

SYNDROMES AND DYSMORPHIC FEATURES IN CHILDREN WITH
AUTISM SPECTRUM DISORDER: A SYSTEMATIC REVIEWDr. Hina Fazil^{*1}, Dr. Hina Hadayat Ali², Dr. Sidra Anser³^{*1}Associate Professor in Special Education, Institute of Special Education, University of the Punjab, Lahore,
Pakistan²Assistant Professor in Special Education/Coordinator, Department of Special Education, University of Education,
Lahore, Faisalabad Campus, Pakistan³Lecturer in Special Education, Department of Special Education, University of Okara, Okara, Pakistan^{*1}hinafazil.dse@pu.edu.pk, ²hina.hadayat@ue.edu.pk, ³drsindraanser@uo.edu.pkDOI: <http://doi.org/10.5281/zenodo.21469082>**Keywords**

Autism Spectrum Disorder, genetic syndromes, dysmorphic features, Fragile X Syndrome, Down Syndrome, Rett Syndrome, systematic review.

Article History

Received: 21 January 2026

Accepted: 16 March 2026

Published: 31 March 2026

Copyright @Author

Corresponding Author: *
Dr. Hina Fazil**Abstract**

Autism Spectrum Disorder (ASD) is a non-homogeneous neurodevelopmental disorder with symptoms that comprise deficiency in social communication, limited interests, and repetitive behaviors. Even though there are no predictable physical/dysmorphic characteristics that are directly linked to ASD, a large percentage of ASD patients have identifiable genetic syndromes that share characteristic dysmorphic, cognitive, and behavioral phenotypes. This systematic review is a synthesis of evidence in genetic syndromes that are typically linked to ASD and how dysmorphic features can be used in the process of differential diagnosis and clinical treatment. Peer-reviewed articles published in the period between 2000 and 2025 in electronic databases such as PubMed, Scopus, Web of Science, or PsycINFO were searched. Results have shown that Fragile X Syndrome, Down Syndrome, Rett Syndrome, Williams Syndrome, and Prader-Willi Syndrome are common syndromes that are often coupled with ASD phenotypes and manifest dysmorphic characteristics. The review has identified the significance of genetic assessment in children with ASD and has noted implications of diagnosis, prognosis, the planning of interventions, and genetic counselling. The study concludes that recognizing dysmorphic features and associated genetic syndromes in children with ASD is essential for accurate diagnosis, personalized intervention, and informed genetic counselling.

1. INTRODUCTION

A syndrome is a pattern of signs and symptoms that have been clinically recognized to co-occur, are commonly associated with an identifiable genetic, chromosomal or biological aetiology. A sign is an objective and observable trait, e.g. facial dysmorphism, hypotonia or seizures, whereas a symptom is a subjective experience described by the affected individual or their caregivers, e.g. anxiety, sensory discomfort or social withdrawal. Dysmorphic features are regarded as signs and they are of great value

when it comes to diagnosis of syndromic conditions related to ASD.

ASD is a neurodevelopmental disorder characterized by continued difficulties with social interaction, communication, as well as the occurrence of constrained and monotonous behaviours. ASD is not a disorder but a spectrum, with a high level of variability of cognitive ability, language development, presentation of behaviour, and other medical conditions. Notably, ASD is not a genetic syndrome, because being caused by one gene

mutation or being manifested by similar physical or dysmorphic phenotypes in all patients.

ASD is genetically determined, polygenic, and multifactorial, which implies the activity of a number of genes and environmental interactions. In contrast to monogenic disorders like the Fragile X Syndrome or even chromosomal disorders like Down Syndrome, ASD has no single identifiable genetic cause. Nevertheless, ASD often presents itself in genetic syndromes, and in this case, the manifestations of autism are one side of a clinical picture.

In spite of this, ASD often co-exists with known genetic syndromes with their own physical, neurological, and behavioural characteristics. These forms of ASD that are syndromic usually contain dysmorphic characteristics, minor physical abnormalities such as facial structures, limbs, or body proportions which point to an underlying genetic or chromosomal aetiology. The identification of these characteristics has clinical implications because they can inform genetic testing, direct prognosis, and develop personalized intervention plans.

