# Diagnosis of Autism in Children With Down Syndrome

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## Synopsis

As many as ten percent of persons with Down syndrome may also suffer from autism. Exact data is difficult to obtain. Many cases go undiagnosed, or are diagnosed at a later age. Many diagnosticians are unawares that the two conditions may exist or are reluctant to apply the second diagnosis. Autism diagnosis is much more complicated than Down syndrome; there is no blood test, genetic marker, facial features, or other characteristics that apply to all autistic persons. The diagnosis is subjective, depending on meeting observations of certain behaviors. Diagnosis and treatment of autism is much more critical than for Down syndrome. Without early detection and intervention the life of a person with autism may be much more limited than that of a person with Down syndrome and often results in autistic children living in a residential school rather than at home because of inability of the parents and the school system to cope with the child's behaviors.

## Symptoms of autism in Down syndrome

The key area affected developmentally in a child with Down syndrome is the cognitive area, the development of thinking, reasoning, and understanding. Delays are expected in this area. Another area of development is the social and emotional development. These areas develop more normally in a child with Down syndrome but not in a child with autism. Coleman and Rogers (1992) give the following account of the expected social and emotional development in a child with Down syndrome.

*Most babies with Down syndrome show the least delay in social and emotional development, smiling when talked to at 2 months (range 1.5-4 months), smiling spontaneously at 3 months (range 2-6 months, and recognizing parents at 3.5 months (range 3-6 months); each of these milestones show only a 1-month delay on average. Although some studies suggest that the intensity of affective responses such as smiling and laughing may be slightly less than that shown by ordinary babies, parents respond warmly to the onset of smiling and eye contact. The Down syndrome babies begin to enjoy pat-a-cake and peek-a-boo games at about 11 months (range 9-16 months), which is about 3 months later than ordinary babies. Studies in the second year of life show the babies to be skilled in social communication even using social skills to attempt to distract*

*an adult from a task the baby does not want to attempt. The babies are warm, cuddly, and normally responsive to physical contact, unlike babies with some other types of disabilities such as autism.*

*This normal emotional responsiveness continues into adult life, and as studies of teenagers have shown, it develops into proper empathy, making the person with Down syndrome a sensitive and socially aware person to live with.*

So the key areas to watch for in a child with Down syndrome suspected of having a complicating behavior disorder such as autism is in the social and emotional areas. Some professionals will argue that social and emotional development can be expected to be affected by delays in cognitive development and it is not evidence of a separate disorder. This is where the subjective nature of autism diagnosis comes in. It's a matter of degree.

Some key behaviors that may point to the possibility of autism in a child with Down syndrome are:

1. Extreme Autistic Aloneness - The child does not relate to people normally and seems to prefer to be left alone. The child seems to consider other persons as objects, not people. He will not join in group play with other children. Unlike children with Down syndrome, who are very lovable and huggable, the autistic child does not want to be held.
2. Anxiously obsessive desire for the preservation of sameness - Any differences in daily routines can cause a large upset.
3. Lack of eye contact - Autistic persons typically do not make eye contact but will look away or "right through" other people.
4. Shows repetitive, "Stereotypical" movement, like sitting for long periods of time with an object in his hand and just waving it back and forth looking at it.

A checklist of autistic behaviors is presented in Figure 1. Some of these characteristics are normal, up to a point, in a child with Down syndrome. This complicates the diagnosis of autism in those children. In the next section, the DSM-IV criteria for diagnosing autism is presented with annotations as to whether each behavior is more prevalent in autism than in Down syndrome.

