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The Practice of Adult Genetics: A 7-Year Experience From a Single Center

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Abstract

The purpose of our study is to familiarize the reader with genetic disorders commonly seen in adults and identify challenges and barriers that limit provision of services. We conducted a retrospective chart analysis of patients seen in the adult Genetics clinics from January 2004 to December 2010 in a metropolitan medical center consisting of an academic private clinic and a county hospital clinic. During the study period, a total of 1,552 patients (n = 1,108 private clinic patients; n = 444 county clinic patients) were evaluated and managed. Of these, 790 and 280 were new patient visits at the private clinic and county clinic, respectively. Approximately 35% (374/1,070) of new patients were seen for cancer-related indications, while neurological indications accounted for approximately 14% (153/1,070) in both clinics. Cardiology-related indications accounted for approximately 13% (145/1,070) of patients, followed closely by chromosomal and syndromic indications for which almost 9% (96/1,070) of new patients were seen. Approximately 8% (90/1,070) of new patients were seen for musculoskeletal indications. We saw increased clinic growth during the study period and found that the most common indications for referral are: (1) Personal/family history of cancer (2) neurological (3) cardiovascular (CV) (4) chromosomal/syndromic and (5) musculoskeletal. A number of challenges were identified, including coordination of services, feasibility of testing, and an overall higher complexity of care with increased clinic scheduling time requirements. Through this review, we demonstrate the demand for adult genetics services and propose some guidelines to address the challenges of management in the adult genetics patient population.

Keywords

adult genetics; cancer genetics; county clinic; academic clinic; clinical genetics; transitional medicine

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INTRODUCTION

Clinical genetics services historically resided primarily in the realm of pediatrics, and hence the focus has been the diagnosis and treatment of genetic conditions affecting children. According to the American Board of Medical Genetics (ABMG), most geneticists are also certified in another specialty. Given the predominance of pediatric genetics clinics, it is not surprising that the majority of geneticists are also board certified in pediatrics (68%) compared to 10% in internal medicine. While 76% of all new patients seen by geneticists are children, adult (non-prenatal) patients comprise only 13% [Cooksey et al., 2005].

The genetic basis of many diseases, including adult-onset diseases, has been uncovered over the past two decades. With rapid advancement in diagnostic techniques, including array comparative genomic hybridization (aCGH) and next generation sequencing, the clinical availability of testing for these conditions is also growing rapidly. Currently, testing is available for more than 2,200 genetic diseases as compared to approximately 750 diseases just 10 years ago [GeneTests, 2010]. There is also an increasing trend toward genetic services, including ordering of diagnostic tests, counseling, and management being provided by non-geneticists and it remains unclear if these health care providers are adequately prepared and educated to render such services [Taylor, 2003]. In addition, better management of pediatric patients with genetic disorders has increased their longevity with many transitioning to adulthood. While the need for healthcare providers prepared to manage adult patients with genetic disorders is growing, there is clearly a paucity of literature on the practice of adult genetics. We present data from our 7-year experience as a separate adult genetics service, operating within a large academic department.

METHODS

Clinic Settings

We operate two adult genetics clinics that are independent from the department's pediatric genetics clinics: A private clinic run by the medical school's faculty group practice, which sees patients who have health insurance coverage or are capable of self-pay, and a medical school-affiliated county clinic that attends to mostly low-income patients who are either uninsured or covered by Medicaid or Medicare. At the private, academic clinic, 61.8% of patients are Caucasian, 6.0% are African-American, 4.9% are Hispanic, 2.2% are Asian, and data were not available for 25% either because the question was left blank, or people selected "I decline to answer this question." At the county clinic 57.1% of patients are Hispanic, 26.5% are African-American, 9.4% are Caucasian, 4.5% are Asian, 0.1% are American-Indian, and data were not available for 2.4%. Our clinics include the services of medical geneticists and genetic counselors. The county clinic is currently served by two American Board of Internal Medicine (ABIM) and ABMG certified medical geneticists and one American Board of Genetic Counseling (ABGC) certified genetic counselor. Each of the providers seeing patients in the county clinic also sees patients in the academic private clinic which is currently staffed by three ABIM and ABMG certified geneticists, one ABMG certified geneticist, and one ABGC certified genetic counselor. The clinical FTEs of the physicians are 10, 15, 20, and 50%. Both clinics operate one half-day per week. By the end of 2010, we were scheduling an average of 11 patients each week at the private academic