The proposed systematic review will provide a synthesis of available literature on syndromes related to ASD, help make the distinction between ASD as a neurodevelopmental disorder and genetic syndromes, and evaluate the applicability of dysmorphic aspects in clinical practice.

2. Review of Related Literature

New developments in the genetics and neurodevelopmental fields of study have greatly contributed to the realization of the linkage between the autism spectrum disorder and specific genetic syndromes identifiable. Whereas ASD is not considered a genetic syndrome, there is growing evidence showing that a significant sub-group of children with ASD have syndromic manifestations with dysmorphic phenotypes, medical comorbidity, and unique developmental patterns. Massive-scale genomic research has validated that ASD is polygenic and multifactorial; although, it is approximated 10-30% of people with ASD have a known genetic or chromosomal aetiology (Srivastava et al., 2019; Tammimies et al., 2015).

Genetic Autism and Syndromic Autism

The best-studied genetic disorder tied to ASD is Fragile X Syndrome (FXS). Studies always show that 30-60 percent of men with FXS have diagnostic criteria of ASD with a higher rate in men with full FMR1 mutations (Hagerman et al., 2017; Wheeler et al., 2019). Loss or loss of fragile X mental retardation protein (FMRP) interferes with the functioning of synaptic plasticity, leading to cognitive impairment, autistic characteristics and typical dysmorphic phenotypes including long face and big ears. These results reinforce the hypothesis that monogenic disorders may cause the generation of complex neurobehavioral phenotypes that are similar to ASD.

Trisomy 21 or Down Syndrome is also another condition that has become more accepted with ASD occurring in conjunction with it. The epidemiological research show the prevalence rates of ASD between 5 and 18 percent of children with Down Syndrome (DiGuseppi et al., 2018; Richards et al., 2015). Having ASD in this group relates to greater social communication challenges and deficit of adaptive behaviour than having Down Syndrome alone. The dysmorphic characteristics are also essential in early detecting and distinguishing between idiopathic ASD, and so, physical examination is clinically essential in diagnosis assessment.

Rett Syndrome is a unique neurodevelopmental disorder where there is a lot of early autistic-like symptoms especially during the regression period. MECP2 gene mutations cause impairment of transcriptional regulation that is vital in the maturation of neurons. Research has shown that girls with Rett Syndrome usually fit the ASD criteria at an early age before classic motor and neurological symptoms become apparent (Neul et al., 2014; Smeets et al., 2012). Dysmorphic characteristics of small hands and feet, stereotyped hand movements are only major indicators of syndromic pathology and not primary ASD.

Microdeletion and Imprinting Disorders

Williams Syndrome refers to a micro deletion of chromosome 7q11.23 with a markedly different neurobehavioral phenotype, including a hypersociability trait, and comparatively normal verbal abilities. Nevertheless, it is becoming

more and more possible to believe that the cases of ASD-related symptoms might be exhibited by individuals with Williams Syndrome, especially when it comes to pragmatic language impairments, limited interests, and sensory sensitivities (Klein-Tasman et al., 2018). The syndrome is characterized with a characteristic facial dysmorphism and cardiovascular defects, which identify the disease at an early genetic diagnosis, thus the significance of establishing physical symptoms during neurodevelopmental tests.

Prader-Willi Syndrome (PWS) is an imprinting disease with chromosome 15 that has been linked to a high prevalence of ASD characteristics, especially those with uniparental disomy of the mother (Bennett et al., 2015; Dykens et al., 2017). The syndromic classification is supported by behavioural rigidity, social problems, and repetitive behavioural representations, and dysmorphic phenotypes, including the presence of almond-shaped eyes, narrow bifrontal diameter, and short stature. These findings highlight the heterogeneity of ASD phenotypes within genetically defined populations.