**Individuals with autism usually exhibit at least half of the traits listed below. These sympthoms can range from mild to severe and vary in intesity from symptom to symptom. In addition, the behavior usually occurs across many different situations and is consistently inappropriate for their age.**

| | | | |
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| **Difficulty in mixing with other children** |  | **Insistence on sameness; resists changes in routine** |  |

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| **Inappropriate laughing and giggling** |  | **No real fear of dangers** |  |
| **Little or no eye contact** |  | **Sustained odd play** |  |
| **Apparent insensitivity to pain** |  | **Echolalia (repeating words or phrases in place of normal language)** |  |
| **Prefers to be alone; aloof manner** |  | **May not want cuddling or act cuddly** |  |
| **Spins objects** |  | **Not responsive to verbal cues; acts as deaf** |  |
| **Inappropriate attachment to objects** |  | **Difficulty in expressing needs; uses gestures or pointing instead of words** |  |
| **Noticeable physical overactivity or extreme underactivity** |  | **Tantrums - displays extreme distress for no apparent reason** |  |
| **Unresponsive to normal teaching methods** |  | **Uneven gross/fine motor skills. (May not want to kick ball but can stack blocks.)** |  |

***Adapted from the original by Professor Rendle-Short, Brisbane Children's Hospital, University of Queensland, Australia.***

**Figure 1. Checklist of autistic behaviors**

# Autism and PDD - Definitions

The official term for autism, as defined by the American Psychiatric Association in their DSM-IV, is Pervasive Developmental Disorders(PDD). The Pervasive Developmental Disorders is a class of five diagnosable disorders including Autistic Disorder, Rett's Disorder, Childhood Disintegrative Disorder, Aspergers's Disorder, and Pervasive Developmental Disorder Not otherwise Specified. Sometimes the term autism is used to describe the Autistic Disorder and sometimes it is used to describe all five PDD's. There are also other related diseases that together are called the autistic spectrum disorders. For purposes of this article I assume autism to include all five PDD's.

# Diagnosis of Autism in Down Syndrome - DSM-IV Checklist

Autism is diagnosed by evaluating the behavior of the patient. There are sixteen descriptive symptoms to look at. If an appropriate combination of eight are displayed, a diagnosis of autism is reached. These symptoms are grouped into four general areas; Severely Impaired Social Interactions, Severely Impaired Communications and Imagination, Extremely Limited Interests and Activities, and First Observed in Infancy or Early Childhood. In some cases, as in Down syndrome, mental retardation may mask the autism and a diagnosis may come later or not at all.

The diagnostic criteria for Autism as given by the American Psychiatric Association is contained in their Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition. (DSM-IV) The diagnosis is quite complex and is recognized only when done by a qualified professional.

Diagnosing autism in a Down syndrome person is a bit trickier than in a non-Down syndrome person. Some of the criteria for autism is normally present in Down's persons and in itself does not mean that autism is present. Following is DSM-IV for the Autistic Disorder with bullets after each criteria to indicate if that criteria is normally found in Down syndrome or if it is more indicative of autism. Diagnosis for the other four PDD's is similar to that for the Autistic Disorder, generally with certain items deleted. When diagnosing autism in a Down's person, the diagnostician must examine each criteria in terms of whether it is a normal trait for the person with Down syndrome and, if it is a normal trait, is it more severe than normally found in a Down's person.

A. A total of six (or more) items from (1), (2), and (3), with at least two from (1) and one each from (2) and (3).
    1. Qualitative impairment in social interaction, as manifested by at least two of the following:
        a. Marked impairment in the use of multiple nonverbal behaviors such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction.

- *Not normally seen in Down syndrome, more indicative of autism.*

b. Failure to develop peer relationships appropriate to developmental level.
   - *Not normally seen in Down syndrome, more indicative of autism.*

c. A lack of spontaneous seeking to share enjoyment, interests, or achievements with other people (e.g., by lack of showing, bringing, or pointing out objects of interest).
   - *Somewhat true in Down syndrome but much more pronounced in autism.*

d. Lack of social or emotional reciprocity.
   - *Somewhat true in Down syndrome but much more pronounced in autism.*

