

My Early Experiences in Establishing Neonatal Screening and the Reason for Regional Meetings of the International Society for Neonatal Screening

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Abstract

When I started the neonatal screening in Japan, I could not obtain the necessary information for establishing the national screening system in my own country. Thus, around 1970, I visited H. Bickel, R. Guthrie and several other experts in the field of the neonatal screening in USA and Europe. Through their help, I could learn: (i) the philosophy of the world of screening, (ii) the way to improve the basic techniques in this field and (iii) the way to improve the level of screening. On the other hand, I realised that in some countries, people received imprecise information from non-authoritative sources. I also realised that it was difficult for people in developing countries to meet experts of other countries. Therefore, when I was appointed the first president of International Society of Neonatal Screening (ISNS), I proposed to have the regional meetings held in many areas. In this report, I explained how we were asked to establish the national screening system in Japan through the support of experts around the world. I hope that people will understand the reason why I proposed that regional meetings of the ISNS be held in various locations.

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In 1966, I had started a regional neonatal screening system for Tokyo based on the Guthrie method. By the end of 1970, several laboratories in Japan had also started screening in their areas. As the head of the Division for Mental Retardation at the National Institute of Mental Health in Japan, I believed that we had to establish a screening system which would give all the babies in Japan the privilege to be screened. This belief in the need for a national screening system in Japan was shared by many colleagues and it encouraged me to take steps to establish such a system.¹

However, I soon realised the inadequacy of my knowledge to pioneer a national screening system. So, I tried to meet world renowned screening professionals and learn from them the way to establish a nationwide screening system. In 1971 summer, I received a special travel grant from my government to visit other screening centres. This was the first and a really important opportunity for me as I could learn all about neonatal screening.

The main purpose of this trip was to visit Professor Horst Bickel (Fig. 1), Director of the Medical School in Heidelberg, West Germany. Over a period of 3 hours, he

very kindly shared with me his expertise and experience and allowed me to discuss issues related to neonatal screening. He told me that he already knew some screening techniques for the early detection of phenylketonuria (PKU) patients but when he had learned the Guthrie method, he recognised that he should cooperate with Guthrie to further develop this method all around the world. Also, he emphasised that neonatal screening should be recognised as a public health initiative and hence should be funded by the government. He also shared his lack of success in establishing a national neonatal screening programme for inborn errors of metabolism in West Germany in spite of many representations. Prof Bickel however encouraged me to establish a national programme in Japan and promised personal and international support. Subsequently, he also responded to us several times when we needed his help.

In the beginning of 1972, I was invited to present a paper reporting on the status of screening in Japan at the “International Symposium on Laboratory Screening Techniques for Inborn errors of Metabolism (IEM) in Newborn and Selected High-risk Infants” that was held in Warsaw. My participation at this meeting of a select group

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Fig. 1. Horst and Barbara Bickel, 1975.



Fig. 2. R. Guthrie in a training Course in Tokyo, 1976.

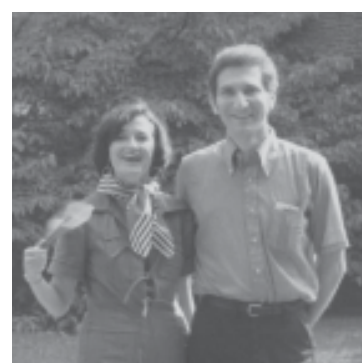


Fig. 3. Harvey and Barbara Levy in USA, 1974.

was facilitated by Horst Bickel and Robert Guthrie. At this meeting, I was able to meet and establish contacts with the screening fraternity of the USA. These included Robert Guthrie from Buffalo, Harvey Levy from Boston, Kenneth Shaw from Los Angeles, Catlin Brandon from Oregon, William Murphy, an associate of Robert Guthrie and Rudolf Hormuth. I also was fortunate to meet with Ronald Gitzelman from Switzerland, Otto Thalhammer from Austria, Barbara Cabalska from Poland, Bohunka Blehova from Czechoslovakia, Nina Carson from Ireland, and Arthur Veale from New Zealand. I also had the opportunities to ask many questions and receive a variety of perspectives on some difficult issues.