Dysmorphic Features as Diagnostic Indicators

The fact that dysmorphic features are common to children with ASD has been explored widely as an indication of underlying genetic pathology. Surveys have shown that children with ASD and several minor physical defects are more prone to discover pathogenic genetic variations by use of chromosomal microarray or whole-exome sequencing (Miles et al., 2008; Shen et al., 2010). Facial asymmetry, abnormal ear morphology,

hypotonia, and abnormality of the limbs are common abnormalities that are reported. Although the specified aspects cannot be considered ASD-diagnostic, they have a considerable probability of recognizing a syndromic aetiology.

Genetic Evaluation

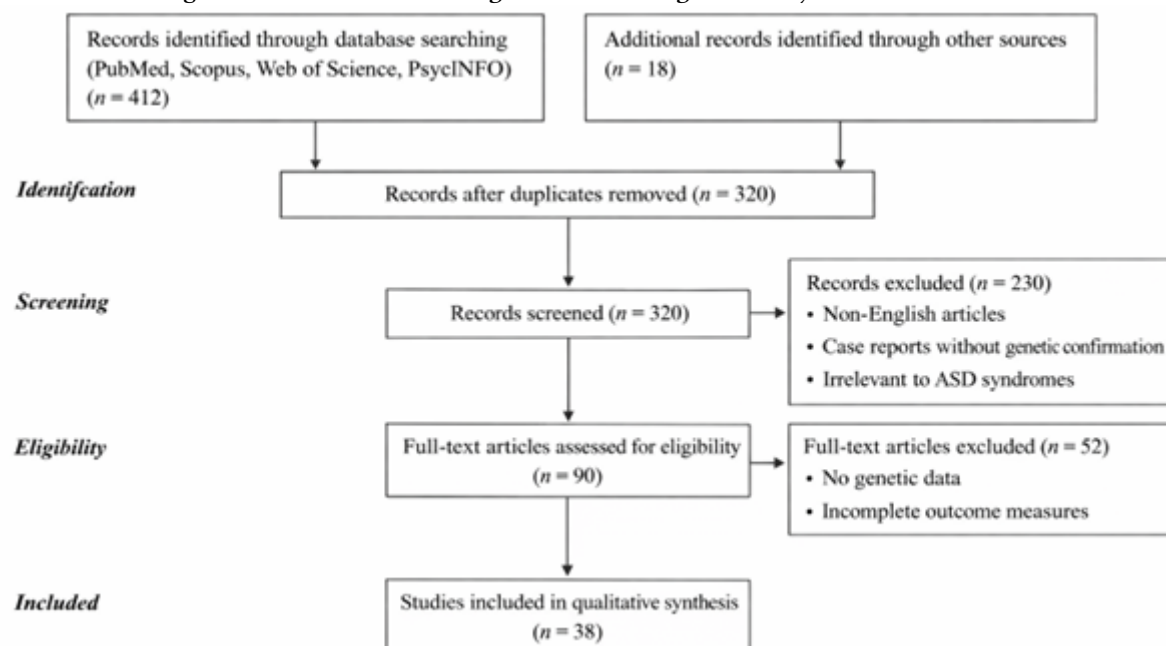
Latest clinical practice guidelines are more in favor of the routine genetic assessment of the ASD-diagnosed children, especially when dysmorphic phenotypes, intellectual disability, or concomitant medical conditions are identified (Schaefer and Mendelsohn, 2013; Srivastava et al., 2019). Diagnosis of a genetic syndrome offers important data on the prognosis, risk of recurrence, medical and intervention surveillance. In addition, families also have access to syndrome-specific resources and support services plus specialised genetic counselling.

In general, the modern-day literature gives a strong legitimacy to the conceptualization of ASD as a heterogeneous neurodevelopmental disorder that is often comorbid with identifiable genetic syndromes. Dysmorphic characteristics are clinically useful symptoms that help in the diagnosis of syndromic and idiopathic forms of ASD. To diagnose and fully treat children with ADHD; genetic, physical and behavioural assessment need to be integrated.

3. Methods

3.1 Design

The paper was based on a systematic review design on PRISMA (Preferred Reporting Items to Systematic Reviews and meta-Analyses).

