

Diagnostic manual of “Regression of Social and Communication Skills in Down syndrome”

Deterioration in daily living abilities, particularly social and communication skills, within a relatively short period (one year or two) has been associated in adolescents and young adults with Down syndrome. Although its clinical characteristics resemble those of “dementia” or “depressive illness”, it is supposed to be a distinct clinical entity with a different etiology, because of the following reasons: it neither seems to progress (as would be expected with dementia) nor resolve on antidepressants (as would be expected with depressive illness). It also seems distinct from a combination of Down syndrome and autism spectrum disorders.

We therefore propose that this deteriorated condition be named “Regression of Social and Communication Skills in Down syndrome”. However, further investigation is essential to understand more details.

Diagnostic criteria : Proposed by Study Group in the Ministry of Health, Labor and Welfare in 2011

- 1. Motor retardation**
- 2. Lack of facial expression**
- 3. Mutism**
- 4. Lack of interpersonal response**
- 5. Markedly diminished interest or pleasure**
- 6. Social withdrawal**
- 7. Sleep disturbance**
- 8. Appetite loss**
- 9. Weight loss**

These symptoms become apparent within a relatively short period, and continue more than several months.

- 5 items or more : determined**
2-4 items : suspected
0-1 items : denied

Differential Diagnosis

1. encephalitis, brain tumor, meningitis, head injury, or these sequela
2. atlas-axis dislocation
3. severe hearing loss or advanced visual impairment
4. thyroid dysfunction
5. serious liver disease
6. arthritis, myositis
7. depression or depressive illness
8. epilepsy
9. pervasive developmental disorder
10. organic disease which a cause except for the above can be specified for