From personal conversations with Guthrie (Fig. 2), I learned many more important things about neonatal screening. Therefore, immediately after my return to Japan, I had a discussion with several Japanese pioneers of neonatal screening. We invited Guthrie to Japan in March 1974 as an official guest. His many lectures and extended discussions remained of great importance to many of us.

In the September 1974, I visited Robert Guthrie and 4 laboratories in Boston, Oregon and Los Angeles, where there were important co-workers of Guthrie who were developing a multiple screening system. I spent several days in Buffalo to learn the details of the Guthrie method. During my stay in his laboratory, I learned about the making of good bacterial inhibition assay (BIA) plates and ensuring an accurate recognition of abnormal samples. I realised that my knowledge of the Guthrie method was not correct and, I decided in my mind that before we started the national screening system with its many regional screening laboratories, we should invite Guthrie to come again and teach the technicians, the precise preparation and accurate reading of the BIA plates. The government thus managed to organise a final training course for the screening laboratories in all areas of Japan at the end of December 1976, just 1 year before the initiation of the nationwide screening system.¹ Also, I had learnt the necessity of

quality control for the Guthrie method as Robert Guthrie had already started to distribute the standards made in his laboratory. I thought that this was the most important step of quality control for the Guthrie method.

In Boston, I visited Harvey Levy (Fig. 3), the expert on the various kinds of IEMs who taught me the crucial technical points and the necessity of early treatment of the different types of IEMs. After I met him, I was convinced to start screening in Japan for as many diseases as we could.

In Los Angeles, I met Richard Koch, Professor of Paediatrics and a specialist in the treatment of PKU and other IEMs and the consultant for the screening system in California. I have also learnt from both Koch and Kenneth Shaw, a chemist who started the Guthrie method in California. At that time, there were many private laboratories for PKU screening in California and they worried about the poor quality of PKU screening in some laboratories. Hence, we discussed about the way to control the quality of the screening process. After this discussion, I considered the establishment of a national quality control system. Also, in LA, I had a chance to meet Robert Phillips, who in the beginning of 1970 developed the Punch index machine when Guthrie and his co-workers decided to start the multiple screening system. Most laboratories in this group used 4 BIA plates for one sample. This machine was very useful for preventing the mistake of not placing the disc in the exact position of each BIA plate. I stayed at his laboratories, learned how to use it and finally concluded that the machine was very reliable. When we started nationwide screening, many laboratories used this kind of machine.

Based on the support from these people, Japan could start the national screening system in 1977. Our federal government supported all regional governments so that all screening laboratories could use the standard prescribed programme. All screening laboratories were covered by the quality control system. Many people, including myself, who were consultants to the screening laboratories were

well trained. After we had started the nationwide screening, the Japanese Society For Mass-Screening was established with about 500 members initially.¹

Had we not received any support from many of the experts mentioned in this article, we might have started Neonatal Screening in Japan with many weak technical points or incorrect procedures based on inaccurate information. I am very grateful to Horst Bickel, Robert Guthrie, Harvey Levy and many others who had helped us tremendously. Based on their support, we were able to start the nationwide screening well without making any serious mistakes.²⁻⁴

I think it will be very important for people who work in the screening laboratories to participate in international meetings, meet experts from different countries and learn from them how to carry out their duties and responsibilities well. Besides Japan, many people in other countries working in screening laboratories also faced difficulty in attending such meetings held overseas.

Therefore, when I was asked to serve as the president of the International Society for Neonatal Screening (ISNS), my first idea was: “Establishment of regional meeting in various parts of the world, so that many people will be able to meet excellent people in the field of screening.” When we had the first business meeting of the ISNS in Brazil in November 1988 I proposed this idea. This was agreed by all and hence we decided to organise regional meetings.^{1,2} In the Asia-Pacific region, the first regional meeting was

held in Sapporo, Japan in June 1993. Since then, 4 more such meetings have been conducted in the Asia-Pacific region, namely in Hong Kong, Thailand, Philippines and China. Besides these 4 countries, the regional meetings were also held in Europe and South America.⁴

I sincerely hope that many neonatal healthcare co-workers in the Asia-Pacific region will have the chance to attend excellent international lectures conducted by the experts. Should they have the opportunities to interact with these overseas experts, it will be definitely a beneficial experience for them.

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