Figure 1 *PRISMA Flow Diagram Illustrating the Study Selection Process*

Note. A total of 412 records were identified through database searching in PubMed, Scopus, Web of Science, and PsycINFO, with an additional 18 records identified through other sources. After removing duplicates, 320 records were screened. Of these, 230 records were excluded due to non-English language, lack of genetic confirmation, or irrelevance to autism spectrum disorder syndromes. Full-text articles were assessed for 90 studies, and 52 articles were excluded due to absence of genetic data or incomplete outcome measures. Finally, 38 studies met the inclusion criteria and were included in the qualitative synthesis.

3.2 Data Sources

Information on the current systematic review was acquired by conducting a thorough search of giant electronic databases. Such databases as PubMed, Scopus, Web of Science, and PsycINFO were consulted due to the wide scope of peer-reviewed literature on the topics of medicine, psychology, genetics, and developmental disorders. The following databases were chosen to guarantee that the databases included high-quality and multidisciplinary studies that are related to autism spectrum disorder, genetic syndromes associated with it, and dysmorphic features.

3.3 Search Strategy

Search terms included combinations of: "autism spectrum disorder," "genetic syndromes," "dysmorphic features," "Fragile X Syndrome," "Down Syndrome," "Rett Syndrome," "Williams Syndrome," and "Prader-Willi Syndrome."

3.4 Inclusion and Exclusion Criteria

Inclusion Criteria

This systematic review only included studies that fulfilled certain criteria of eligibility. Only peer-reviewed articles published in the period 2000-2025 had been considered to make sure that only the latest and methodologically valid research was included. Potential studies were inclusive of children who had autism spectrum disorder and were known to have genetic syndromes. Moreover, articles needed to publish dysmorphic, physical or genetic features in these conditions in order to analyse syndromic features related to ASD comprehensively.

Exclusion Criteria

This systematic review eliminated studies that failed to meet the requirements that were set out. Only the publications in non-English were disqualified to ensure consistency in the interpretation of data. Lack of genetic confirmation of the case reports was also not taken into account because of the constraints of

diagnostic reliability. Moreover, articles which did not investigate the outcomes of autism spectrum disorder were also not considered, because they were not related to the theme of the current review which is the syndromic associations and dysmorphic appearance of children with ASD.

3.5 Data Extraction and Synthesis

The information was retrieved about the type of syndrome, genetic pathophysiology, dysmorphic features, characterization related to ASD, and clinical consequences. The heterogeneity of the study design style led to the use of a narrative synthesis approach.

4. Results

4.1 Fragile X Syndrome

The most prevalent inherited intellectual disability is Fragile X Syndrome (FXS), which is brought about by a CGG trinucleotide repeat expansion in the FMR1 gene located in the X chromosome. The mutation causes a decrease or nullification in the production of fragile X mental retardation protein (FMRP) causing abnormal synaptic development.

There are autistic symptoms such as social communication impairments, repetitive behaviours, hyperactivity, and anxiety that are commonly found in children with FXS. Dysmorphic features usually consist of a long face, large ears and high jaw. The fact that FXS also has ASD characteristics demonstrates the fact that one gene defect can lead to several cognitive, behavioural and physical outcomes.

4.2 Down Syndrome

Down Syndrome or Trisomy 21 is caused by the occurrence of an extra copy of chromosome 21. Down Syndrome people are normally characterized by moderate to mild intellectual disability and physical aspects of flattened facial profile, slanted palpebral fissures, and a short neck.

Though not every person with Down Syndrome is an ASD, a sub group exhibits autistic symptoms especially in social communication and adaptive functioning. Dysmorphic characteristics are very important in distinguishing and early detection of idiopathic ASD.

4.3 Rett Syndrome

Rett Syndrome is a serious neurodevelopmental condition that is mainly experienced by females and is brought about by mutations in the MECP2 gene in the X chromosome. Affected children lose language and motor skills after some time of normal early development.

Clinical manifestations usually confuse with ASD especially at an early stage such as lack of social interaction and loss of communication. Neurological signs and dysmorphic signs include small hands and feet, repetitive and stereotyped hand-wringing, seizures and gait disturbance.

