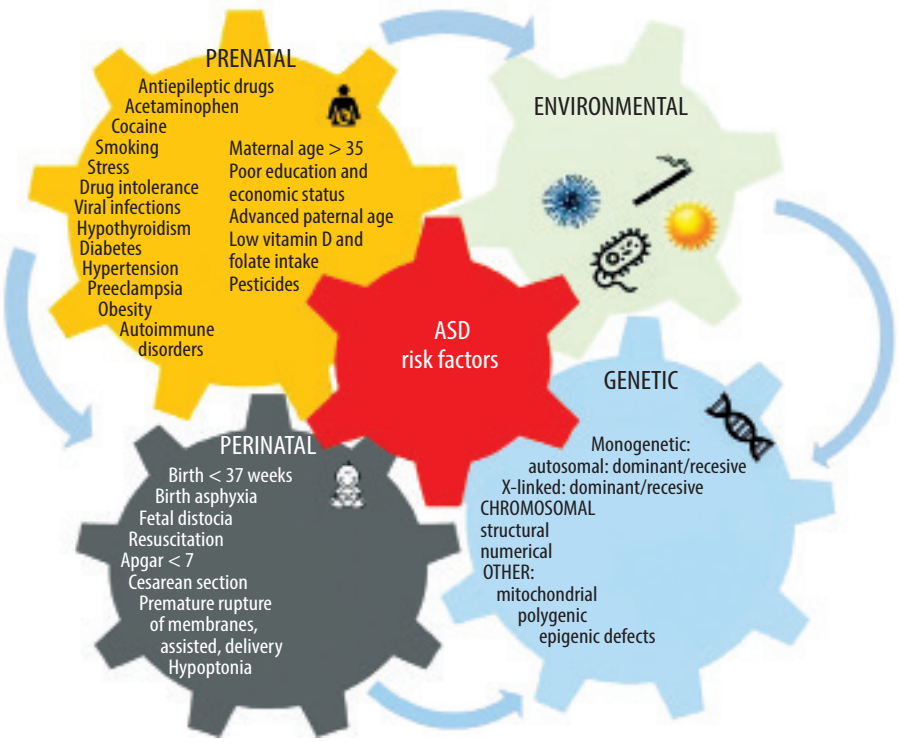

FIGURE 1. Autism spectrum disorders risk factors [6–10]

ASD – autism spectrum disorders
Environmental risk factors include prenatal viral and bacterial infections, mother's sun exposure, and cigarette smoking during pregnancy.

In contrast, in syndromic ASD, autism is one of several features of an identifiable genetic syndrome. These patients typically present with an array of additional symptoms, such as dysmorphic features, congenital malformations, intellectual disability, and specific behavioral phenotypes. These features are not coincidental but arise from a shared genetic etiology. It is estimated that syndromic forms of ASD account for approximately 20% of all autism cases [14].

The main goal of this article is to summarize the genetic causes of ASD. Authors focus on the clinical assessment of ASD patients, from phenotypical examination through pedigree analysis and molecular verification, up to prognosis of development and risk assessment for the proband and other family members.

STEPS OF CLINICAL GENETIC COUNSELING

Figure 2 illustrates steps of genetic counseling pathway, highlighting key objectives at each stage of this diagnostic process.

Genetic counseling is a very important part of the diagnostic process in patients with ASD. It follows a structured approach aimed at evaluating possible genetic etiology and guiding the family through the entire process of testing and result interpretation. A typical appointment usually begins with gathering a detailed medical history, including developmental, neurological, and general health information. The individual's prenatal and perinatal data are also investigated. Then, pedigree analysis is performed in order to identify any potential inheritance patterns and estimate recurrence risk within the patient's family. Based on the collected data, genetic tests are administered after being consulted with patient's parents or guardians. Each test is presented along with its diagnostic value, limitations, and relevance to the individual case. After receiving the results, the geneticist analyses them during a subsequent visit. The test results and their implications should be explained in a sensitive and understandable manner as this information often has a great impact on the patient's life as the diagnosis will determine patient's entire future including further therapeutic procedures or decisions concerning planning offspring [15]. Once a genetic diagnosis is established, recommendations for further management, medical follow-up, and recurrence risk in future pregnancies will be provided. Even if a clear genetic etiology is not identified, a clearly structured counseling process supports families throughout the entire diagnostic pathway.

