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Genetic Conditions Among Patients Receiving Genetic Services in Middle Tennessee

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Abstract

We reviewed genetics charts of 2235 patients seen from 1985 to 1990 at Vanderbilt University Medical Center, Nashville, Tennessee, and summarized the 20 most common reasons for referral (occurring in 1138 of the patients) and the diagnoses or conditions among patients receiving genetic services in one of four clinical settings (prenatal counseling clinics, general genetics clinics, outreach genetics clinics, and ward consultations). The five most common reasons for referral were advanced maternal age (≥ 35 years) (203/1138, or 18% of patients), followed by dysmorphic features/multiple congenital anomalies (MCA) (185/1138; 16%), developmental delay/mental retardation (MR) (168/1138; 15%), Down's syndrome (103/1138; 9%), and abnormal maternal serum α -fetoprotein (MSAFP) (74/1138; 7%). The five most common diagnoses or conditions identified for all genetics patients were advanced maternal age (≥ 35 years) (195/906; 22%), developmental delay/MR (111/906; 12%), dysmorphic features/MCA (107/906; 12%), Down's syndrome (88/906; 10%), and multiple fetal losses (57/906; 6%). Of the 20 most common diagnoses or conditions categorized in 602 of the 906 patients, a multifactorial cause was observed in 25% of those patients; a chromosomal cause was observed in 26% of cases of Down's syndrome, accounting for 55% of the chromosomal disorders; a single gene disorder was observed in 17% of patients; an environmental cause was seen in 4%; and an unknown cause was noted in 28%. We hope this study will help physicians in middle Tennessee and surrounding areas by increasing their awareness of the types and frequencies of genetic diseases so that misdiagnoses and delayed referrals can be avoided.

To estimate the frequency and types of genetic problems that occur in patients referred to a large, full-service medical center, we reviewed the charts of genetics patients over a 5-year period. Analysis of the frequency and types of genetic problems among individuals referred for genetic services at Vanderbilt University should be useful for comparison with other geographic regions of the country as well as with hospitalized patients, birth or population-based health surveys, or registries of individuals at certain ages. Previous studies of hospitalized patients in the United States showed that approximately 33% of children and 7% of adults had a genetic disorder diagnosed.^{1–4} In addition, the frequency of genetic diseases in Canadian children and young adults was reported recently from birth or

population-based health registries and was also ascertained from the general population in the United States.²⁻⁵

Because of the lack of published data in the literature on the frequency and types of genetic problems and the need to inform clinicians about common reasons for referrals and diagnoses seen in general genetics, prenatal counseling, outreach genetics clinics, and ward consultations, we gathered data from our genetics files from 1985 to 1990.

MATERIALS AND METHODS

The data for this study were obtained from a review of patients referred for genetic services to Vanderbilt University Medical Center, Nashville, Tennessee, and patients seen in outreach genetics clinics from 1985 to 1990. Our referral base consists of patients and their families from middle Tennessee, southern Kentucky, and northern Alabama. Approximately 30 000 babies are born each year in this catchment area.

The 20 most common reasons for referral and the 20 most common diagnoses reported in our prenatal counseling clinics, general genetics clinics, outreach genetics clinics, and ward consultations were recorded after reviewing genetics files over this 5-year period.

RESULTS

In our study, 2235 patients came to Vanderbilt University Medical Center for genetic services between 1985 and 1990. The 20 most common reasons for referral and the 20 most common diagnoses for patients seen in the prenatal counseling, general genetics, outreach genetics clinics, and ward consultations as well as the number of patients in each clinical setting are summarized in Tables 1 and 2. The causes of the most common conditions diagnosed are also listed in Table 2. The figure shows the total number of genetics patients seen in each of the four clinical settings for the 5-year period.

DISCUSSION

In our analysis of patients receiving genetics services, it was not surprising that advanced maternal age (> 35 years) was the most common reason for referral, since patients from the prenatal counseling clinics were included and followed up by dysmorphic features/multiple congenital anomalies (MCA). Almost equal numbers of patients referred for this reason were found in ward consultations and the general genetics clinics. From the list of 20 most common reasons for referral in our study, the most common reason for referral to the prenatal counseling clinics was advanced maternal age (181/317; 57%); for referral for ward consultations, MCA (73/216; 34%); for referral to general genetics clinics, developmental delay/mental retardation (MR) (105/472; 22%); and for referral to the outreach genetics clinics, also developmental delay/MR (42/133; 32%). Of our 20 most common diagnoses or conditions identified in patients in all four clinical settings, a multifactorial cause was observed in 148 individuals (25% of the 602 patients categorized); a chromosomal cause was observed in 159 individuals (26%); with Down's syndrome accounting for 88 patients or 55% of patients with chromosomal disorders; a single gene disorder was observed in 101

patients (17%); an environmental cause was observed in 23 patients (4%); and the cause was unknown in 171 patients (28%).

