Toward a Europe-wide Patient-centric Approach to Autism Spectrum Disorder

Elisabetta Vaudano

AUTISM SPECTRUM DISORDER (ASD) is a broad grouping of neurodevelopmental disorders, including autism, Asperger’s syndrome, and pervasive developmental disorder—not otherwise specified. Nearly 75% of ASD patients suffer from a range of comorbid psychiatric illnesses or conditions, which may include attention-deficit hyperactivity disorder, anxiety, bipolar disorder, depression, Tourette syndrome, and others. In addition, there is a significant co-occurrence of autism and epilepsy.

Although genetic factors may play a major causative role in ASD, other multifactorial mechanisms are also involved in etiology. The interaction of all these factors results in considerable diversity in the manifestation of the core clinical features. Given that ASD is essentially a developmental disorder, presentation will also vary with age, cognitive and learning abilities, and experience. There is also a significant sex dimension in ASD. ASD is a lifelong condition, and as such, needs will change dynamically and in respect of environmental circumstances.

Prevalence studies of ASD are complicated due to the fact that the concept of autism has been changing since its inception, from childhood schizophrenia to neuro-variation. These changes in concept have been accompanied by changes in the diagnostic threshold through which the “case” of autism is identified. The new diagnostic criteria of ASD focus on two core domains: social communication impairment and restricted interests/repetitive behaviors.

Still, the evidence is clear that the prevalence of ASD globally has been steadily increasing over the past two decades, with current estimates reaching around 1–1.5% internationally. In Europe, there is lack of an up-to-date and comprehensive evaluation of ASD prevalence, but most recent studies in member states show prevalence levels overall in alignment with global ones. Autism-specific legislation in different EU member states varies in both form and scope: from single policies in specific areas to all-encompassing national autism plans or strategies. There is also discrepancy in member states’ abilities to implement their policies.

Individuals with autism show a range of abilities and disabilities within the umbrella of an autism diagnosis. Thus, not surprisingly, the outcome in autism is highly variable. Most individuals with autism and severe cognitive and language impairments require specialist educational provision as children and will continue to need support throughout adulthood. Even among individuals with an average IQ or above (IQ 70+) and ASD, research indicates that less than a third of adults achieve a “good” social outcome in terms of employment, relationships, and independent living. Indeed, the employment rate for people with autism is between 50% and 75% lower than that for people with intellectual disability without autism and nearly all other populations with special needs. This has many costs at the societal as well as individual level, as unemployment decreases quality of life, reduces financial independence, and impairs dignity. Across the life-span, people with autism have been reported to experience a much lower quality of life compared to people without autism, and there is evidence for increased premature mortality in ASD due to a multitude of medical conditions.

As regards available treatments, there are both pharmacological and non-pharmacological options, but these are only partially effective on some of the ASD symptoms or on the comorbidities, without disease modification. Recent reviews of interventions for autism conclude that relatively few meet the methodological standards required for assessing medical or pharmacological treatments. There is also a real need and increasing evidence for home-based intervention—something rarely available in the majority of European countries.

Autism Europe has provided a recent summary of current evidence-based knowledge about autism with a wide audience in order to improve general understanding and impact positively on practice and policy in Europe.

The diversity of people with autism means a “one size fits all” solution to improving the outcomes for people with ASD is unlikely. A highly personalized approach may be needed, with tailor-made solutions based not on group-defining characteristics but rather on the appreciation of cross-domain bio-behavioral profiles at the individual level. In this context, as shown by the articles in this special issue, digital solutions such as virtual reality (VR) may have real potential for an innovative impact on ASD, be this an improvement in daily living activities, on social interactions, or for life outcomes such as education or employment retention.

A further untapped opportunity for VR is in the ASD diagnostic field. A personalized ASD diagnostic must reflect the relevant domains that may be impaired in people with ASD, and such tests need to be comparable, engaging, and sensitive across large age and ability ranges. VR can create tailor-made convincingly realistic simulations of experiences that may provoke symptoms, which otherwise would not be easily observable at the doctor’s office. These coupled with sensors and other digital solutions may offer further opportunity for unbiased measuring and prolonged monitoring of
disease status or even in the future for seamlessly combining treatment with the diagnosis.

Many challenges still have to be addressed successfully for people with ASD to benefit from such solutions. First, any solution has to be specifically adapted for people with ASD (e.g., consideration of the potential higher risk of seizures in this population). Analytical validation of these tests will require creating age norms and optimizing psychometric properties. Validated biomarkers will be needed to support regulatory acceptance and access for patients. Most importantly, any solution has to be demonstrated to have a positive impact on the health outcomes and quality of life for those with ASD.2

For this endeavor to be a success, a co-creation and open collaboration model is needed, keeping the users—the people with ASD—at the center. They need to be engaged with the other stakeholders from day 1 as full and equal partners in research. In addition, a large amount of high-quality data is necessary to ensure the necessary independent validation.

The Innovative Medicines Initiative a has been a pioneer in the approach to research in the field of innovative ASD treatments via its project EU-AIMS b and now the even more ambitious AIMS2-TRIALS project. c

AIMS-2-TRIALS brings together people with autism and their families, academic institutions, charities, and pharmaceutical companies to study autism and provide an infrastructure for developing and testing new therapies. In line with the autism community’s priorities, the consortium also focuses on why some people with autism develop additional health problems that severely impact both the quality and length of life.

AIMS-2-TRIALS has created the world’s largest database of multimodal data on people with ASD. The project is developing the first European clinical trials network for autism, as well as allowing for an internationally integrated partnership with charities, government agencies, and industry to determine rapidly if therapies are effective. Partnership with people with autism and their families and carers will be a crucial part of developing therapies that achieve the outcomes that matter most to people with ASD. There is a clear opportunity for the work of the consortium to inform and support the further development of non-medical treatment options such as VR. d

Elisabetta Vaudano
The Innovative Medicines Initiative

Notes

b. https://www.eu-aims.eu/
c. https://www.aims-2-trials.eu/

References