clinic and 12 patients at the county clinic. Since January 2010, the academic, private clinic branched out to include a separate clinic for metabolic and genetic disorders of bone served by one medical geneticist and a genetic counselor and in November 2010 a cardiovascular (CV) genetics clinic was opened and care is provided by a medical geneticist, a genetic counselor, and a cardiologist. Each of these specialty clinics occurs on two half-days per month.

Chart Review

In accordance with IRB approval, we retrospectively reviewed the charts and clinic records for all patients seen at the two clinic locations between January of 2004 and December of 2010. Diagnoses and referral patterns were analyzed. We also calculated the proportion of patients who were scheduled but did not attend their appointments in 2009 and 2010, the years for which this data was available.

RESULTS

A total of 1,552 patients were evaluated and managed in both clinics. The number of patients seen each year has grown significantly over the time period analyzed (Fig. 1), so that a majority of the clinic visits in the 7-year period (820/1,552; 52.8%) has occurred in the last 2 years. Table I elaborates on the indications in the various categories for referral to adult genetics and the percentage of patients seen for each indication category is summarized in Table II for both clinics.

Private, Academic Clinic

One thousand one hundred and eight patients were seen in the academic private clinic during the study period. Of these, approximately 71% (790/1,108) were new patients. Cancer-related indications accounted for 28% (222/790) of new encounters. Visits in the Cardiology category accounted for approximately 17% (134/790) of new encounters and this included patients seen in the CV genetics clinic at the private academic clinic. The majority of our patients seen for neurological conditions were seen in the academic private clinic. In total 153 new patients were seen at both clinics for neurological conditions, and of these, 130 were seen at the private clinic. The proportion of new patients seen at the private clinic for neurological conditions was approximately 16% (130/790). Musculoskeletal indications overall comprised approximately 10% (77/790) of new visits. We experienced a rapid rise in the proportion of new clinic visits in this category in the period reviewed: 1.4% (1/72) in 2004 versus 30.1% (47/156) in 2010. A suspicion of Ehlers-Danlos syndrome (EDS) is one of the most common reasons for referral, accounting for 55% (26/47) of new patients being seen for musculoskeletal indications in 2010. Of these, 20 patients qualified for a diagnosis of EDS. Thirteen were given the clinical diagnosis of EDS, hypermobility type, three were found to have mutations in *COL5A1*, associated with classic EDS, one patient did not have testing due to financial constraints but was given the diagnosis of classic EDS, two patients (mother and daughter) had a previously identified mutation in *COL3A1* associated with EDS, vascular type and one person was given a diagnosis of EDS but the type has not yet been determined. Patients with EDS form a very tightly knit community. We suspect that our sudden increase in the number of patients seen for musculoskeletal conditions from 2009 to

2010 may be attributable to word of mouth recommendations within this community. Of the 47 new patients seen for musculoskeletal indications in 2010, 15 (31.9%) were seen at the clinic for metabolic and genetic disorders of bone, with indications including osteopenia, osteoporosis, osteogenesis imperfecta, and Paget disease. Chromosomal and syndromic indications accounted for approximately 9% (68/790) of new patients. While the number of referrals increased every year, the no show rate was somewhat consistent at 15% in 2009 and 19% in 2010.

County Clinic

Four hundred and forty-four patients were seen in the county clinic during the study period. Sixty-three percent (280/444) of these encounters were with new patients. Cancer-related indications accounted for 54% (152/280) of new visits. The trend has been a steady yearly increase in the proportion of cancer-related visits, from 15% (2/13) in 2006 to 67% (83/124) in 2010. Neurological conditions, while the second most common indication when combining the two clinics, accounted for only 8% (23/280) of new visits at the county clinic. Chromosomal and syndromic indications such as aneuploidies and duplication/deletion syndromes accounted for 10% (28/280) of new patient visits. Approximately 4.6% (13/280) of visits were for musculoskeletal indications. The fifth most common indication category for new patient encounters was endocrinology, which accounted for 4.3% (12/280) of visits. The no show rate was much higher compared to the academic private clinic: 52% in 2009 and 43% in 2010.