CLINICAL DIAGNOSIS

The most important part of the clinical evaluation of a patient with ASD is phenotypical assessment. As it provides valuable indicators for establishing the clinical diagnosis of a potential genetic disorder, it should be performed with the highest accuracy.

A physical examination is an essential part of the diagnostic process since many genetic syndromes have a typical "gestalt" (specific phenotype seen at a first glance) that helps to establish the "strasse diagnoze" (e.g.

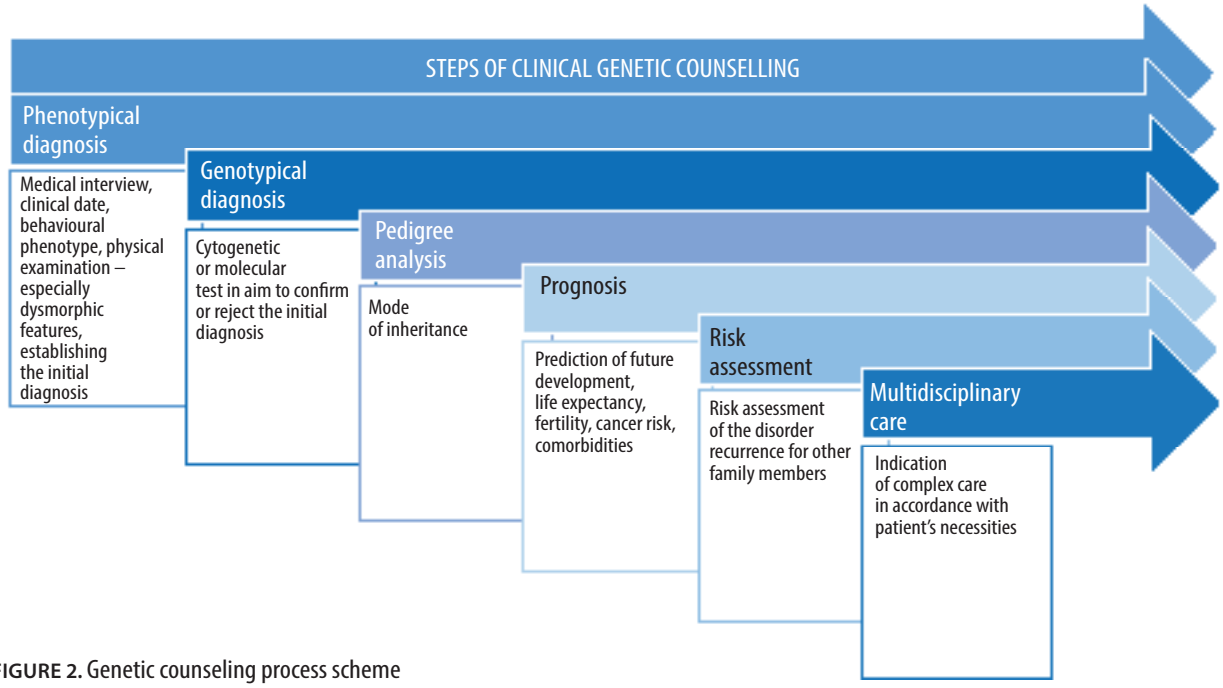

FIGURE 2. Genetic counseling process scheme

Down syndrome, Williams syndrome, *etc.*) [16]. In ASD patients, multiple features suggesting a syndromic ASD may also be encountered. Table 1 lists some of the main signs of syndromic ASD patients. Table 2 provides a brief guideline on when the patient should be referred to a clinical genetics department.

The examination begins with anthropometric measurements. The patient's height, weight, head circumference should be taken and compared with percentile grids appropriate for the patient's sex, age and population of origin. Some studies indicate that early generalized overgrowth (an increase of 2–3 standard deviations from normal height, weight, or head circumference) can be a predictor of later ASD symptoms, indicating inappropriate brain and mental development [17].

Behavioral phenotype can be observed from the first moments of the appointment. The healthcare providers can observe lack of eye contact or interest in social communication, hyperactivity, problems with concentrating (e.g. in Fragile X syndrome) or stereotypical movements (e.g. Rett syndrome). Even the manner of walking may be indicative of an underlying disease (for example: atactic gait suggests the Angelman or Rett syndrome) (Table 3, 4).

MOLECULAR DIAGNOSIS

The etiology of most cases of ASD remains unknown. Genetic tests provide an answer only in some percentage of diagnosed individuals (Table 5).

