Attitudes towards Down's syndrome: follow up of a cohort of 280 cases

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Abstract

A follow up study of a cohort of 280 Down's syndrome (DS) fetuses was initiated in order to estimate the percentage of terminations of pregnancy, the prevalence at birth, the survival of DS children, the attitude of the parents at birth, and the medical and surgical care provided. The present study is a preliminary analysis of the data collected up to the age of 1 year. It shows that 43% of the 280 DS fetuses had died by the end of the first year of life, owing to termination of pregnancy (27%), late spontaneous abortion or stillbirth (4%), or death during the first year of life (12%). Among the 33 children who died, 12 had a common atrioventricular canal, six had another major malformation, three died from infection, one from respiratory distress, two were cot deaths, and one was an infanticide, but eight deaths were unexplained, occurring in children with no known malformation or disease. Among the 185 children still alive after 2 days, 23 (12%) were available for adoption, their mothers having elected to remain anonymous. These results show that in some instances parents or professionals feel justified on one hand in not providing DS children with the necessary care and on the other hand to transfer their responsibilities to the public health system. The influence of prenatal diagnosis of chromosome disorders as a determining factor of the social acceptance of DS is still questionable.

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the issue of what becomes of DS children, a follow up study of a cohort of 280 DS fetuses was initiated in order to find out the survival rate of DS children, acceptance at birth by their parents, and medical and surgical care. The present study is an analysis of the data collected up to the age of 1 year.

Materials and methods

DEFINITION OF THE COHORT AND STUDY POPULATION

The cohort consisted of all DS fetuses detected pre- and postnatally between 1 January 1984 and 31 December 1990. The study population consisted of a total of 164 506 livebirths, stillbirths, and induced abortions surveyed during the same period of time by the Birth Defects Registry of the Marseille district (Eurocat 22). In the study area, stillbirths were systematically examined using a postmortem protocol¹¹ including karyotyping. When karyotyping was unsuccessful for technical reasons, mainly maceration, the stillbirths with typical clinical features of DS without any doubt were classified as DS.

DATA COLLECTION AND ANALYSIS

Systematic notifications to the Registry include the child's birth date, gestational age, type of birth, place of birth, clinical data, prenatal procedures performed, results of karyotype, when done, parents' birth date, and place of residence. A complementary questionnaire was set up to collect information on the follow up of the DS children and was sent to all physicians who had notified a case or were likely to be in charge of them.

Data about placement for adoption were obtained from the Department of Health and Social Welfare and matched with the data from the Registry through the date of birth of the child. The data corresponding to the total number of births in the area were obtained from the National Institute of Health and Economic Studies (special analysis).

Results

PREGNANCY OUTCOME AND SURVIVAL

In this cohort of 280 DS fetuses, 43% had died by the end of the first year of life, owing to termination of pregnancy (27%), late spontaneous abortion or stillbirth (4%), or death during the first year of life (12%).

As can be seen in table 1, abortions were performed on 76/280 fetuses (27%), which corresponds to all the cases prenatally diag-

The cultural approach to Down's syndrome (DS) has changed over time, since the discovery of its chromosomal origin.¹ At present, concepts such as acceptance of different people, fight against discrimination, and complete integration are shared by most people. At the same time, industrialised countries have developed policies on prenatal screening for DS based on maternal age limits, previous family history, prenatal sonographic signs, and serological markers, which have a high level of social acceptability.²⁻⁶ The possible effects of the availability of prenatal screening on the social acceptance of disabled persons have been questioned,^{8,9} but never evaluated, either in the past or since the availability of prenatal screening. A survey conducted in Paris showed that 10% of all the children available for adoption were DS children.¹⁰ To start exploring

Table 1 Distribution of pregnancy outcome among Down's syndrome cases over the years 1984–1990 (Bouches du Rhône Birth Defects Registry)

Year of pregnancy outcome	Terminations of pregnancy		Spontaneous abortions* and stillbirths		Livebirths		Total	Prevalence (per 1000)
	No	%	No	%	No	%		
1984	6	22	1	4	20	74	27	1.17
1985	6	18	1	3	26	79	33	1.41
1986	4	10	1	3	34	87	39	1.65
1987	13	30	3	7	27	63	43	1.84
1988	11	29	2	5	25	66	38	1.59
1989	13	27	3	6	33	67	49	2.04
1990	23	45	1	2	27	53	51	2.18
Total	76	27	12	4	192	69	280	1.70

* From the 20th week of gestational age.

Table 2 Down's syndrome fetuses: distribution of pregnancy outcome depending on maternal age, Bouches du Rhône Birth Defects Registry (1984–1990)

Maternal age	Terminations of pregnancy		Spontaneous abortions and stillbirths		Livebirths		Total	Prevalence (per 1000 births)
	No	%	No	%	No	%		
<35	11	8	7	5	114	87	132	0.91
35–37	18	32	1	2	37	66	56	4.64
38–39	16	48	0	0	17	52	33	8.02
≥ 40	31	61	3	6	17	33	51	16.25

nosed, as all the women elected for abortion. Despite an increase over time of the proportion of women having their fetus karyotyped, the total number of DS livebirths was higher in 1990 than in 1984 because of the increase in the prevalence at birth. The prevalence increased as a result of the increase in the average maternal age at birth in the region (27.5 in 1984 and 28.7 in 1990). The proportion of prenatal diagnoses increased with maternal age: under the age of 35, 8% of DS were screened prenatally through ultrasound detection of abnormalities. The majority (61%) of DS were diagnosed prenatally in women over 40 (table 2).