DISCUSSION

The field of adult clinical genetics has experienced rapid and continual growth in recent years [Taylor et al., 2006]. The data on the evolution of an adult genetics service presented here clearly reflect this overall trend in genetic medicine. The total number of patients seen nearly tripled from 2004 to 2010 at the academic private clinic, while in the county clinic the total patient load increased by a factor of 18 between 2006 and 2010. During this time period, clinic times have been increased resulting in more available appointment slots, more physicians have joined the practice, and efforts have been made to increase awareness amongst patients and providers about genetics and genetic testing. The patient population included adults with new onset of disease, individuals who were diagnosed with genetic conditions in childhood who are now transitioning into adulthood, adults with genetic conditions that went undiagnosed in childhood, and patients seeking risk assessment because of a family history of a genetic condition. Besides the general advances of knowledge and technology, our experience suggests that physician awareness and education also played a significant role in the rapid rise in the number of referrals to the adult genetics clinics.

Hereditary breast and ovarian cancer and hereditary colorectal cancer were the two most common referrals related to cancer genetics. The large percentage of patients seen for cancer-related indications likely results from several factors: (1) Increasing availability of genetic tests for hereditary cancer syndromes, (2) the results of studies suggesting that treatment decisions based on genetic test results can have a significant impact on patient outcomes (e.g., bilateral prophylactic mastectomy in a *BRCA* mutation carrier), and (3)

increased media coverage and direct-to-consumer testing. The large difference in the proportion of new patients seen for cancer-related indications at the private and county clinics (28% vs. 54%) is likely the result of location-specific factors, including the larger size of the oncology service at the county clinic, and the presence of other institutions in the Houston area that offer cancer-related services to the same patient population as our private clinic. Also, at the private academic clinic, only one geneticist saw all of the patients with cancer-related indications and appointment slots for these patients were limited.

Neurological conditions were the second most common referral indication when data from the two clinics were combined. Again, the discrepancy between the proportion of patients seen at the academic private and county clinics (16% vs. 8%) is most likely the result of local factors, as well as referrals from other specialized clinics such as movement disorder clinic, neuromuscular clinic, and pediatric genetics clinic. The third most commonly seen category was CV indications, which is not surprising considering our affiliation with the Texas Heart Institute, and the presence of a well-established pediatric CV genetics clinic at Texas Children's Hospital from where patients are transitioned to us upon reaching adulthood, and that the genetic basis of conditions like Marfan syndrome, cardiomyopathy, and long QT are well known to non-genetics providers. Syndromic indications such as aneuploidies and other duplication/deletion syndromes accounted for roughly 9% of all visits with approximately 8.6 and 10% at the private and county clinics, respectively. Some reasons for this may be that referring physicians find it easier to identify the need for genetic services in this population and the fact that many of these individuals are transitioning from a pediatric genetics clinic, where there is greater awareness of the availability of adult genetics services. Patients seen for musculoskeletal conditions comprise the fifth most common category. For the last year, since its inception, the Clinic for Metabolic and Genetic Disorders of Bone is included in the academic private clinic data; however, that clinic meets only two half-days per month and is staffed by only one clinical geneticist, who has a research interest in metabolic and genetic disorders of the bone. In our experience, the demand for evaluation for musculoskeletal disorders, particularly EDS, appears to be much higher in the private setting. We hypothesize that certain musculoskeletal problems are being under-diagnosed at the county clinic due to the lack of physician awareness as well as cultural differences in the reasons to seek medical care between the two patient populations. Also, many of our EDS patients self-refer after having read about the condition on the internet.

One large gap in the services we provide pertains to patients with metabolic conditions. As we do not currently have a nurse or a dietitian in our clinic, we are not actively seeking these patients, and except in extremely straight-forward cases, we continue to refer patients that require long-term management of metabolic conditions to the well-established pediatric metabolic clinic at our affiliated institution.