Based on known genetic ASD markers, many authors have suggested different diagnostic pathways. However, a generally approved scheme is displayed in Figure 3.

TABLE 1. Syndromic autism suggesting features [22]

What suggests syndromic autism?
Facial dysmorphism
Congenital defects
CNS defects
Intellectual disability
Epilepsy

ASD – autism spectrum disorders, CNS – central nervous system

TABLE 2. Criteria for referral to clinical genetics department [7, 35, 83]

Dysmorphic features
Congenital malformations
Intellectual disability or global developmental delay
Family history of ASD or other neurodevelopmental disorders
Neurological abnormalities
Specific syndromic features suggestive of known genetic syndromes

ASD – autism spectrum disorders

PEDIGREE ANALYSIS

Pedigree construction should cover at least three generations of the patient's ancestors. The proband's siblings, parents, and grandparents should be taken into consideration as well as the parents' siblings and their children. Collecting history of pregnancy loss, congenital defects, cases of abnormal psychomotor development, neurodevelopmental and psychiatric disorders, and identified genetic conditions is important since ASD may coexist with many comorbidities, and previous familial ASD cases may have gone undiagnosed.

TABLE 3. Monogenic syndromes associated with – autism spectrum disorders features

Syndrome	Clinical features	Gene	Locus geni	Inheritance	Prevalence	References
Fragile X syndrome	Macrocephaly, long face, broad forehead, large ears, hiperextensible joints, macroorchidism after puberty	*FMR1*	Xq27.3	XLD	1/5 000 males 1/4 000–8 000 females	[18]
Rett syndrome	Normal development in first 6 months of life subsequently developmental regress, secondary microcephaly, stereotypical hand movements, loss of speech, atactic gait, hyperventilation and breath holding	*MeCP2*	Xq28	XLD	1/10 000 females lethal for males	[19]
PTEN – associated macrocephaly syndromes Cowden syndrome Macrocephaly/autism syndrome	Multiple breast, skin, thyroid, oral mucosa, gastrointestinal hamartomas and malignancies, lipomas, macrocephaly Macrocephaly, broad forehead, midface hypoplasia, depressed nasal bridge, short nose, developmental delay	*PTEN*	10q23.31	AD	1/200 000–250 000	[20]
Tuberous sclerosis complex	CNS tumours, cardiac rhabdomyoma, kidney angiomyolipoma, polycstic kidney disease, lung lymphangioleiomiomatosis, skin angiofibroma, shagreen patches, ungual and oral fibromas	*TSC1* *TSC2*	9q34.13 16p13.3	AD	1/6000–10 0000	[21]
Neurofibromatosis type 1	Skin and CNS neurofibromas, plexiform neurofibromas, Lisch nodules, cafe au lait spots, skinfold freckling, optic glioma	*NF1*	1q11.2	AD	1/3000	[22]
Sotos syndrome	Overgrowth, macrocephaly, advanced bone age, round face in infancy but longer in later age, prominent forehead, hypertelorism, prominent jaw, large ears	*NSD1*	5q35.3	AD	1/14 000	[23]
Cornelia de Lange syndrome	Short stature, low weight, microcephaly, synophrys, long eyelashes, short nose, long philtrum, thin lips, small hands, oligodactyly, hirsutism	*NIPBL* *SMC3* *SMC1L1* *HDAC8* *RAD21*	5p13.2 10q25.2 Xp11.22 Xq13.1 8q24.11	AD AD XLD XLD AD	1/10 000	[24, 25]
Noonan syndrome	Short stature, heart defect, short webbed neck, low posterior hairline, chest defect, hypertelorism, downslanting palpebral fissures, low-set ears	*PTPN11* and many others	12q24.13	AD	1/1000–2500	[26]
Timothy syndrome	Developmental delay, mental retardation, small teeth, low set ears, syndactyly, cardiac defects, cardiac arrythmias, recurrent infections	*CACNA1C* sometimes somatic mosaicism	12p13.33	AD	Less than 100 worldwide	[27]

TABLE 3. Cont.

