



CASE CHALLENGE



A 14-month-old female infant presented with multiple skin coloured hyperpigmented papular lesions on her left hand for past month. The lesions have been gradually increasing in size, coalescing to form plaques in a linear manner.



What's your diagnosis?

For Answer: Please refer to the last page

Sampoorna Swasthya Sanwardhan. (संपूर्ण स्वास्थ्य संवर्धन)

(In this section, we share a short article on Child Psychology, Mental Health & Wellness, Neuroscience, Emotional Intelligence & Effective Parenting for Holistic Health of Children)



Advancements in Understanding and Addressing Autism Spectrum Disorder: Insights and Innovations

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Introduction

Autism Spectrum Disorders (ASD) are complex puzzles that have intrigued researchers, doctors, and families worldwide. These disorders are characterised by difficulties in social communication and repetitive/restricted interests and behaviours. ASD impacts individuals from diverse backgrounds, transcending geographical and cultural divides. Despite its complexity, ongoing research and innovation offer hope.

As the number of ASD diagnoses continues to rise, there is a growing urgency to understand and address the challenges individuals with ASD face. Recent years have witnessed significant research progress, driven by advanced technologies and collaborative efforts. Genetic studies, brain imaging, and innovative interventions are shedding light on ASD, moving us toward a comprehensive understanding.

In this article, we explore a few such latest ASD breakthroughs.

1. Genetics and Genomics

The genetic roots of ASD emphasise the interplay between genetic predisposition and environmental factors. Traditionally, chromosomal microarray (CMA) has been a first-tier investigation test, but recent advances in whole exome sequencing (WES) have revolutionised in identifying many genes directly linked to causes of ASD. WES not only surpasses CMA in detecting relevant genetic variants but also unveils de novo variants crucial to understanding ASD's molecular basis.

In a groundbreaking study¹, Indian researchers analysed 101 patient-parent trios, revealing diverse genetic variants. De novo variants in genes linked to synaptic function, transcription regulation, and chromatin remodelling emerged as key players. This study advocates for WES as the preferred test, echoing global trends.

This research not only enhances our knowledge about genetic roles but also opens doors to targeted therapies. Insights from diverse populations, like India, enrich our understanding of ASD's genetic landscape, advancing global efforts in precision medicine for neurodevelopmental disorders.

2. Neuroimaging and Neurophysiological Approaches for Early Detection

Early detection and intervention are crucial for managing ASD. Current diagnostic method relies on standardised questionnaires or observational assessments, often identifying ASD after 24 months. Recent advancements in neuroimaging and neurophysiological testing provide promising avenues for early detection, especially in high-risk infants.

Researchers are actively exploring the utilisation of magnetic resonance imaging (MRI), electroencephalogram (EEG), and functional near-infrared spectroscopy (fNIRS) to assess neurodevelopmental differences in high-risk infants, showing promising prospects. Recent studies reveal structural and functional brain disparities linked to ASD, including altered connectivity, unusual social stimuli specialisation, and aberrant lateralization². These findings indicate the potential of neuroimaging and electrophysiological methods to accurately predict ASD diagnosis, even in infancy. Despite limitations like the absence of quantitative analysis and exclusive focus on neuroimaging and neurophysiological biomarkers, the review underscores the significant promise of these techniques in early ASD screening.

3. Medical intervention

Current medications for ASD focus on managing symptoms like irritability, aggression, anxiety, and repetitive behaviours. FDA-approved drugs include antipsychotics like risperidone and aripiprazole, which address irritability and aggression, and SSRIs like sertraline for anxiety.

The main focus of research on medications for ASD is on improving core symptoms and quality of life³. Drugs targeting glutamate receptors, particularly memantine, D-cycloserine, and acamprosate, demonstrated improvement in behavioral symptoms in clinical trials. Inhibitors/downregulators for proinflammatory cytokines like IL-6 and IL-17, such as tyrphostin AG126, resveratrol, and suramin, showed positive effects in reducing repetitive behavior and improving social interaction, core symptoms of ASD. These advancements offer hope for more effective treatments, but further research is needed to understand their mechanisms and long-term effects.

4. Embracing Neurodiversity and Advocacy

As we delve into the final section of this article, it's important to acknowledge that while it may not directly reflect the latest advancements in medical science, it embodies a significant advancement in our collective mindset toward acceptance and inclusion. The shift towards viewing autism through a neurodiversity lens is a profound step forward, fostering acceptance, appreciation, and celebration of the unique strengths and perspectives inherent in neurodiverse individuals. Here's how this approach promotes a more inclusive and equitable society:

- Shift in Perspective: Educate and raise awareness to change perceptions of autism as a natural variation in human cognition.
- Acceptance and Appreciation: Showcase the talents and strengths of individuals with autism, promoting a culture of appreciation and celebration.

- **Inclusion and Equity:** Implement policies for equal access and reasonable accommodations in education, employment, and healthcare.
- **Empowerment:** Provide opportunities for self-advocacy, skill-building, and autonomy, enabling individuals with autism to thrive and contribute.

Conclusion:

In conclusion, while the future holds promise for advancements in understanding and addressing ASD, it's crucial to highlight the significance of early diagnosis, multidisciplinary therapeutic intervention, and regular follow-up assessments. Early identification through screening enhances intervention effectiveness, while ongoing assessments ensure treatments adapt to evolving needs. Prioritising early diagnosis, tailored therapies, and consistent evaluations optimises outcomes and empowers individuals with ASD. Let's remain steadfast in our commitment to providing comprehensive care and support for those with ASD.

Resources:

1. Sheth, F., Shah, J., Jain, D. et al. Comparative yield of molecular diagnostic algorithms for autism spectrum disorder diagnosis in India: evidence supporting whole exome sequencing as first-tier test. *BMC Neurol* 23, 292 (2023).
2. Clairmont C, Wang J, Tariq S, Sherman HT, Zhao M, Kong XJ. The Value of Brain Imaging and Electrophysiological Testing for Early Screening of Autism Spectrum Disorder: A Systematic Review. *Front Neurosci*. 2022 Feb 3;15:812946. doi: 10.3389/fnins.2021.812946. PMID: 35185452;PMCID:PMC8851356.
3. Lim S, Lee S. Chemical Modulators for Targeting Autism Spectrum Disorders: From Bench to Clinic. *Molecules*. 2022 Aug 10;27(16):5088. doi: 10.3390/molecules27165088. PMID: 36014340;PMCID:PMC9414776.



CASE CHALLENGE

Answer: **LINEAR LICHEN** **SCLEROSUS**

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