

Presentation and Discussion of a Japanese Paper entitled "Autism detection in children under 18 months - sensitivity of medical exam screening in infancy and early childhood"

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We will discuss the study entitled "Autism detection in children under 18 months - sensitivity of medical exam screening in infancy and early childhood" (Yasuda Mutual Life Insurance Company "Investigation and Study Promotion" Vol.32 (Page 63-70) 1996).

This study attempts to actively diagnose autism in its initial stages and evaluate standard screening sensitivity based on the current difficulties encountered when detecting autism. It is very important to diagnose autism early on, and to carry out early intervention, thus the importance of this study.

In the north zone of the city of Yokohama, the fail-safe clinical system is established. It permits the introduction of false negative screening examples in early intervention through the alliance of related organizations, carrying out mass screening of infantile autism in 18 month old children. In this study the sensitivity of autism screening is sought in the medical examination of children under 18 months of age. It covers 50 cases in which infantile autism (ICD-10) was diagnosed after 4 years, and which had received medical examination at the age of 18 months, in public

health offices of this zone. Consequently, the sensitivity of the screening was 74.0%. As the screening medical exam of children under 18 months allowed for an early start in treatment, the importance of early intervention could be confirmed.

With the publication of the DSM-III (1980), the diffusion of operational diagnosis began, and at present the ICD-10 (1993) and DSM-IV (1994) are taken into consideration. Japan also follows that tendency.

According to these, characteristic symptoms of autism show up before the age of 3. However, in most cases parents visited a doctor when their children were as young as 4 or 5 years of age, and the clinical picture before the age of 3 depended only of the retrospective information given by the parents. In the 90's, studies in which professionals directly observed the clinical image of autism in children under 3 years of age were started. But the operational diagnostic criterion of autism is established according to the clinical picture of children between 4 and 5 years of age. Therefore it cannot be applied to children younger than 3. For children under 3, mass screening with indicators of

behavioral characteristic whose development can be affected by autism is required. When the system of early clinical intervention is applied, it is important that autism be detected in its early stages by the mass screening method. Therefore, one must first examine the relevance of mass screening.

Generally, among the indicators with which the relevance of mass screening is examined, sensitivity and specificity are of outmost importance. The sensitivity involves the ability to judge an ill patient as positive, and specificity indicates the capacity of judging a healthy patient as negative. In the screening system of the city of Yokohama, the existence of false negative examples in the medical exam of children under 18 months was confirmed. Therefore, in order to define the sensitivity of the system, it is necessary to define the percentage of false negative cases. As far as the authors know, there is no study designed to guarantee the detection of false negative cases.

This study attempts to clear up the sensitivity of mass screening of autism in children under 3. To do this, 3 conditions are required: (1) Existence of mass screening that can be applied before 3 years of age (2) Final diagnosis after 3 years of age by a child psychiatrist (3) The design of a study that allows certain detection not only of the actual positive cases, but of the false negative cases.

The exam consists of the observation of the child's behavior, diagnosis through communication questions and personal relation, language, gaze agreement, interest in other children, imitation of actions, finger indication when questioned, and answers using photos and drawings. A public health nurse carries out the exam. The children in which the possibility of development problems is detected are observed for a specific period of time in the corresponding public

health offices, and then are examined in the department of Pediatrics of the Yokohama Integral Rehabilitation Center (YRC). If a diagnosis is defined, they can undergo the early intervention system. As for the false negative cases, early intervention begins when the difficulty is solved.

In this study, among the children who received medical examination in YRC, 50 cases which were diagnosed with infantile autism by a child psychiatrist after 4 years of age and who had received medical examination at 18 months of age are examined. As for the method, based on the data of the medical exam of 18 month children, the child subjects were divided into 2 groups: a group of real positives who were judged as needing observation, and a group of false negatives in which no abnormality was noted in the exam. This illustrated the sensitivity of the screening exam (the percentage of real positives in the 50 cases).