Syndrome	Clinical features	Gene	Locus geni	Inheritance	Prevalence	References
Smith-Lemli-Opitz syndrome	Short stature, microcephaly, ptosis, short nasal root, anteverted nares, micrognathia, syndactyly of 2nd and 3rd toes, cardiac, genitourinary and gastrointestinal defects	*DHCR7*	11q13.4	AR	1/10 000–50 000	[28]
Duchenne muscular dystrophy	First symptoms about 4 year of life associated with progressive muscle weakness, inability to walk about the age of 10, calf muscle pseudohypertrophy, cardiomyopathy, respiratory complications	*DMD*	Xp21.2-21.1	XLR	1/3500 males	[29]
Myotonic dystrophy type 1	Myotonia, facial weakness, ptosis, ECG abnormalities, sleep disturbances	*DMPK*	19q13.32	AD	1/8000	[30]
Cohen syndrome	Hypotonia, developmental delay, microcephaly, downslanting palpebral fissures, thick eyebrows and hair, visual impairment, leukopenia obesity in adulthood	*VPS13B*	8q22.2	AR	Unknown	[31]

AD – autosomal dominant, AR – autosomal recessive, CNS – central nervous system, XLD – X-linked dominant, XLR – X-linked recessive

PROGNOSIS

Malignancies

It is widely known that some genetic syndromes are associated with frequent malignancy and some of them are characterized by autistic features (Table 6) [48]. The awareness of the aforementioned correlation gives an opportunity for regular focused screening, early cancer diagnosis and therefore early and effective oncologic treatment (Table 7).

Development

Human development is closely related to language skills. The speech progress varies between patients and impacts development prediction. Presumably, language acquisition decelerates after the age of 11, so early speech therapy is crucial for higher efficacy [49]. It is estimated that 55–95% of children with autism spectrum disorder will achieve speech ability, almost at the same level as their unaffected peers, if verbality and intelligence level are within a normal range [49, 50]. Communication and language development are extremely important for adult patient functioning. Adults with autism present various levels of language skills which impact their vocational independence and friendships making. The higher IQ, richer vocabulary, more fluent grammar use and utterance complexity, the higher chances for better professional opportunities and more satisfying social life [51]. Fragile X syndrome is always associated with a certain level of speech development delay. Echolalia, difficulty with understanding non-literal language, and articulation problems are very common. Early and intensive speech therapy focusing on both expressive and receptive language is recommended. Therapeutic strategies include structured language intervention, social communication training, and the use of augmentative and alternative communication (AAC) devices when verbal output is limited [55]. In Rett syndrome, after a period of seemingly normal language and speech development, a regression of already acquired skills is observed. Patients usually do not achieve a level of speech that is sufficient for normal everyday communication. The use of AAC such as communication boards or eye-gaze technology is implemented to enable contact with the patient [56]. Similarly, in Angelman syndrome (AS), minimal to absent verbal speech is typical, so alternative communication is crucial. A newly published article suggests that patients with AS do not use verbal communication but are able to understand speech, and eye tracking methods can be used in their speech therapy [57]. In Williams syndrome, patients usually preserve expressive speech and present rich vocabulary. At the same time, pragmatic language use and social deficits are observed.

TABLE 4. Chromosomal syndromes associated with autism spectrum disorders features

Syndrome	Clinical features	Chromosomal abnormality	Prevalence	References
Down syndrome	Infantile hypotonia, nuchal fold, flat nasal bridge, upslanting palpebral fissures, epicanthal folds, single palmar crease, short stature, congenital heart defects, hearing and vision problems, hematologic disorders, hypothyroidism	Trisomy 21 Partial trisomy 21 Mosaic trisomy 21	1/800	[32]
Turner syndrome	Short stature, webbed neck, low set ears, low posterior hairline, ovarian failure, infertility, middle ear infections, hearing loss, heart defects, hypothyroidism	Monosomy X Partial monosomy X Mosaic monosomy X	1/2000 females	[33]
Klinefelter syndrome	Tall stature, gynecomastia, low gonadotropin and testosterone levels, small testes, infertility, increased fat tissue, delayed speech, learning difficulties	Extra X chromosome in male	1/660 men	[34]
XYY syndrome	Tall stature, macrocephaly, hypertelorism, hypotonia, macroorchidism, tremor, delayed development	Extra Y chromosome in male	1/1000	[35]
Di George syndrome	Delayed development, learning difficulties, seizures, micrognathia, palatal abnormalities, hypertelorism, eyelid hooding, cardiac defects, parathyroid and thymic hypoplasia/aplasia, hypothyroidism, frequent infections	22q11.2 deletion	1/4000	[36]

TABLE 5. Autism spectrum disorders genetics background detectability in different test [23, 37–41]

Test	Detectability (%)	
	Syndromic	Non-syndromic
Karyotype	2–3	< 1
CMA	Up to 20	13
FMR1	0.46–5	–
MeCP2	4 of girls	–
PTEN	5 of patients with macrocephaly	–
WES	25–40	10–20
WES + CMA	Up to 50	Up to 30

CMA – chromosomal microarray, FMR1 – cytozine-guanine-guanine trinucleotide repeats test in FMR1 gene, MeCP2 – MECP2 gene sequence analysis, WES – whole exome sequencing

In such cases, therapy focuses on understanding abstract language and reducing stereotyped or socially inappropriate language behaviours [58].

