

# Current Situation of Infants with Trisomy 18 Syndrome in Japan

## Summary of Research by the Trisomy 18 Support Group

### What is trisomy 18 syndrome?

Trisomy 18 syndrome is a congenital disorder characterized by growth and developmental delays and various complications due to a chromosomal aberration: chromosome 18 appears three times (trisomy) rather than twice in cells of the body. It was reported for the first time in the 1960s by Dr John H. Edwards. It is a frequent syndrome of chromosomal aberration, affecting one out of 3,000 - 8,000 newborns. You can see an explanation - how does trisomy occur - on next page of this leaflet.

### Issues related to trisomy 18 syndrome.

Although much research has described Trisomy 18 syndrome related to its complications and the rarity of long survival, information connected to family care or clinical practice has been very poor in Japan. Overlooked issues included, "How is it to live at home with infants with trisomy 18?" or "What kind of treatment do they need?" The treatment policy for trisomy 18 syndrome was also in confusion. For example, one infant might be treated intensively in a hospital, and another might be restricted to ordinary treatments from the time the infant had a confirmed diagnosis of trisomy 18. Surely, this wide variation in attitudes towards the infant patients was confusing and painful for the parents.

### Research in Japan about situation of trisomy 18 patients and their families.

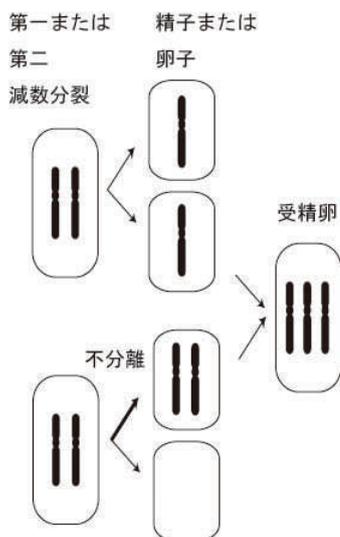
The Japanese Society for Trisomy 18 thinks that research about the actual situation facing patients and their families is very important for establish good practices for medical treatments, nursing and care. We surveyed the members of the Japanese Society for Trisomy 18 in 2003 to clarify these areas: 1) birth, 2) symptoms, 3) treatments, 4) life at home or at the hospital, 5) the passing away of patients, 6) parent's feelings. This leaflet is a summary of the research.



*This clenched hand is a characteristic feature of an infant with Trisomy 18.*

# Summary of Research by the Trisomy 18 Support Group

*Trisomy is a phenomenon that any male or female may encounter at the beginning of life.*



## How does trisomy occur?

Chromosomes are bundles of DNA, containing plans for the human body, i.e., genes. In each human cell, there are usually 46 chromosomes. These come in pairs, of which number 1 to 22, the autosomal chromosomes, are common between men and women. In addition, there is a pair of sex chromosomes, XY for men and XX for women. When trisomy 18 occurs, a cell has three copies of chromosome 18 instead of a pair.

Most infants with trisomy 18 have three copies of the 18th chromosome in all cells of the body (full trisomy), but some infants with trisomy 18 have double copies of the chromosome in some cells and triple copies in others (mosaicism). Chromosomes are equally divided when sperm cells or egg cells are made. (That is, an egg cell or a sperm cell

usually contains 23 chromosomes: the 1st to 22nd autosomes plus an X or Y sex chromosome, for a woman or a man, respectively.) In this process, if chromosome 18 does not divide equally (nondisjunction), it will result in an additional 18th chromosome, so the sperm or egg cell will have 24 chromosomes. If this cell, then, contributes to conception, the fertilized egg will have 47 chromosomes as a result of the three copies of the 18th chromosome.

It is a fact that there are many cells with chromosomal aberrations among the sperm or egg cells of every adult. Thus, there is always a chance with a pregnancy that the fertilized egg will have a chromosomal aberration.

## Research methods

The subjects of the study were members of the Japanese Society for Trisomy 18, who participated with free will and in anonymity. The response rate was 70%; out of 125 questionnaires, 88 families answered. Twenty-nine of the respondents were the guardians of a boy and 59 were the guardians of a girl. Seven of responses came from families with a stillbirth. This research was performed from July 2003 to February 2004. The

results were presented by supporting members of Japanese Society for Trisomy 18 at several academic meetings in 2004, i.e., those of the Japan Society of Perinatal and Neonatal Medicine, the Japan Society for Premature and Newborn Medicine, and the Japanese Society of Genetic Nursing.