The relation between the screening sensitivity and whether the setback phenomenon alters the result or not was also studied. As for the setback phenomenon, for this study the definition "a phenomenon that shows disappearance, involution, or developmental arrest that lasts for more than 1 year, after acquiring some speech or the non-verbal function of communication" was adopted. To define whether this phenomenon exists or not, the data of the exam of 18 month children was consulted, as well as the opinion of the YRC, and the comments of the parents. To define if there is relation to other intellectual handicaps, those scoring over IQ70 in the Tanaka Binet test (High-functioning autism - HFA group) were included, while those who obtained less than IQ70 (Low-functioning autism - LFA group) were ignored.

Consequently, as for the screening sensitivity of the examination of 18-month-old children, in the 50 subject

cases there were 37 real positives and 13 false negatives. Therefore, as the screening sensitivity of autism is 74.0% (37/50), the fail-safe function was considered high, and confirmed the importance of the function of mass screening that is carried out in public health offices of the city of Yokohama.

As for the setback phenomenon, there were 11 cases in which this phenomenon was spotted after the 18th month of age. There were 4 real positive cases and therefore the screening sensitivity was of 36.4%. There were 39 cases in which the setback phenomenon wasn't spotted after the 18th month, and 33 cases that had been diagnosed as real positives. Therefore the sensitivity of screening is 84.6%, and there is a significant difference between the 2 groups.

14 HFA were found and among them 9 cases were real positives. The screening sensitivity was of 64.3%. 36 LFA were found and among them 28 were real positives. Therefore, the sensitivity of screening was 77.8% and no significant differences between the 2 groups were found.

As for the beginning phase of early intervention, the group which was diagnosed as real positive in the exam of the children of 18 months of age had an average age of 2 years and 8 months when intervention was begun, and for the false negative group the average age was 3 years and 7 months of age. In both cases they received early intervention at YRC.

With this result in mind the authors considered that among the existing studies addressing the early diagnostic of autism that were conducted based on direct observation by specialists, there are various cases in which mass screening was carried out, such as examination of children of 18 months of age, but none of them in which the relevance of screening itself was analyzed. For example, Baron-Cohen carried out mass

screening using a checklist that he himself developed.

This exam showed a highly specific result, but follow-up was only carried out for the real positive cases and nothing about the screening sensitivity was studied.

Among the epidemiological studies there are many reports in which the autism rate reaches 0.1-0.2%, but one must consider the possibility of the existence of false negative cases.

The screening sensitivity of the exam of 18-month-old children in the public health offices of the city of Yokohama is 74%, and therefore this exam proves to be useful for the screening of autism. At the same time, it is very important that the existence of false negative cases was considered and calculated to correct its participation. False negatives could be detected thanks to the early detection system based on a tight network of related organizations such as the public health office, provided by the YRC, this system functions like a fail-safe for screening.

Something that stands out is the fact that among the cases that displayed the setback phenomenon after the 18th month, 36.4% of these children were diagnosed as real positives before the 18th month of age. In these cases, despite receiving comments from parents saying that they never noticed any signs of involution or developmental arrest, the public health nurses had already detected some sort of developmental difficulty. Generally, it is hard to detect autism in cases that display the setback phenomenon in the examination test that is carried out during the child's 18th month. As for the cases of high functioning autism, there is a possibility of diagnosis by a specialist at a later age when autism that is accompanied by intellectual difficulties. However, the examination of 18 month olds (with or without intellectual difficulties) that is

carried out in the public health offices of the city of Yokohama detects, to a certain level, early symptoms of autism.

The authors arrived at the conclusion that screening of the examinations of 18 month old children has a high influence on the timely delivery of early intervention. Through the high sensitivity screening, autistic children are diagnosed, and it allows for an even earlier intervention. The average age of false negatives who start having appointments with specialists is 3 years and 7 months, and as an age to start receiving intervention, it is quite early. The high sensitivity of the examination of 18 month old children strengthens fail-safe function against the false negative cases. Therefore, the beginning phase of early intervention advances for these cases.