2. Qualitative impairments in communication as manifested by at least one of the following:
   a. Delay in, or total lack of, the development of spoken language (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime).
      - *Not normally seen in Down syndrome.*

   b. In individuals with adequate speech, marked impairment in the ability to initiate or sustain a conversation with others.
      - *Normally seen in Down syndrome, although not to the extent seen in autism.*

   c. Stereotyped and repetitive use of language or idiosyncratic language.
      - *Seen somewhat in Down syndrome but not to the extent seen in autism.*

   d. Lack of varied, spontaneous make-believe play or social initiative play appropriate to developmental level.
      - *Not normally seen in Down syndrome.*

3. Restricted, repetitive, and stereotyped patterns of behavior, interests, and activities, as manifested by at least one of the following:
   a. Encompassing preoccupation with one or more stereotyped and restricted patterns of interest that is abnormal in either in intensity or focus.
      - *Not normally seen in Down syndrome.*

   b. Apparently inflexible adherence to specific, nonfunctional routines or rituals.
      - *Not normally seen in Down syndrome.*

   c. Stereotyped and repetitive motor mannerisms (e.g., hand or finger flapping or twisting or complex whole body movements).
      - *Not normally seen in Down syndrome.*

   d. Persistent preoccupation with parts of objects.
      - *Not normally seen in Down syndrome.*

B. Delays or abnormal functioning in at least one of the following areas, with onset prior to age 3 years: (1) Social interaction, (2) Language as used in social communication, or (3) Symbolic or imaginative play.
    - *Children with Down syndrome will normally display some deficits in these traits depending on the severity of mental retardation. Since this is not entirely unexpected, it is usually ignored in the records and not so noted. Therefore when autism is diagnosed at a later age and the records or memory is searched for this evidence it is either not found in the official records of the child or it is forgotten about because it was not deemed all that unusual at the time. The diagnostician may then discard the possible diagnosis of autism because the appearance is given (falsely) that these traits were not present prior to 36 months of age. Because of this quirk in the diagnosis, there probably are many persons with Down syndrome with autism but it is not so diagnosed.*

C. The disturbance is not better counted for by Rett's Disorder or Childhood Disintegrative Disorder.

# Issues in diagnosing autism in persons with Down syndrome

There are a number of reasons why there are not very many reported cases of autism in persons with Down syndrome.

One issue in the diagnostic criteria is the requirement for onset before age three. In the case of Down syndrome, the parents of a young child with Down syndrome and the professionals attending them are concerned with the Down syndrome itself with its possible complications of cardiac problems, leukemia, and other problems. Delays in development are expected. Given the expected delays in development, it simply does not occur to parents or professionals that autism may be present. The parents have probably never heard of autism and are completely unawares of symptoms. The same can be true for some of the professionals dealing with child with Down syndrome. Only the most astute of diagnosticians is likely to recognize autism in a child with Down syndrome before age three. Only when the child becomes older, or if the parent persists, may it become clear that autism is present. But some diagnosticians will apply the three year rule and decline the autism diagnosis later on because the records do not show the presence of the symptoms before age three. The issue of early onset, then, becomes a major obstacle to the diagnosis.

Another issue is that persons with Down syndrome come with a few of the DSM-IV criteria already there as a part of the Down syndrome. When looking for eight of sixteen specified criteria the diagnostician may be tempted to excuse those symptoms

characteristic of Down syndrome and not garner enough symptoms to give the autism diagnosis.

Also, some professionals may consider only the Kanner autism, where there is little or no mental retardation, to be the true autism and decline a diagnosis in the Down syndrome case where there is usually some mental retardation. Others may decline the diagnosis because autism causes are usually cloudy or unknown in an individual case, and given the mental retardation in Down syndrome, may say that since there is a known cause, they decline the diagnosis.

Some diagnosticians may not wish to burden the family with an additional label, i.e. autism. This denies the family the opportunity to join autism support groups and to seek out autism interventions. While the autism and Down syndrome treatment protocols overlap and have many similar characteristics, the autism treatments are much more intense. And treatment for autism is more critical in averting lifetime consequences .

