

CA AE 070523



**Severe Autism**  
**High support needs**



Outline:

Introduction

Etiopathology

Treatments

Training

On behalf of: Eng. Giovanni Marino  
Prof. Carlo Hanau

ANGSA Italy  
APRI Italy

Prof. Ettore Focardi  
University of Florence  
ANGSA Toscana, Italy

# INTRODUCTION

**The autism is a disorder not limited to the childhood but it's all lifetime long.**

DSM5 Autism classification is in 3 levels:.

- 1) Level 1, low level support needs
- 2) Level 2, medium support needs
- 3) Level 3, high support needs

The autism is originated by a genetic disorder with multisystemic involvement (gut, immune system,...), referred as monogenic condition. In other cases there is a susceptibility to autism due to a combination of genes interacting with the environment. We can report the recent words from Dr. Roberto Keller, Italian psychiatrist, director of regional autism center in Turin (Italy):

*It is worth to subdivide 'autisms' on the basis of cognitive level. People on the spectrum and with intellectual disability have poor prospects to reach a good level of autonomy and integration compared with 'high functioning' people.*

*The luckiest, can stay in family with the help of educated operators or eventually go into dedicated structures, rare event.*

*Until few years ago those people would be destined to psychiatric hospital, now they can be mostly referred in residences for disabled. Unfortunately these residences are almost **empty containers** without education & rehabilitation/habilitation programs.*

## Autism etiopathology (origin):

Since more than 50 years the genetic contribution to autism has been suggested, but only at the beginning of 21th century the first genes have been identified.

Actually ~**133 genes** are involved in monogenic conditions. Many others genes are involved as susceptibility factors.

The analysis of cellular and animals models have shown that the majority of these genes plays an important role in the brain development.

In particular these genes modulate the number and the functioning of the contact points between neurons (synapsis).

The use of recent investigation methodologies has allowed to identify a genetic cause of autism in ~14-20% of persons with autism.

Recently (2022), an additional technological improvement (whole genome sequencing, WGS) allows the characterisation of different typologies of variants (sequential, structural, nuclear and mitochondrial) in the same experiment.

In this study, the analysis of more than 5K individuals with autistic spectrum disorders has identified 67 new candidates genes with high level of confidence.

## Autism etiopathology (origin) 2:

At this point, next important step would be to share the work in different groups at 'world level' studying each a monogenic condition to possibly arrive to personalized therapies.

The molecular targets arising from this study may also eventually imply pharmacological interventions.

Recently arrived a drug proposal for the Rett syndrome.

In addition, some research is under way, in haploinsufficiency condition, to try to stimulate the working gene to produce additional proteins coded by that gene when the other allele is not working.

**This is one point that our community has to take into account and strengthen.**

## Treatments for autistic persons with high support needs:

- **Regulatory enforceability of people with autism does not distinguish needs**
- Normally, treatments for autistic people is addressed to persons with less problematic conditions because is easier and more sustainable (funds). The hope is to try to increase the efforts in projects addressed to high support needs. This could help to find solutions when these persons loose their family support and avoid the easier way of Institutionalization.
- EU disability platform has to include research, treatments development
- Appropriate structures have to be developed to meet these objectives
- **AE has to strengthen these aspects on the transition from Institutional to community based care for high support needs autistic people**
- Some attention has to be taken into account to the selection of people inside a particular committee ( find somebody working in the field )
- **Promote training at every level (specialists, operators, families..) to provide adequate and appropriate support.**

## Aim: avoid this

Avoid situations like **Sabine**, autistic (level 1) french girl at age 20 (left picture), then at age 28 enters in a psychiatric hospital for 5 years, later (right picture) you see the effects of probable wrong treatments.



Age 20



Age 38

# Problem position

The involvement of major specialists in Italy guided me in building an appropriate program:

Autism manifests itself in childhood and during this period it is necessary to carry out constant educational activities, **evaluating best available therapies** through the creation of multidisciplinary teams of specialists who propose and measure the outcomes of the **therapeutic Individual Program 'tailored' to each child taken in charge.**

**Recommendation has to be done to perform genetic research to eventually identifies etiopathology.**

Only in this way can the patient be guaranteed a better quality of life and an easier capacity for social integration and autonomy

# Problem position: adult life

For many years, in fact, the topic of autism in adulthood has been underestimated both in the clinical and academic fields.

Moreover, the scientific misunderstanding preserved, alas, in relation to adults with autism, they are regarded as people with social integration difficulties or psychopaths.

The "educational/habilitative" care and approach must remain constant, as is the case for all other psychiatric disorders with a chronic course.

The real problem, in fact, is that after reaching the age of majority (18 y), patients are taken care of by psychiatric facilities that deal with little of the pathology, thus not ensuring continuity of care.

The shortcomings lead, therefore, to limiting treatment to the prescription of psychotropic drugs, underestimating the better and longer-lasting effects of psychotherapy and rehabilitation.



# Way to go



Currently, data indicate that there is a percentage of adult patients with autism who were **not previously diagnosed**.

In these cases, a clear assessment is not easy and even more difficult is the identification of a therapeutic pathway aimed at improving the relationship w others. For these people, **it is good to start with targeted interventions with consistent sessions of cognitive-behavioural psychotherapy given by appropriate trained operators, integrated, eventually, to drug therapy,** so as to evaluate the mid- and long-term effects.

Of course, it is necessary to consider the person, his or her needs as an adult and think about support that gives space for the possibility of independent living and a possible job placement.

No less important, then, is **the support to be provided to families**, with the possibility of targeted therapeutic interventions, to meet the requirements of the **Individual Life Project** indicated by a multidisciplinary team in charge of the patients. The designation of a **'case manager'** is essential to follow and evaluate the outcomes of the actions taken.

# Professional figure training



All this implies the necessary and appropriate training of the professional figures involved from the onset of the syndrome:

**pediatrician, child neuropsychiatrist, psychologist, primary care physician, support teacher, pedagogist and then psychiatry, rehabilitation therapists, professional educators, staff already working in the contexts of Mental Health Departments.**

There is still a long way to go to achieve these goals and help overcome the small and large difficulties that, every day,

**1 in 60 people present with autism**

Participation in community life is possible: the secret is in courage and confidence, in the skills and seriousness with which to approach the disorder.

# Master Course



The master course on autism started at Pisa University on January 2018.

Duration: 1 year. Lectures: 256 hours (3 days/month). Traineeship: 216 Hours

Final exam: Thesis discussion on a clinical case.

Application with master degree on Psychiatry, child neuropsychiatry, psychology, Pedagogy etc..

Participants: ~ 14/year, majority of psychiatrists. More than 60 specialists trained.

this year: VI edition

Outcome: **in several Tuscany territories, mental health medical services created multiprofessional groups to satisfy the needs of adult autistic population.**

# Conclusion

The conditions of autistic people with high support needs require more attention from our group of associations.

We discussed with few of them and we found agreement on this point.

We are available to take part in this effort and we ask Autism Europe Council of Administration to discuss on this point and eventually endorse this position.

We propose to create a group of people interested and discuss how to attack this question.

Thanks for your attention

[ettore.focardi@gmail.com](mailto:ettore.focardi@gmail.com)