A task that remains is conducting an analysis of the screening process carried out by public health nurses in the examination of 18 month old children to establish a more precise method of screening to detect autism. The method for examining children carried out by the city of Yokohama may be applied to any city and zone, but an organized early intervention system is necessary. If early detection advances on its own, it presents grave ethical issues. Therefore, aside from the diagnosis and processing there is need to establish a global social system, such as family support.

These are the aspects covered by the study. Thanks to this paper it is known that the city of Yokohama is making great efforts to detect and support autistic children. But the concept of subgroups of autism is expanding. Besides autism of the Kanner type, there is high functioning autism, Asperger Syndrome, and others. There are differences among the opinions of specialists regarding the concept and criterion to be

judged. In reality there are many cases in which the child psychiatrist may not know how to diagnose his/her patient. And we must point out that there is a skeptic opinion as for the diagnostic criterion of DSM-IV or ICD-10.

Lorna Wing, writer of "The Spectrum of Autism", stated: Many things have been cleared regarding autism, but not its cause or mechanisms. Regarding autism diagnosis and sub-group definitions, no tests have been developed. Many uncertainties remain regarding autism diagnosis. Most autistic children seem physically normal, and diagnosis is based upon behavioral patterns present in their infancy. Current classification systems, such as ICD and DSM, considered social interrelation impediments, communication impediments, character superposition disorders, and repetitive patterns as indispensable diagnostic characteristics. However, misdiagnosis can easily occur due to the following reasons:

- 1) A broad variety of similar conditions exist. Some are very hard to diagnose.
- 2) Autism spectrum disorders can occur together with any level of general intelligence, from profound disability to well above average.
- 3) Physical diseases or developmental anomalies can coexist with autism. Epilepsy is commonly associated.
- 4) The behavioral pattern varies with age.
- 5) Environmental conditions cause behavioral changes.
- 6) Surrounding individuals cause behavioral changes.
- 7) Education affects the behavioral pattern.
- 8) The individual's personality reflects in and influences his behavior.

Therefore, the diagnosis requires full analysis of past personal history based on all acquirable information. This requires time, and jumping to conclusions can easily result in misdiagnosis.

It is currently difficult to discern mild intellectual developmental arrest from high functioning autism, Asperger type autism, ADHD, LD, etc. This study diagnosed infants based on clinical pictures, but a detailed description of the examination is lacking. Therefore it cannot be denied that doubt remains regarding screening sensitivity, the diagnostic criteria for autism, and their relevance.

Problems will arise when diagnosing the disease based on screening alone. When considering the degree of disturbance the diagnosis can cause the parents, it should only be communicated after establishing an adequate family support system. In this regard, the city of Yokohama has already established separate clinical and support systems, and we expect future strengthening of the support component.

During the past 10 years the concept of infantile autism has expanded. That is to say, the existence of the infantile autism sub-group other than typical Kanner type, was determined and included in the concept of "autism" in the broad wide sense of the word. As mentioned previously, Asperger syndrome is one example. So there is a widespread tendency to diagnose 'autism' liberally, even when the clinical picture is diverse. We must agree to such tendency in many clinical aspects. Different cases of autisms are found in some special education schools and institutions. In the cases for those special educational support and sometimes medical intervention is required to diagnose as having autistic tendencies, some children can be judged as autistic at first sight, some children who can communicate visually are diagnosed as autistic, and some children have adequate language capacity and expression.

We admit to state that an epidemiological study is the base upon which to construct a regional treatment, early

intervention, and special education project. Once the importance of such studies is recognized, the importance of intervention is emphasized, not according to the specific diagnosis or name of the disease, but according to the individual patient's condition and special needs. This applies to clinical fields dealing with intervention, treatment and educational support.