Despite the advances in the field, our experience also exemplifies many hurdles that are likely to be common to other nascent adult genetics practices. Almost half of medical geneticists rate access to genetic services as poor or fair and approximately 75% of reimbursement for genetics services as poor or fair [Cooksey et al., 2005]. In response to another survey, 23% of family physicians stated that genetics consults are difficult or

impossible to obtain. This was particularly true in rural areas and may overestimate the difficulty in obtaining genetics consultation in a major medical center. While Acheson et al. [2005] reported that this difficulty was sometimes due to logistical reasons, such as the geneticists being located several hours away from the family physician, 18% of respondents cited ethical and social reasons for not pursuing a genetic diagnosis. The most common response was that patients fear insurance discrimination. To allay such fears in our genetics clinics we routinely provide extensive genetic counseling to patients prior to testing including information about the Genetic Information Non-discrimination act (GINA) [National Human Genome Research Institute, 2011]. Prior to GINA, patients were often given the option to have testing on a self-pay basis if they wished to avoid testing through their insurance company. However, young adults are encouraged to obtain life insurance prior to undergoing genetic testing, as GINA offers no protection from discrimination involving life insurance.

Another challenge relates in a variety of ways to the high cost of genetic testing. In many cases, patients are unwilling or unable to pay for testing. Some insurance policies do not cover the cost of genetic testing, there is wide variability in coverage criteria and the criteria for coverage are not always consistent with physician recommendations [Myers et al., 1999; Latchaw et al., 2010]. Several studies suggest that insurance coverage of testing and expected out-of-pocket expense is crucial among breast cancer survivors in determining who will have genetic testing [Bosompra et al., 2001; Velicer and Taplin, 2001]. As differences have been noted in the type of insurance coverage by race/ethnicity, we expect and observe disparate access to genetic services for minorities [Suther and Kiros, 2009]. Reducing patients' perceived barriers to testing is important, and might be accomplished by obtaining prior insurance authorization for testing or assisting patients in applying for financial assistance programs to cover the cost of testing. Both of these strategies have been employed in the clinics presented here. Whenever necessary, we also try to serve as patient advocates and routinely write letters of medical necessity. We also seek out opportunities for patients to receive free testing or treatment through research protocols. In 2010, we offered genetic testing to 32.6% (89/273) and 36.9% (93/252) of patients seen at the academic, private clinic, and the county clinic, respectively. We found that at the academic, private clinic approximately 86% (77/89) of patients elected to proceed with testing while the remainder did not undergo testing due to financial constraints. At the county clinic we are required to get approval from the hospital district for any testing not performed at the hospital as these patients rarely have insurance and the cost of testing is absorbed by the hospital. We also utilize the financial assistance programs available at some laboratories. Through these approaches we have been able to arrange for testing for approximately 83% (77/93) of patients offered testing in 2010.

We have also developed relationships with social workers at each clinic site who can help with the needs of our genetic patient population. Patients transitioning from a pediatric genetics clinic to an adult genetics clinic may find it bewildering to navigate through the new type of health delivery system; hence, we try to help them to become more accountable for their health care while providing the support that they need. Another issue that has arisen is related to in-home management and concerns about long-term care.

Health care providers in the adult genetics clinic also face some challenges. High no-show rates and non-compliance are an issue, primarily at the county clinic. The provision of genetic services for these complex conditions requires a large amount of time and resources and reimbursement for services is often poor. On average, total professional time per new patient is estimated to be 7 hr, while follow-up patients receive 3.5 hr of professional time per visit including both direct patient care, preparation and after visit follow-up, with only 15–25% of that time estimated to be potentially reimbursable professional time [McPherson et al., 2008].

Lack of awareness among referring physicians is another limitation of adult genetics. To this end we have maintained a webpage, updated our listing with GeneClinics and the Texas Department of Public Health, and created a marketing brochure targeting providers. An outreach plan of lectures to residents and faculty in internal medicine and primary care physicians in the community is ongoing. We have also tried to make the public more aware of genetic services through our participation in an annual wellness fair hosted at the hospital where the county clinic is located. Similarly, we have participated in community outreach programs organized by our department (www.bcm.edu/eveninggenetics).