Analysis of trends across the 5-year period showed a large increase in the overall numbers of patients seen. This was illustrated most dramatically in the prenatal counseling clinics, which had 24 patients in 1985 and 130 patients in 1990, representing an increase of 442%. We speculate that this increase, as well as the increase in the number of patients receiving genetic services in the other clinical settings, may be due to a heightened awareness of prenatal screening, particularly in women over the age of 35 years, and the increased recognition of genetic diseases by health care providers and the general population.

In reference to longitudinal studies in the literature, we have found our results to be comparable to those obtained in similar studies, for example, in the state of Nebraska.⁶ It is hoped that this study will inform clinicians about the most common reasons for referral and diagnoses or conditions identified in patients referred for genetic services at a major medical center. This study should help physicians in middle Tennessee and surrounding areas increase their awareness of the types and frequencies of genetic diseases and thereby avoid possible misdiagnoses and delayed referrals.

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Total number of genetics patients seen in four clinical settings at Vanderbilt University Medical Center. Breakdown by 1-year intervals.

Figure 1.
Total number of genetics patients seen in four clinical settings at Vanderbilt University Medical Center. Breakdown by 1-year intervals.

TABLE 1
Most Common Reasons for Referral of Patients for Genetic Counseling (Vanderbilt University Medical Center, 1985 to 1990)

Reason for Referral	Prenatal Counseling Clinics	Ward Consultations	General Genetics Clinics	Outreach Clinics	Total
1. Advanced maternal age (> 35 yr)	181	4	13	5	203
2. Dysmorphic features/multiple congenital anomalies	20	73	74	18	185
3. Developmental delay/mental retardation	0	21	105	42	168
4. Down's syndrome	12	30	49	12	103
5. Abnormal maternal serum α -fetoprotein	74	0	0	0	74
6. Multiple fetal losses	19	0	33	12	64
7. Marfan's syndrome	0	5	39	7	51
8. Failure to thrive	0	30	16	2	48
9. Neurofibromatosis	0	0	42	4	46
10. Short stature	0	6	19	8	33
11. Microcephaly	0	9	14	4	27
12. Cleft lip/cleft palate	0	8	9	8	25
13. Drugs/environment	11	0	9	4	24
14. Turner's syndrome	0	0	20	3	23
15. Spina bifida	0	12	0	2	14
16. Fragile X syndrome	0	3	8	2	13
17. Congenital heart defect	0	6	4	0	10
18. Muscular dystrophy	0	0	10	0	10
19. Hydrocephaly	0	9	0	0	9
20. Phenylketonuria	0	0	8	0	8
	317	216	472	133	1138

TABLE 2

Most Common Diagnoses Made in Patients Receiving Genetic Counseling

Diagnoses	Prenatal Counseling Clinics	Ward Consultations	General Genetics Clinics	Outreach Clinics	Total	Etiology
1. Advanced Maternal Age (≥ 35 yr)	177	0	13	5	195	—
2. Developmental delay/mental retardation	0	16	64	31	111	Multifactorial
3. Dysmorphic features/multiple congenital anomalies	7	41	43	16	107	Unknown
4. Down's syndrome	11	26	41	10	88	Chromosomal
5. Multiple fetal losses	16	0	29	12	57	—
6. Abnormal maternal serum α -fetoprotein	52	0	0	0	52	—
7. Neurofibromatosis	0	0	37	6	43	Single gene (AD)
8. Neural tube defect	9	20	6	2	37	Multifactorial
9. Marfan's syndrome	0	3	27	3	33	Single gene (AD)
10. Chromosomal abnormality (other than trisomy 13/18/21)	8	0	15	0	23	Chromosomal
11. Drugs/environment	10	5	6	2	23	Environmental
12. Turner's syndrome	0	0	18	4	22	Chromosomal
13. Failure to thrive	0	20	0	0	20	Unknown
14. Microcephaly	0	8	10	0	18	Unknown
15. Trisomy 18	5	7	0	2	14	Chromosomal
16. Ehlers-Danlos syndrome	0	3	10	0	13	Single gene (AD, AR, XLR)
17. Hydrocephaly	5	8	0	0	13	Unknown
18. Short stature	0	0	13	0	13	Unknown
19. Cystic fibrosis	0	12	0	0	12	Single gene (AR)
20. Trisomy 13	2	10	0	0	12	Chromosomal
	302	179	332	93	906	