## Situation of pregnancies and deliveries

Of the 81 liveborn infants in the study, the average time of gestation was 38 weeks, 5 days and the average birth weight was 1,868g. 42% of the births were transvaginal and 58% were by cesarean section. 68% of the parents had been given the opportunity by a doctor to discuss what to do when their baby developed bad symptoms or experienced acute deterioration, and 24% parents had not been given that opportunity. (8% of the respondents gave no answer to this question.)

Regarding the contents of explanations, according to 39% of the respondents, doctors “set a limit to treatment, for example, no aggressive resuscitation,” and, according to 32%, they offered to “treat as much as we possibly can.” (29% of the respondents gave no answer to this question.) 53% of the parents felt that their opinion had been respected

adequately during the determination of the treatment plan; 12% of the parents did not feel respected; and 1% could not judge what they felt. (34% gave no answer.)

Not a few parents had experienced that medical staff related to them that the uniform policy for a baby with trisomy 18 was to not treat the child actively, and, moreover, failed to give a sufficient explanation about babies with trisomy 18. Some parents felt a sense of discomfort because doctor’s attitudes towards babies with trisomy 18 differed from one hospital to another. On the other hand, some parent stated that the medical staff respected the parent’s opinion enough and that they made a treatment plan with due consideration of the infant’s individual disease condition.

# Summary of Research by the Trisomy 18 Support Group

*Infants with Trisomy 18 grow and develop slowly.*

## Trisomy 18 leads to what kind of medical complications?

Various complications are possible with trisomy 18, in particular heart disease. In the results of this study, complications occurred in the following percentages (of all 88 patients).

Heart: ventricular septal defect, 81%; patent ductus arteriosus, 44%; atrial septal defect, 35%; aortic coarctation, 18%.

Digestive system: esophageal atresia, 15%; omphaloceles, 9%; anal atresia, 6%.

Urinary system: hydronephrosis, 8%; horseshoe kidney, 7%.

Nervous system: cerebellar hypoplasia, 44%; seizure, 20%; apnea attack, 42%.

Others: ear deformities, 52%; cleft lip, 8%; cleft palate, 7%; syndactyly, 25%.

## What kinds of treatments are given for children with trisomy 18?

Respiratory support, including intubation and nasal continuous positive airway pressure (CPAP)/directional positive airway pressure (DPAP), was performed for 50% of the infants (the percentage of intubation among them was 76%), while 49% of infant were not given or did not need respiratory support. (No answer, 1%). Among the infants who underwent ventilation, 42% were respirator dependent until they died. On the other hand, 45% of the infants were able to be independent from ventilation for a while or forever. (Unknown, 13%). Not a few infants were able to leave from artificial ventilation, and not a few infants had no need respiratory support at birth, among the liveborn infants.

Fourteen patients experienced operations, including those for esophageal atresia in 3 patients, for ompha-

locele in 3 patients, and for heart disease in 4 patients (most of which were pulmonary artery banding).

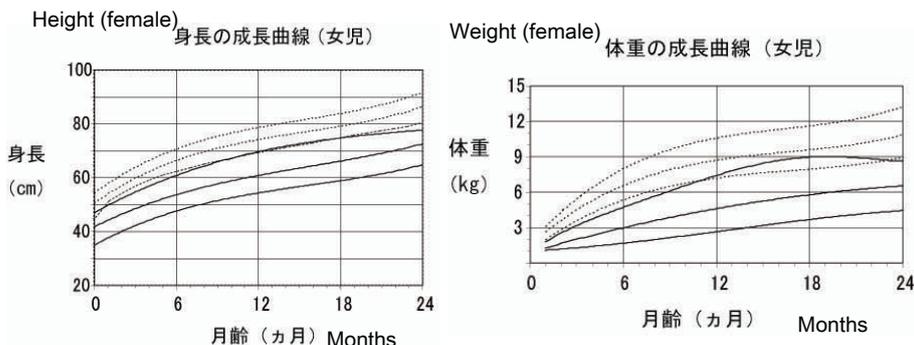
BCG vaccine was given to 14 infants; DPT (Diphtheria, Pertussis, Tetanus) vaccine, 17 infants; oral poliovirus vaccine, 12 infants; measles vaccine, 13 infants; rubella vaccine, 13 infants; mumps vaccine, 5 infants; varicella vaccine, 7 infants; influenza vaccine, 12 infants; anti-RS (respiratory syncytial) virus monoclonal antibody globulin, 2 infants. As for vaccination side effects, one infant experienced a lump and feeling of heat from a DPT vaccine. A fever was detected in four infants after vaccination: 2 from measles, 1 from rubella, and 1 from varicella.