Comorbidities

Coexisting disorders can veil ASD and cause the delay of the proper clinical diagnosis. Multiple kinds of neurologic, psychiatric, and gastrointestinal problems may be observed along with autism. Among numerous symptoms about which patients complain are also sleep disturbances. The most frequent signs and symptoms present in common genetic syndromes associated with autism are presented in Tables 3, 4. Awareness of the most common symptoms that accompany autistic features in known genetic syndromes allows the early screening and vigilance for frequent comorbidities and therefore early, tailor-made therapy and developmental support for the patient. For instance, epilepsy is observed in over 80% of patients with AS [59]. In the treatment of epilepsy in AS, levetiracetam is highly effective. Simultaneously, in drug-resistant seizures ketogenic and low glycemic index diets are used [60]. Until recently, Duchenne muscular dystrophy used to be treated only in a symptomatic way (physical treatment, corticosteroids, cardiac and respiratory support) [61]. In June 2023, the U.S. Food and Drug Administration approved delandistrogene moxeparvovec (marketed as Elevidys) for children aged 4 and 5 with a confirmed mutation in the *DMD* gene. Later, the indications were expanded to ambulatory individuals aged 4 and older [62]. Gene therapy for Duchenne muscular dystrophy represents a significant breakthrough in the treatment of genetic disorders, offering hope for the development of drugs for other genetic syndromes. Short stature is a key characteristic of Noonan syndrome (NS). Several studies have demonstrated the efficacy of growth hormone (GH) treatment in improving height in children with NS. The timing of GH implementation plays a crucial role and gives the best treatment outcomes when administered before puberty [63, 64].

Life expectancy

Nowadays, it is believed that patients with autistic symptoms without comorbid disorders have life expectancy similar to normal population. However, comorbidities common in autism and congenital defects such

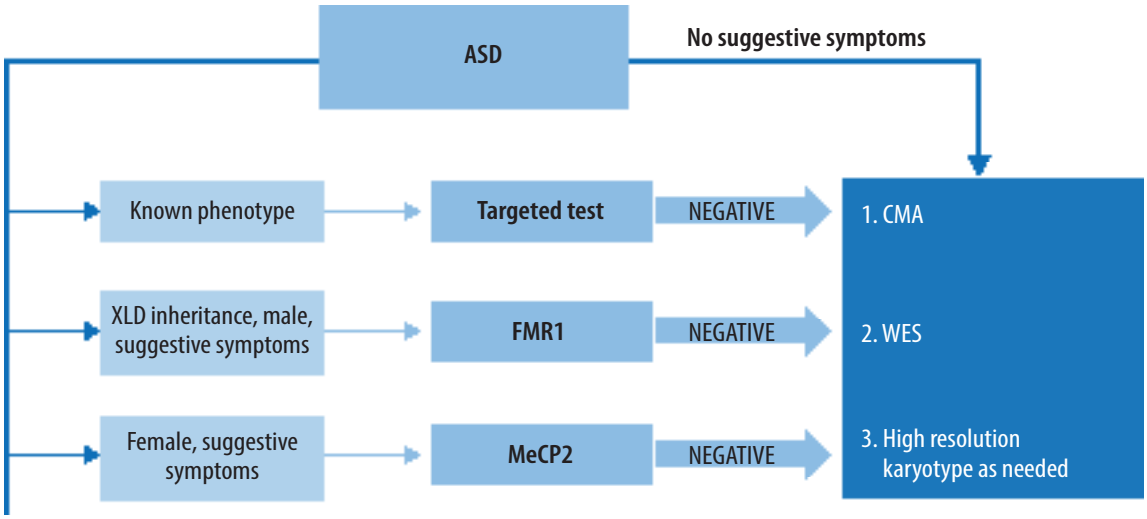

FIGURE 3. Suggested diagnostic pathway in autism spectrum disorders [38, 42]