Genomic medicine is predicted to revolutionize health care in all aspects of clinical medicine. Beyond the diagnosis of genetic conditions, advances in DNA-based testing are now allowing for assessment of patients' expected response to treatment and targeted therapy through pharmacogenetics [Hamburg and Collins, 2010]. Though not yet mainstream, as the field of personalized genomic medicine expands, impacting all medical specialties, the need for adult genetics services for adequate interpretation of complex and often complicated results is expected to increase [Scheuner et al., 2008]. Strong collaboration between geneticists and primary care physicians will be key to reaping the full benefits of genomic test results [Knotnenerus, 2003].

Despite these challenges, our data suggest that adult genetics clinics are a feasible model for the fulfillment of the widely perceived gap in the quality of care between children and adults with genetic conditions. We believe there are ample opportunities for expansion as the number of patients seen each year continues to grow. In order for the model to be successful, our experience suggests that the training and hiring of a greater number of genetics professionals (physicians and counselors), active outreach to physicians and patients, the involvement of support staff including insurance specialists and social workers, and coordination with pediatric genetics clinics for smooth transitioning of patients, are essential.

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FIG. 1.
Growth of the adult genetics clinics over the course of the study period.

Indications for Referral to Adult Genetics, Separated by Category

TABLE I.

Category	Examples of diagnoses
Cancer	Breast and ovarian, colorectal, renal cell carcinoma, melanoma, pancreatic, von Hippel—Lindau, Birt—Hogg—Dube
Cardiology	Marfan, congenital heart defects, long QT, MTHFR, cardiomyopathies
Chromosomal/Syndromic/ Dysmorphology	Deletions/duplications, Williams, Noonan, Turner, Klinefelter, Proteus, Bardet-Biedl, Stickler, Prader-Willi, Velo-Cardio-Facial syndrome
Endocrine	Congenital adrenal hyperplasia, short stature, acromegaly, Kallmann, multiple endocrine neoplasia, panhypopituitarism
Gastro-intestinal	Hemochromatosis, Wilson, non-alcoholic fatty liver disease
Hematology	Hereditary hemorrhagic telangiectasia, thrombocytopenia, porphyria, hemophilia, thrombophilia
Metabolic/Mitochondrial	Ornithine transcarbamylase deficiency, mitochondrial, trimethylaminuria, diabetes
Miscellaneous	Ashkenazi Jewish screening, reed, psychiatric, ectodermal dysplasia
Musculoskeletal	Ehlers—Danlos, skeletal dysplasia, dwarfism, rhabdomyolysis, craniosynostosis, myopathy, muscular dystrophy, osteoporosis, osteogenesis imperfecta, Paget disease
Neurology	Neurofibromatosis, ataxia, intellectual disability/autism, Charcot—Marie—Tooth, Huntington disease, amyotrophic lateral sclerosis, tuberous sclerosis complex
Ophthalmology/ENT	Retinitis pigmentosa, pendred, usher, hearing loss, cone dystrophy, lebers congenital amaurosis
Pulmonary	Cystic fibrosis, restrictive lung disease, bronchiectasis
Renal/Genito-urinary	Polycystic kidney disease, kidney failure, premature ovarian failure, alport
Recurrent SABs	2 or more spontaneous abortions

This list is not exhaustive of all possible indications for referral, but demonstrates the diversity of indications that were seen during the study period.

TABLE II.

Percentages of New Patients Seen in Each Indication Category in the Clinics

Diagnosis category	Private, academic clinic	County clinic
Cancer	28.1	54.3
Cardiology	17.0	3.9
Chromosomal/dysmorphology/recurrent SABs	8.6	10.0
Endocrine	1.1	4.3
Gastro-intestinal	1.6	1.1
Hematology	3.0	1.1
Metabolic/Mitochondrial	6.2	1.4
Miscellaneous	3.0	4.3
Musculoskeletal	97	4.6
Neurology	16.5	8.2
Ophthalmology/ENT	2.3	1.1
Pulmonary	0.5	0.4
Renal/Genito-urinary	2.2	3.6
Recurrent spontaneous abortions	0.1	1.8