## How do infants with trisomy 18 grow and develop?

A growth chart of Trisomy 18 could be made for girls due to the size of sample. Dotted lines are curves of the height/weight of the unaffected girls, and solid lines are that of trisomy 18 (3, 50, 97 percentile, respectively).

Development stages and their achieved age for the infants were as follows: Head control, 8 infants (average achievement point 1 year and 5 months); Roll over, 13 infants (1 year and 4 months); Crawl, 2 infants (4 years and 2 months); Walk alone, 3 infants (3 years and 3 months). As for responses: a responsive smile, 18 infants (average achievement at 7 months); babbling, 9 infants (average 1 year 6 months); understanding words, 3 infants (average 5 years old); talking, 1 infant (average 4 years old). As you can see they grow and develop slowly.

Growth Curve



*These infants have some constitutional predispositions and complications, but they have their own uniqueness and charms.*



With Permission  
from the Parents

### What is the prognosis for a child with trisomy 18?

As for the prognoses of the 78 infants in the study who were able to be followed over their lifetime, the survival rate was: one day, 94%; one week, 90%; one month, 80%; one year, 31%, 3 years, 15%; 5 years, 9%; and 10 years, 8% (5 patients). The causes of death were: heart failure or cardiac disorder, 53%; infectious disease, 17%; respiratory failure, 6%; hepatoblastoma, 5%; and others. Some parents (6%) were not informed enough about the cause of death.

It is important to note that population-based data in Western countries report a survival rate for 1 year

olds that is between 0 - 10%. The discrepancy with this report's findings may depend on the fact that many members of the Japanese Society for Trisomy 18, and in particular many of those who responded to the questionnaires, are families of long-term survivors, and it may also depend on the fact that not a few pediatricians in Japan sufficiently treat infants with trisomy 18.

### What are the babies' charming points? What makes the babies happy?

In response to the question, "Please tell us your child's charming points" some of the parents answers were: "all", "large eyes", "full cheeks", and "smile." In addition, polydactyly and clenched fingers, which are characteristics of trisomy 18, were remarked as charming points by some parents.

"When do you find your baby happy?" Parents gave various answers: "hugging", "bathing", "playing with the family", "feeding/meals", "going out", and "kangaroo-care".

"When do you feel happy to be with your baby

and your baby's birth?" Some answers were: "the baby's birth itself", "being beside the baby", "during smiles", "at every moment", and "family pleasures of home life".

Healthcare providers had a tendency to think of treatment for complications of trisomy 18 as a kind of invasive action, with comments such as "an infant with trisomy 18 will suffer from life lengthening, and this will burden their family." But we now know that greatness of the infant's being or the family's love can overcome physical impairment or a severe prognosis.

### What is it like to care for a child with trisomy 18?

The percentage of infants who were hospitalized and never discharged was 47%, and the percentage of infants who were discharged and able to stay at home was 50%. (No answer, 2%). Parents felt burdens regarding their lives with their infants, such as "difficulty in feeding", "crying", "apnea", "insomnia/day and night reversal", "dyspnea", and "convulsions".

It seemed that tube feeding or hospital visits took much time and were burdens for mothers. On the other hand, some parents answered that they did not feel them to be burdens. Twenty infants were able to have milk or meals fully via the oral route, 16 infants were able partially via the oral route, and 36 infants fully depended on tube feeding. (No answer, 12).

Physical disability certificates for social services were issued for 21 infants. Mental disability cer-

tificates for social services were issued for 12 infants. Among them, 7 infants got both.

A few guardians had used a support system for home care; short stay, 3 families used a system of short stays; 2 families made use of nurse visits. Coordination between healthcare providers and social services was not established enough. Some of the guardians comments were: "We cannot get knowledge about welfare services unless we actively search for information"; "Procedures for receiving services from the welfare system were always fiddly"; "There are many requirements to meet for social services"; "Social service staff did not understand well the medical conditions of our child."