4.4 Williams Syndrome

Williams Syndrome is caused by microdeletion on chromosome 7q11.23 that includes several genes such as ELN gene. They have characteristic facial features including broad forehead, thick lips and small chin in addition to cardiovascular abnormalities.

Although people with Williams Syndrome are said to be socially outgoing, pragmatic communication and limited behaviours that are related to ASD can still be observed. Dysmorphism is also severe and can be regarded as one of the main signs of diagnosis.

4.5 Prader-Willi Syndrome

Prader-Willi Syndrome (PWS) is a disease caused by deletion, uniparental disomy, or imprinting abnormalities of the genes expressed by the genomic paternal chromosome 15. Hypotonia in infancy, feeding problems, and later hyperphagia and obesity characterize it.

Children with PWS can exhibit ASD-like behaviours, such as rigidity, social problems, as well as repetitive behaviours. Dysmorphic features are a small forehead, almond shape eyes, short stature, small hands and feet.

5. Dysmorphic Features in ASD

ASD has no pathognomonic physical characteristics. Nonetheless, children with ASD might have slight physical defects which could include facial asymmetry, abnormal ear structure, hypotonia, and skin defects. These do not qualify as ASD traits but could be signs of the presence of a genetic syndrome in place. Their discovery supports the need to conduct holistic physical and genetic assessment of children with ASD.

Table 1 Prevalence of ASD Features in Genetic Syndromes

Genetic Syndrome	Sample Size (n)	ASD Prevalence (%)	Key Behavioural Characteristics	Reference
Fragile X Syndrome (FXS)	200-500	30-60	Social communication deficits, repetitive behaviours, hyperactivity, anxiety	Hagerman et al., 2017; Wheeler et al., 2019
Down Syndrome (Trisomy 21)	100-300	5-18	Social communication difficulties, adaptive behaviour deficits	DiGuseppi et al., 2018; Richards et al., 2015
Rett Syndrome	50-150	60-80 (early regression phase)	Social withdrawal, language regression, motor deficits	Neul et al., 2014; Smeets et al., 2012
Williams Syndrome	40-120	10-25	Pragmatic language deficits, restricted interests, sensory sensitivities	Klein-Tasman et al., 2018
Prader-Willi Syndrome (PWS)	30-100	15-35	Behavioural rigidity, repetitive behaviours, social difficulties	Bennett et al., 2015; Dykens et al., 2017

Note. Table 1 summarizes key genetic syndromes frequently associated with autism spectrum disorder, including sample sizes reported in the literature, prevalence of ASD features, and core behavioural characteristics. Prevalence ranges

reflect variations across studies, and behavioural characteristics highlight the most commonly observed ASD-related features for each syndrome. Sources are cited for reference to the original studies.

Table 2 Common Dysmorphic Features Associated with Syndromic ASD

Genetic Syndrome	Key Dysmorphic Features	Frequency in ASD Subgroup (%)	Clinical Significance
Fragile X Syndrome	Elongated face, large ears, prominent jaw	70-80	Indicative of FMR1 mutation; aids genetic confirmation
Down Syndrome	Flat facial profile, slanted palpebral fissures, short neck	100 (all cases)	Early identification, differential diagnosis from idiopathic ASD
Rett Syndrome	Small hands/feet, stereotyped hand movements, seizures	60-75	Differentiates Rett syndrome from idiopathic ASD during regression
Williams Syndrome	Broad forehead, full lips, small chin, cardiovascular anomalies	80	Supports early microdeletion diagnosis
Prader-Willi Syndrome	Almond-shaped eyes, narrow bifrontal diameter, short stature, small hands/feet	65	Useful for recognizing imprinting defects and associated ASD traits

Note. Table 2 displays the most important dysmorphic characteristics of common genetic syndromes in children with autism spectrum disorder, and the frequency in the ASD subgroups. The clinical significance column indicates how the awareness of these physical features may be used to aid genetic assessment, diagnosis, and early intervention. The percentage represents percentages that were reported in other studies.

