CHROMOSOMAL ABERRATIONS DETECTED AT BIRTH

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Malformations due to chromosomal aberrations constitute a major part of fetal demise in the first and second trimesters. Whoever a certain number of newborns will be delivered alive and the most common detected abnormality is Trisomy 21 in all studies. In our practice karyotype was performed upon strong clinical indication. The type of anomaly diagnosed concerned the most common ones using the G-banding technique and not small deletions and minor abnormalities described with advanced technique. The young age of mothers in this series is an important factor for the interpretation of these results.

RESULTS

Trisomy 21 was the most common diagnosed anomaly (Table I) with a surprising male predominance.

Table I

MALFORMATION	M	F	TX	SB	ND	M/1000D	M/N DEL
TRISOMY 21	**30**	**9**	**39**	**1**	**3**	**1.15**	**871**
-Duodenal atresia	1	2	3	0	0		
-Imperforated anus	1	0	1	0	0		
-Cardiac malformation(not VSD)	1	4	5	0	0		
-VSD	8	2	10	0	0		
-TEF	1	0	1	0	0		
-Bilateral Hydronephrosis	1	0	1	0	0		
-Diaphragmatic hernia & omphalocele	1	0	1	0	0		
TRISOMY 13	**3**	**4**	**7**	**0**	**7**	**0.21**	**4852**
TRISOMY 18	**2**	**5**	**7**	**0**	**7**	**0.21**	**4852**
TURNER	**0**	**6**	**6**	**5**	**0**	**0.18**	**5661**
NOONAN	**3**	**0**	**3**	**0**	**2**	**0.09**	**11323**
TRANSLOCATION P4-7	**0**	**1**	**1**	**0**	**1**		
44 Del 18(ptr)HPSEN	**0**	**1**	**1**	**0**	**1**		
T chromosomal aberrations	**38**	**26**	**64**	**6**	**21**	**1.9**	**531**
T malformed NB	**449**	**363**	**812**	**156**	**306**	**23.90**	**41.8**

The prevalence of trisomy 21 in this series was 1 in 871 total births and 1 in 875 live births. This figure is lower than reported in Libya as 1 in 516 live births [1] or 1 in 554 live births in Saudi Arabia [2]. It is to be mentioned that during the study period there was no real possibility in this population to develop ante natal diagnosis at large scale in this population and reduce the rate of trisomy 21 by abortion. The influence of antenatal diagnosis on the incidence of trisomy 21 has been reported in many countries of different cultural backgrounds [3, 4, 5].

Male predominance in trisomy 21 was noticed also in a Russian study [6] and in New York [7].

In a series [8] of comparative number, 33 562 newborns (live and stillborn), chromosome aberrations in malformed newborns was evaluated (Spanish Collaborative Study of Congenital Malformations) from March 1982 -September 1996. Of the 1,409 (4.1%) malformed infants identified, the frequency at birth of chromosome abnormalities was 5.4% of malformed newborns (7.9% in our series). There were 59 (1.76/1000) infants with Down's syndrome, 6 (0.18 /1000) with Trisomy 18, 3 with Turner's syndrome, 2 with Trisomy 13 and 4 with other anomalies. The prevalence of Down's syndrome was higher in the Spanish population compared to this series but other abnormalities were lower. The age distribution in the 2 series can be a determinant factor in the difference in results. The prevalence of Trisomy 13 was relatively high. Further studies on larger populations through time might confirm or infirm these initial findings.

The incidence of congenital abnormalities due to chromosomal abnormalities showed also ethnic variations. In a cytogenetic study of Malaysian [9] neonates with congenital abnormalities it was found to be 1:838 live births. Frequency of newborns having abnormal chromosomes, majority with Trisomy 21 was 0.14% for Malaysia, 0.12% for Chinese and 0.06% for Indians. Ethnic variation could be seen in nearly all malformations but the methodology of case ascertainment is variable and precautions must be taken before unfounded explanations.

The availability of genetic counseling services in Palestine will permit us to have a more clear idea about the real incidence of chromosomal aberrations. At the same time the development of antenatal diagnosis and the public acceptance of abortion for cases diagnosed in the early antenatal period will produce the same effect on the reduction of the number of newborns with chromosomal anomalies. This effect has already been noticed in Tunisia, a country with common cultural background as Palestine.

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