# Summary of Research by the Trisomy 18 Support Group

## How do parents first learn of their child's disease?

As for the time that parent(s) were told of the possibility of trisomy 18 or a chromosomal aberration: among the 88 questionnaires, prenatal stage, 28%; at birth, 27%; in the days after birth, 40%; no answer, 4%. The conditions of this first explanation were as follows: both parents in attendance, 41%; father only, 34%; mother only, 10%; (others and not described, 15%). Only 40% people were given the explanation in a place conducive to maintaining privacy.

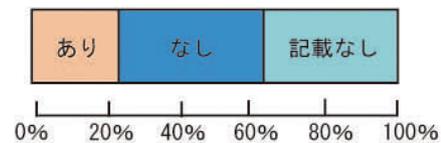
At the time, 42% people were given opinions such as “the baby will be alive for only a short time,” “we will not resuscitate the baby aggressively,” etc. Twenty two percent replied that they received mental support from the medical staff at that time, and 41% stated that there was no such help. (No answer, 37%).

There were some situations in which only the father or mother was told about the baby's condition far from a private place. This situation, in which one parent was not informed of his or her own baby's

correct condition, made the parent feel alienation. Besides, the parent who was told alone felt unendurably burdened by the fact. When a doctor did not explain anything of aftertime during pregnancy, a mother felt completely at a loss with a comment with “I had a hard time. Because I didn't know how the pregnancy progressed and how the delivery came with my situation, there was nothing dependable for me.”

Another problem was healthcare providers showing a tone of resignation in their language and behavior regarding a baby with trisomy 18 as if the baby will die soon.

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## What are the feelings of the parents of stillborn babies?

Seven answers were from parents with a stillborn infant. All of them had a prenatal diagnosis. It brought pain to the parents that the baby's life, as close as it was to being born, might have been abandoned by the healthcare providers due to trisomy 18, and that healthcare providers might have changed their relationship with the family to avoid them.

In addition, some mothers felt unbearable loneliness after the stillbirth, because the healthcare providers had her stay in the same hos-

pital ward where the sound of babies crying reached her ears. The parents felt emotional support through feelings of compassion or of tender care for baby, opportunities prepared by healthcare provider to spend time with the baby, and peer information from a peer support-group.

## Necessity of emotional support

Everyone imagines themselves to give birth to a healthy baby during pregnancy. It is an unexpected and shocking event to be informed that one's baby has a disease with an unfamiliar name and a poor survival rate at birth or after birth. Since most people can hardly image their lives with a handicapped child, they get more and more anxious. The situation is as if you one suddenly lost a course and wandered off into a trackless place.

The first step of support for these families by healthcare providers is to treat the baby in accordance with the infant patient's disease condition, to inform the parents about the disease fairly and softly and to respect the parents' opinion when considering the treatment plan.

Counselors can help families to talk about their worries. The numbers of counselors were not enough around hospital wards but the number is increasing recently. Also, the peer counseling of the Society for Trisomy 18 may be a beneficial way that parents can share their emotions with people who have experienced in the same situation.

## Acknowledgements

The data shown on this leaflet are only a fraction of the investigation. Each person's experience and complex emotions can never adequately expressed as a numerical value. In this questionnaire survey, members of the Japanese Society for Trisomy 18, by filling out the many questions carefully, would

have tracing painful thoughts in recalling their difficult experiences. We are grateful to all the people in the Japanese Society for Trisomy 18 who overcome painful experiences answer the questionnaire, those who could not answer for various reasons, and the coordinating people.



## **The Trisomy 18 Support Group**

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Please refer to the following website.  
<http://18trisomy.com/>

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Where to Contact  
E-mail: [questions@18trisomy.com](mailto:questions@18trisomy.com)

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### Our Group Activities

Family Members

Supporting Members: Medical Staffs and Welfare Workers for Children

Issue of Pamphlets

Distribution of Pamphlets (to Hospitals etc.)

Issue of Newsletters (Three Times per Year)

Support for Members that Do not Have Internet Access (by Mail or Fax)

Provision and Share of Information, Circular of Notes

Communication on Homepage

Bulletin Board for Family Members Only

Peer Counseling

Mailing List

Those who have email addresses can register for this mailing list.

Provision of Real-time Information

Issue of "Guu Guu Handbook"

Holding of Public Seminars (Once a Year)

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