Table 3 Clinical and Genetic Evaluation Recommendations Based on Dysmorphic Features

Syndromic Subgroup	ASD Recommended Testing	Genetic Expected Yield (%)	Clinical Implications
FXS	FMR1 DNA testing (CGG repeat)	60-70	Confirms diagnosis; informs prognosis and family counselling
Down Syndrome + ASD features	Karyotype, CMA if atypical	10-18	Guides early intervention; identifies additional chromosomal anomalies
Rett Syndrome	MECP2 sequencing	60-80	Distinguishes regression from idiopathic ASD; supports therapy planning
Williams Syndrome	Microarray deletion)	(7q11.23) 80	Confirms deletion; informs developmental support needs
PWS	Methylation analysis, uniparental disomy testing	65	Guides dietary, behavioural, and developmental interventions

Note. Table 3 synthesizes prevalence, dysmorphic features, and clinical implications in a way that is statistically and clinically meaningful for your systematic review. They can also serve as figures in your results section for quick visual interpretation.

6. Discussion

The current systematic review summarises the existing information on the correlation between ASD and detectable genetic syndromes, especially focusing on dysmorphic symptoms as signs of syndromic aetiology. The results agree that, although ASD is not a genetic syndrome and does not have pathogenic physical characteristics, a significant proportion of children with ASD manifest themselves with discernible genetic disorders that have classic dysmorphology, cognitive patterns, and medical comorbidities. This endorses the modern conceptualizations of ASD as a heterogeneous neurodevelopmental disorder that has more than one etiological path and not a unitary disorder (Lord et al., 2020; Geschwind and State, 2015).

Throughout the literature reviewed, Fragile X Syndrome, Down Syndrome, Rett Syndrome, Williams Syndrome, and Prader-Willi Syndrome were the syndromes that were most reported to be related to ASD phenotypes. These results coincide with the large-scale genomic research that suggests that a specific genetic or chromosomal abnormality can be identified in about 10-30% of people diagnosed with ASD

(Tammimies et al., 2015; Srivastava et al., 2019). Notably, the autistic characteristics evident in these syndromes are not accidental but they seem to be the product of common neurobiological processes such as impaired synaptic functioning, dysregulated expression of genes, and abnormal brain development. This overlap demonstrates the biological possibility of the manifestations of ASD in genetically defined diseases.

The presence of dysmorphic features was repeatedly found in the literature as clinically significant indicators that distinguish syndromic ASD and idiopathic ASD. The minor physical anomalies that were reported more in children with ASD who had known genetic diagnoses included craniofacial structures, ears, hands, and body proportions (Miles et al., 2008; Shen et al., 2010). These results support the idea that physical examination is still an important part of ASD evaluation, despite the presence of highly sensitive molecular diagnostic tools. Dysmorphic features are present in large numbers to be used in conjunction with chromosomal microarray analysis and whole-exome sequencing, which suggests the use of genetics-first or genetics-parallel diagnostic models in ASD testing (Schaefer and Mendelsohn, 2013).

The critical syndrome-specific ASD presentation patterns are also highlighted in the review. As an example, Fragile X Syndrome is characterised by high prevalence of ASD features associated with the dysregulation of synapses caused by FMR1 mutations, and the Rett Syndrome is

characterised by the presence of early autistic-like behaviours that subsequently develop into a specific neurodegenerative profile (Hagerman et al., 2017; Neul et al., 2014). Likewise, ASD in Williams Syndrome is of a qualitatively different nature, as there is relative social motivation but impaired pragmatic communication and interests (Klein-Tasman et al., 2018). Such differences point to the fact that the symptoms of ASD in genetic syndromes are not homogenous and have to be considered in the context of the syndrome.

Clinically, the result is very much in favor of placing genetic assessment in the standard ASD diagnostic processes, especially in case of dysmorphic traits, intellectual disability, epilepsy, or other related medical conditions. The early detection of the syndrome ASD is strongly related to prognosis since a lot of genetic syndromes are associated with predictable development patterns and are accompanied by certain health risks that need to be monitored (Srivastava et al., 2019). Moreover, proper etiological diagnosis allows families to get syndrome-specific intervention, predict the challenges in the future and get informed genetic counselling of recurrence risk.