ASD – autism spectrum disorders, CMA – chromosomal microarray, WES – whole-exome sequencing, XLD – X-linked dominant

as heart or respiratory abnormalities may cause an early demise [71, 72]. Shorter life is expected in patients with Down syndrome due to heart defects and premature aging, including early-onset Alzheimer's disease [73]. Patients suffering from Rett syndrome experience recurrent infections and breathing problems, which, according to recent observations, may be reconciled by proper medical treatment and rehabilitation [74]. Other examples of life shortening factors are epilepsy and trauma in Angelman syndrome [75], or the complications and diseases resulting from obesity in Prader-Willi syndrome [76].

Fertility

There is scarce data concerning fertility in patients affected by autism spectrum disorders. The researchers focus on maternal and paternal fertility disturbances in terms of ASD hereditability rather than individuals' fertility potential itself. Nevertheless, some autism connected genetic syndromes, such as Klinefelter or Turner syndromes, may affect patients' fertility [33, 34]. Hypogonadism, both central and gonadal, is common in Prader-Willi syndrome and leads to delayed puberty, underdeveloped genitalia, and reduced fertility in both males and females [54]. Although females with Rett syndrome typically reach normal puberty, secondary hypogonadism can occur, resulting in irregular menstruation and decreased fertility [19]. In many genetic syndromes associated with autism, patients are unable to live independently and usually have no chance of conceiving [77]. Nevertheless, infertility treatment should be offered to people who want to start the family [78]. Infertility can be treated in different approaches depending on the underlying cause. Hormonal therapies, such as gonadotropins or estrogen/progesterone supplementation, can help induce puberty or stimulate gametogenesis in individuals with Turner syndrome or Prader-Willi syndrome [79, 80]. In males with

TABLE 6. Autism spectrum disorders comorbidities [43–47]

Parameters	Symptoms
Neurologic	Epilepsy Macrocephaly Hydrocephalus Cerebral palsy Headache Migraine CNS defects
Gastrointestinal	Constipation Diarrhea Gastroesophageal reflux Nausea Vommiting Food intolerance Poor growth Pica Abdominal pain Food selectivity Food refusal Celiac disease
Psychiatric	Anxiety Mood disorders ADHD Depression Obsessive-compulsive disorder Psychosis Intellectual disability Gender dysphoria Disruptive disorders Schizophrenia
Other	Sleep disturbances Refractory defects Strabismus Autoimmune diseases Obesity Incontinence Self-inflicted oral soft tissue injuries Atopy Dental caries

TABLE 7. Syndromic autism and correlated malignancies [21, 22, 52–54, 65–70]

Syndrome	Malignancies
Cowden	Breast Thyroid Kidneys Uterus
Tuberous sclerosis complex	Renal cell carcinoma Cardiac rhabdomyoma Ependymoma Giant cell astrocytoma Chordoma (children!)
Neurofibromatosis type 1	Optic glioma Gliomas Pheochromocytoma Leukaemia Gastrointestinal stromal tumour (gist) Malignant peripheral nerve shift tumour Rhabdomyosarcoma Breast
Sotos	Wilms tumour Hepatocarcinoma Lung Leukaemia Neuroblastoma
Noonan	Malinant schwannoma Neuroblastoma Pheochromocytoma Leukaemia Rhabdomyosarcoma
Down	Leukaemia Testicles
Turner	Melanoma CNS Colon
Klinefelter	Non-Hodgkin lymphoma Leukaemia Hematological
Prader-Willi syndrome	Melanoma Skin Leukaemia

CNS – central nervous system

Klinefelter syndrome, intracytoplasmic sperm injection enables affected men to become biological fathers. Sperm, retrieved directly from the testes, is used to fertilize an egg [81]. Assisted reproductive technologies, such as *in vitro* fertilization can also help bypass structural or functional reproductive impairments [82].

Risk assessment

After analysis of genetic testing results and pedigree analysis, a genetic counselor is able to estimate the risk of ASD recurrence in a patient's family members. The risk assessment may be easy to predict by means of a monogenic disorder diagnosis. In autosomal dominant disorders, the recurrence risk is 50% for each child. In autosomal recessive disorders, the risk is 25% with a 50% risk of being a carrier. In the case of X-linked recessive inheritance the risk differs depending on the parent gender. When the mother is a carrier, the risk of children developing the disorder is 50% for male offspring, while the female descendants may be gene carriers with 50% probability. When the father is affected, there is 100% certainty that his daughter will be a gene carrier. His male offspring, on the other hand, will never be affected. In X-linked dominant inheritance mode, the mother will pass a gene statistically to half of her offspring regardless of their sex, while the father's genes will be passed onto all his daughters. In X-linked inheritance, there is no transmission from father to son [83]. In addition, genetic advice should include phenomena and exceptions like *de novo* mutation, anticipation, penetrance and consanguinity [84].