Among the 192 children alive at birth, 17% died during the first year, nearly half of them during the first month. The causes of death, either direct or indirect, are listed in table 3. Among the 33 children who died, 12 had a common atrioventricular canal, six had another major malformation, three died from infection, and one from respiratory distress after inhalation, but eight deaths were unexplained, occurring in children with no known malformation or disease, two were cot deaths, and one was an infanticide. The survival rate at one year was 79% for the children with a common atrioventricular canal surgically treated and 43% for those who were treated medically (log-rank test, $p=0.08$).

Table 3 Causes of death among the 33 DS children who died during their first year of life

Age at death	Cause (Direct or indirect)	No
First week		
1st day	Ascites	1
	Oesophageal atresia	1
	Unexplained	2
2 days	Infanticide	1
5 days	Unexplained	2
6 days	Non-specific infection	1
2nd week–1 month		
15 days	Unexplained	1
18 days	Lung infection (after meconium inhalation)	1
1 month	Atrioventricular canal (without surgery)	1
	Sudden infant death syndrome	1
	Respiratory distress (inhalation of food)	1
	Vertebral malformation/triplets	1
	Exomphalos	1
2–3 months		
2 months	Atrioventricular canal (without surgery)	1
	Unexplained	2
	Major ascites	1
	Duodenal stenosis	1
3 months	Atrioventricular canal (with surgery)	1
4–6 months		
4 months	Sudden infant death syndrome	1
4 months	Atrioventricular canal (without surgery)	3
5 months	Unexplained	1
	Atrioventricular canal (with surgery)	1
6 months–1 year		
6 months	Atrioventricular canal (without surgery)	2
7 months	Meningitis	1
	Atrioventricular canal (with surgery)	1
9 months	Atrioventricular canal (without surgery)	1
11 months	Atrioventricular canal (without surgery)	1

DS CHILDREN AVAILABLE FOR ADOPTION AT BIRTH

In France, there is the possibility for parents who choose not to raise their child to make the baby free for adoption at delivery time or later on. If the choice is made at delivery, the mother may deliver anonymously so that no link can possibly be established later between her and the baby. The question of whether this procedure was used by DS parents was explored. Among the 185 children still alive after 2 days and not lost to follow up, 23 (12%) were available for adoption, their mothers having elected to remain anonymous (table 4).

Table 4 Number of DS children available for adoption at birth, per year of birth

	DS children free for adoption		Total of DS still alive after 48 hours
	No	%	
1984	0	0	18
1985	1	4	26
1986	5	16	31
1987	3	12	26
1988	2	8	25
1989	7	21	33
1990	5	19	26
Total	23	12	185

Discussion

This study confirms the trends already observed in different regions of France^{12,13} and in other European countries¹⁴ as regards the increase in the prevalence of DS conceptions resulting from the shift in the maternal age distribution and the increase in the uptake rate of prenatal diagnosis. The fact that the uptake of amniocentesis has been increasing regularly since the early 1980s shows that a large proportion of the population wish to benefit from prenatal testing. The extent of the use of fetal karyotyping is not sufficient to decrease the prevalence at birth, leading to a contradictory situation where more and more couples have to face the birth of a DS child at a time when most people think it is a screenable condition.

The one year survival rate observed in this study (81%) is comparable to the results obtained in recent studies,^{15,16} as well as the fact that cardiac defects are the most common causes of death. In our study, a larger than expected number of unexplained deaths was observed including one infanticide.¹⁷ Our data set is not detailed enough to estimate how many of all the deaths were avoidable with appropriate medical or surgical care, but the question needs to be addressed.

As regards the abandonment at birth of 12% of the DS children in this cohort, it is necessary to interpret this in the light of the peculiarities of French legislation, established in 1941 to protect illegitimate children whose mothers could not care for them, as opposed to the British law (Adoption Act 1976), for instance, where agreement to adoption is ineffective if given by the mother less than six weeks after the child's birth. In addition, in the French legislation, there is the possibility to protect the identity of the parents; the child is then declared as born to unknown parents. For this reason, considering adoption at birth by a willing family is presented to the parents by the medical teams at the birth as an option.

As no previous studies have been published on the proportion of DS children raised in their family, placed in institutions, or given up for adoption by their parents, it is not possible to say whether this 12% is higher than it was in the past. However, it is necessary to look at whether similar attitudes are observed in other regions and countries, what are the variables linked to the type of parental attitude, such as age, parity, educational level, and religion, whether DS children are adopted, and who are the adoptive families.

The development of prenatal diagnosis for chromosomal disorders may have influenced the social acceptance of Down's syndrome. Parents and professionals may feel more justified than in the past in not providing DS children with the necessary care.^{18,19} For this reason also, parents and professionals may feel more justified in transferring their responsibilities to the public health system.

Besides the possible effect of prenatal diagnosis, the acceptability of DS births is likely to be linked to many factors, such as the decrease in the size of sibships, the geographical spread of families, the increase in the number of di-

vorces, the insufficient number and quality of the services available for infants and adults, and the economic crisis. Another important variable is likely to be the way the parents were "first told". Published studies²⁰⁻²² have extensively described parents' and professionals' reactions to the birth of a DS child. Maternity hospital teams must take enough time to help the parents to undergo the initial trauma.

As public health screening policies are developed to prevent the birth of children with severe disabilities, the authorities should counterbalance these policies by developing appropriate services for the care and social integration of disabled people, which meet the needs of both patients and their families.

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