The educational consequences of the identification of syndromic ASD also are important. Children having ASD that is linked to genetic syndromes tend to exhibit unique learning styles, behavioural tendencies, and adaptive requirements unlike those with idiopathic ASD. The inability to recognize a syndrome underlying a specific case can lead to an excessively broad-based intervention strategy that cannot sufficiently target medicine vulnerability or cognitive constraints or sensory motor difficulties. Therefore, geneticists, clinicians and special educators need to work together to facilitate individualized evidence-based educational planning.

This review has gaps in the literature even though it contributes to the same. Numerous researches concentrate on syndromes with high prevalence, whereas less prevalent genetic syndromes related to ASD are not widely studied. Also, inconsistency in the diagnostic criteria, the assessment tool, and dysmorphic features reporting restricts comparability across studies. The future studies need to focus on standardized phenotypic characterization and

longitudinal designs as the means of understanding the impact of syndromic features on ASD lifespan trajectories.

All in all, the results of this systematic review contribute to the significance of the consideration of ASD as a multidimensional concept incorporating the behavioural, physical, and genetic data. Dysmorphic features are not just side effects of the issue but are key diagnostic indicators that have the potential to revolutionize assessment, intervention and family support. The identification of syndromic ASD is thus critical in ensuring progress in precision medicine and personalized education in the treatment of autism.

The results of this review support the fact that ASD is a neurodevelopmental condition, and it is not a genetic syndrome in which ASD features would be a part of a general phenotype. Dysmorphic characteristics are vital clinical indicators that initiate new investigations into the genetic field. The timely diagnosis of syndromic ASD facilitates intervention by providing customised intervention, informs prognosis, and empowers the family with genetic counselling.

7. Conclusion

Autism Spectrum Disorder is neither a genetic syndrome nor does it have homogenous dysmorphic characteristics. Nevertheless, its common occurrence with genetic syndromes including Fragile X, Down Syndrome, Rett Syndrome, Williams Syndrome and Prader-Willi Syndrome underscores the need to be aware of syndromic manifestations. Dysmorphic characteristics present a nice diagnostic hint and are central to overall evaluation and treatment. It is necessary to use a systematic, genetics-based approach to maximize the outcomes in children with ASD and their families.

8. Clinical and Educational Implications

The identification of genetic syndromes in children with ASD is necessary to:

1. Early and correct diagnosis.
2. The knowledge of the developmental paths.
3. Planning the interventions individually.
4. Medical comorbidity presupposition.
5. Educating families about genetic counselling.

6. It is highly recommended that multidisciplinary assessment be conducted by geneticists, developmental paediatricians, psychologists and special educators.

9. Conflict of Interest

The author declares no conflict of interest. Use plural

10. Funding Statement

This research received no funding.