Non-syndromic autism is more complicated to diagnose as the genetic factor is usually not recognizable. As a result, the risk estimation cannot be done with the same level of accuracy as in the case of mendelian inheritance patterns. In non-syndromic ASD no specific pathogenic variant is identified and the underlying mechanism is typically multifactorial. This means an interplay of both genetic predisposition, and environmental factors. Although no single cause has been found, empirical data from recent studies may help the estimation of the recurrence risk for counseling purposes. According to data from the Baby Siblings Research Consortium, approximately 18.7% of younger siblings of children with ASD receive an autism diagnosis by the age of three [85]. Recent follow-up research from 2024 suggests that this number increases to nearly 37% when assessments are extended into the school-age period [86]. A large-scale population study also reported a relative recurrence risk of 8.4 for full siblings, which further supports the strong familial aggregation in ASD even in the absence of a defined genetic diagnosis [87]. These findings emphasize that while genetic testing may not always reveal a specific etiology, families should be aware that the recurrence risk for siblings is significantly higher than that of the general population (about 1%). Including this empirical data enhances the clarity and clinical utility of genetic counseling, particularly when supporting decision-making in families with one affected child.

Multidisciplinary care

The high etiologic and clinical heterogeneity of patients with ASD requires a multidisciplinary and individualized approach. Extreme variability of associated disorders and symptoms, different levels of social functioning, speech development and learning difficulties preclude a universal care proposal. Each patient copes with several miscellaneous problems, demanding an independent and tailor-made solution.

It is obvious that parents and healthcare providers should react to a child's symptoms individually. Because of multiple comorbidities, mental and neurological disturbances may occur. Those patients should be offered consultation with a psychiatrist and neurologist. Consultations with gastroenterologist, endocrinologist, dentist, allergist, psychologist and many others should be provided accordingly.

Social deficits and speech development abnormalities are core ASD symptoms. Speech therapy should be performed by professionals familiar with ASD. Sensory integration disturbances are also observed and require adequate intervention. School-related social problems, hyperactivity, and learning difficulties require specific interventions and significant effort from parents and teachers, who face numerous challenges at each educational stage.

There are several social support websites and organizations dedicated to children with autism and their families. These provide resources, peer support, advocacy, and community connections (e.g. Autism Speaks – global organization offering support, research updates, and advocacy; MyAutismTeam – a social network for parents of children with autism to connect and share advice; in Poland – Krajowe Towarzystwo Autyzmu or Porozumienie Autyzm – Polska; on Facebook search for groups like "Autism Parents Support" or "Autism Support Network").

FOLLOW-UP

Genetic counseling is crucial for follow-up care of children with ASD and their families. Identification of pathogenic variants can inform recurrence risk assessments, guide reproductive decisions and enable early ASD diagnosis or monitoring of siblings at risk [39, 88]. Genetic conditions associated with ASD are frequently related to accompanying comorbidities. The awareness of their occurrence opens the door to tailored surveillance and interventions [89]. The prognosis in ASD is highly variable and depends on the genetic diagnosis, but revealing an etiology of a patient's disorder can refine prognostic predictions and help clinicians develop individualized care plans. Integrating genetic results into clinical practice offers a chance for comprehensive care, supports families in decision-making, and may improve long-term outcomes through targeted therapies and monitoring [35].

CONCLUSIONS

The aim of this review was to help general practitioners, paediatricians, and other non-geneticists healthcare providers become familiar with the role of genetics in all stages of management of patients diagnosed with ASD. This article may be a handy guide of how to incoprorate genetic insights into everyday practice. Autism spectrum disorders is a highly variable group of neurodevelopmental disorders and requires an individualized approach. Medical providers should be able to recognize children at risk of autistic symptoms and guide their further care.

DISCLOSURES

1. Institutional review board statement: Not applicable.
2. Assistance with the article: None.
3. Financial support and sponsorship: None.
4. Conflicts of interest: None.

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