Most of the above issues are addressed in the textual portion of DSM-IV, in favor of giving the autism diagnosis when the behaviors are present. There is nothing in either the textual matter or in the diagnostic criteria which suggests any aversion to diagnosing autism in a person with Down syndrome and/or mental retardation. The textual matter specifically states that many autistic persons are also mentally retarded. It also states that if there is a loss of data from early years or the data is unclear as to whether the symptoms were present before age three, that the autism diagnosis should be given.

I am in contact with a number of parents with children having the dual diagnosis of Down syndrome and autism. All have reported extreme difficulty in obtaining the dual diagnosis. They could not understand the failure of their children to develop socially and emotionally, with deficits beyond that normally found in a child with Down syndrome. They had to shop around and practically beg the professionals to tell them what was wrong with their child. These are not cases of hypochondriac parents trying to pile up diagnoses but rather concerned parents who want to know what is wrong with their child.

# Studies

Studies of autism in Down syndrome are quite limited. Ghaziuddin (1992) found two children who fulfilled DSMIII-R criteria for autism from an estimated total of forty with Down syndrome. Wing and Gould (1979) diagnosed four children from a group of thirty child with Down syndrome as having autism or were within the spectrum of the autistic group. Turk (1992) reported 9% of his series of children with Down syndrome met the full criteria for autism. Lund (1988) diagnosed five adults with autism from a group of forty four with Down syndrome. Bregman (1988) and Wakabayashi (1979) also described children with the dual diagnosis. Howlin, et al. (1995) described four boys with Down syndrome who were diagnosed with autism at ages eight to eleven. The authors report that, although there is no epidemiological study of the prevalence of autistic spectrum disorders in a large population of children with Down syndrome, the evidence currently available suggests that it is in the order of 10%. Also in the article, the authors

reported that M. Ghazziuddin had personally communicated that he found autism in10% of his subjects with Down syndrome. Dr. Ghazziddin also communicated personally to me that 10% would be a reasonable number. Ghaziuddin (1997) describes three persons with Down syndrome and autism. Gillberg, et al. (1986) found 5% of subjects with autism in 20 persons with Down syndrome.

A number of studies of very large Down syndrome populations were conducted which resulted in very low rates of autism. Gath and Gumley (1986) found 1% of subjects with autism in 193 persons with Down syndrome. Myers and Pueschel (1991) found 1% of subjects with autism in 497 persons with Down syndrome. Collacut, et al. (1992) found 2.2% of subjects with autism in 371 persons with Down syndrome. The lower autism rates in these studies may be deceptive. The key issue in these studies was to determine susceptibility of persons with Down syndrome to psychiatric disorders. The studies were done by researchers looking for occurrences of psychiatric disorders in mentally retarded children and adults. They were not specifically looking for autism. Diagnoses of Depression, Psychoses, Personality Disorders, Conduct Disorders, Schizophrenia, Emotional Disorders, autism, and many others were made. Some were done using 1980 versions of DSM III or ICD-9. One study was done by examining old records and making a retroactive diagnosis based on observations made years before by people laboring under older concepts, such as when autism was routinely diagnosed as schizophrenia. Two authors admitted that the persons diagnosed with psychoses might also have been grouped as having autism. One author assigned the diagnosis of psychoses in several cases of autistic-like behavior because the behavior wasn't reported before the age of 30 months. I went through these studies and reassigned those cases where the author suggested autism could have been assigned instead of other disorders and recalculated the autism rates . They came out as high as 16%.

As mentioned earlier, diagnosis of autism is rather subjective to begin with, and gets even murkier in persons with Down syndrome and other known forms of mental retardation. Finding clear and valid statistical data is very difficult due to the vagaries of autism diagnosis. Establishment of a clear scientific statistic is not the point. The point is that the two disorders do indeed co-concur, and in significant numbers. Since the stakes are much higher in autism as mentioned earlier, it is critical that autism be recognized when it is present in children with Down syndrome so that appropriate treatment protocols can be pursued and the parents can look outside the Down syndrome community.

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Revised: April 26, 1999