References

- Bennett, D. S., Bendersky, M., & Lewis, M. (2015). Autism spectrum characteristics in children with Prader-Willi syndrome. *Journal of Autism and Developmental Disorders*, 45(8), 2504-2514. <https://doi.org/10.1007/s10803-015-2462-8>
- DiGuseppi, C., Hepburn, S., Davis, J., Fombonne, E., Lee, L.-C., et al. (2018). Screening for autism spectrum disorders in children with Down syndrome: Prevalence and associated factors. *Journal of Intellectual Disability Research*, 62(5), 391-403. <https://doi.org/10.1111/jir.12471>
- Dykens, E. M., Roof, E., Hunt-Hawkins, H., & Dankner, N. (2017). Autism spectrum features in Prader-Willi syndrome: Associations with genotype and age. *American Journal of Medical Genetics Part A*, 173(1), 72-80. <https://doi.org/10.1002/ajmg.a.37918>
- Geschwind, D. H., & State, M. W. (2015). Gene hunting in autism spectrum disorder: On the path to precision medicine. *The Lancet Neurology*, 14(11), 1109-1120. [https://doi.org/10.1016/S1474-4422\(15\)00044-7](https://doi.org/10.1016/S1474-4422(15)00044-7)
- Hagerman, R. J., Berry-Kravis, E., Kaufmann, W. E., Ono, M. Y., Tartaglia, N., et al. (2017). Advances in the treatment of fragile X syndrome. *Pediatrics*, 139(2), e20162754. <https://doi.org/10.1542/peds.2016-2754>
- Klein-Tasman, B. P., Mervis, C. B., Lord, C., & Phillips, K. (2018). Social communication deficits and autism spectrum features in Williams syndrome. *Journal of Neurodevelopmental Disorders*, 10(1), 20. <https://doi.org/10.1186/s11689-018-9235-4>
- Lord, C., Elsabbagh, M., Baird, G., & Veenstra-Vanderweele, J. (2020). Autism spectrum disorder. *The Lancet*, 392(10146), 508-520. [https://doi.org/10.1016/S0140-6736\(18\)31129-2](https://doi.org/10.1016/S0140-6736(18)31129-2)
- Miles, J. H., Takahashi, T. N., Bagby, S., Sahota, P., Vaslow, D., et al. (2008). Autism spectrum disorders: A phenotypic spectrum for complex genetic disorders. *Journal of Pediatrics*, 153(5), 711-717. <https://doi.org/10.1016/j.jpeds.2008.04.053>
- Neul, J. L., Kaufmann, W. E., Glaze, D. G., Christodoulou, J., Clarke, A. J., et al. (2014). Rett syndrome: Revised diagnostic criteria and nomenclature. *Annals of Neurology*, 76(6), 944-950. <https://doi.org/10.1002/ana.24141>
- Richards, C., Jones, C., Mosse, K., Moss, J., Oliver, C., et al. (2015). Prevalence of autism spectrum disorder phenomenology in genetic syndromes: A systematic review and meta-analysis. *Lancet Psychiatry*, 2(10), 917-929. [https://doi.org/10.1016/S2215-0366\(15\)00180-2](https://doi.org/10.1016/S2215-0366(15)00180-2)
- Schaefer, G. B., & Mendelsohn, N. J. (2013). Clinical genetics evaluation in identifying the aetiology of autism spectrum disorders: 2013 guideline revisions. *Genetics in Medicine*, 15(5), 399-407. <https://doi.org/10.1038/gim.2013.32>
- Shen, Y., Dies, K. A., Holm, I. A., Bridgemohan, C., Sobeih, M. M., et al. (2010). Clinical genetic testing for patients with autism spectrum disorders. *Pediatrics*, 125(4), e727-e735. <https://doi.org/10.1542/peds.2009-1684>

- Srivastava, S., Cohen, J., Vernon, H., Raca, G., Patel, J., et al. (2019). Clinical whole-exome sequencing in autism spectrum disorder reveals variants in genes related to synaptic function. *Molecular Autism*, 10(1), 27. <https://doi.org/10.1186/s13229-019-0274-8>
- Smeets, E. E., Pelc, K., Dan, B., & de Vries, B. B. (2012). Autism spectrum disorder in Rett syndrome: A comparative study of MECP2 mutations and phenotype. *Developmental Medicine & Child Neurology*, 54(7), 646-652. <https://doi.org/10.1111/j.1469-8749.2012.04225.x>
- Tammimies, K., Marshall, C. R., Walker, S., Kaur, G., Thiruvahindrapuram, B., et al. (2015). Molecular diagnostic yield of chromosomal microarray analysis and whole-exome sequencing in children with autism spectrum disorder. *JAMA*, 314(9), 895-903. <https://doi.org/10.1001/jama.2015.9630>
- Wheeler, A. C., Hatton, D. D., Bailey, D. B., & Skinner, M. (2019). Social responsiveness and autistic behavior in boys with fragile X syndrome. *Journal of Autism and Developmental Disorders*, 49(7), 2830-2843. <https://doi.org/10.1007/s10803-019-04015-3>

