

Research Article

Institutional capacity of health care institutes for diagnosis and management of common genetic diseases - A study from a north coastal district of Andhra Pradesh

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Abstract

Background: In India, the genetic disease is a disregarded service element in the community health- protection system. This study aims to gauge the accessibility of services for treating genetic disorders and also to evaluate the practices on deterrence and management services in the district health system.

Methods: A cross-sectional survey of selected health amenities from 454 medical officers (MO's), 94 accredited social health activist (ASHAs) workers, 86 multipurpose health assistant-female (MPHA-F), 34 multipurpose health assistant-male (MPHA-M), 14 multipurpose health supervisors-female (MPHS-F), 10 multipurpose health supervisors-male (MPHS-M), 6 multipurpose health extension officer/ community health officer (MPHEO/CHO), 10 public health nurse (PHN), 45 lab technicians (LT's) working in the government health sector and 254 in the private health sector, 409 nursing staff working in the government health sector and 995 in the private health sector, 15 primary health centers (PHC's), 4 community health centers (CHC's), 1 district government hospital (DGH), 3 referral hospitals (RH's). From the side of private health institutions 25 corporate hospitals (CH's), 3 medical colleges (MC's), and 25 diagnostic laboratories (DL's) were conducted.

Results: The findings show that adequate staff was in place at more than 70% of health centers, but none of the staff have obtained any operative training on genetic disease management. The largest part of the DH's had rudimentary infrastructural and diagnostic facilities. However, the greater part of the CHC's and PHC's had inadequate diagnostic facilities related to genetic disease management. Biochemical, molecular, and cytogenetic services were not available at PHC's and CHC's. DH's, RH's, and all selected medical colleges were found to have offered the basic Biochemical genetics units during the survey. In 24% of CH's, the basic biochemical units are available and 32% (8 out of 25) of DL's have the advanced biochemical genetics units by study. Molecular genetics units were found to be available in 28% (7 out of 25) of DL's during the study. About 6 (24%) diagnostic centers of cytogenetic laboratories were located in the Visakhapatnam district under the private sector.

Conclusion: The district health care infrastructure in India has a shortage of basic services to be provided for the genetic disorder. With some policy resolutions and facility strengthening, it is possible to provide advanced services for a genetic disorder in the district health system.

Introduction

India is dwelling for genetically varied populations. Due to endogamous practices, there is a high incidence of recessive alleles due to which rare diseases form a serious burden in India. The public health system comprises tertiary level hospitals

established in bigger cities, secondary stage hospitals which are the district and taluka hospitals, and the primary health centers for both the rural and urban populace. Usually, the tertiary level hospitals are associated with a medical school and are concerned with the training of doctors and other medical personnel [1]. The arrangement of health service deliverance,

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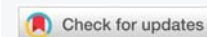
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Keywords: District health system; Genetic disorder; Management practices; Deterrence; Infrastructural amenities





training of personnel, and allotment of the budget is governed by the Ministry of Health & Family Welfare under the Central government. However, the implementation of the health system delivery is predominantly governed by the respective state governments of the different states of India. Shortage of budgetary allotments leads to many gaps in service delivery, and only 25% - 30% of the population is in fact able to access this system. Hence, Private health services accommodate the health needs of the majority of the population [2]. This system is vastly heterogeneous and its pattern differs across different areas of the country. Many urban areas have super-specialty hospitals that are governed by various corporate bodies and these are mostly accessed by the economically privileged class. In general, the majority of Indians both in rural and urban areas approach the individual and small nursing home practices for meeting health care needs [3]. Based on the capability of the doctors and the infrastructure availability the level of care in these health care centers is diversified. Most health care is funded by individuals themselves and in a few cases by the employer while on the other side private medical insurance is also taking roots in India. The Medical Council of India (MCI) regulates the quality of medical practice by establishing guidelines for the training and professional conduct of doctors [4]. Before or after the pregnancy period, if there are problems identified in respect of development such as maturing delays, heart deficiencies, thinking disability, malfunction to thrive, manifold malformations, short size, etc., genetic testing is required. Genetic testing will help in ruling out chromosomal abnormalities in pregnancy and hence is performed while there is an identified family history of chromosomal abnormality [5].

Different types of diagnostic techniques for genetic disorders are applied in clinical laboratories to identify chromosomal abnormalities. In these general cytogenetic analysis methods like chromosome perusal, multiplex ligation-dependent probe amplification, molecular cytogenetic analysis methods, next-generation sequencing, and restriction enzyme fragment length analysis [6,7]. The laboratories for clinical genetic testing are present only in few cities in the country. For laboratory genetic services, patients from rural and suburban areas need to travel long distances. Now a day's, genetic disorders are becoming rising communal health distress in India [8]. Indian Council of Medical Research has launched a website related to medical genetics in India which brings forth a catalog of genetic laboratories and counseling centers for use by patients as well as clinicians. Further, guidelines are also provided for biomedical research related to prenatal diagnostics techniques [9].

This study was conducted to assess the availability and practices of prevention and management services for genetic disorders and to fulfill the gap in these services in the district health system.

Methods

The survey refers to the medical genetic activities

performed in the Visakhapatnam district from 7th June 2019 to 6th April 2021. A descriptive cross-sectional survey of a representative sample of institutional health facilities from Visakhapatnam district, Andhra Pradesh, India was conducted. 12 mandals were selected for the study from the Visakhapatnam district, namely, Anandapuram, Bheemunipatnam, Gajuwaka, Padmanabham, Paravada, Pedagantyada, Pendurthi, Sabbavaram under Visakhapatnam rural region, and, Visakhapatnam urban-1,2,3 and 4 under Visakhapatnam urban region. A cross-sectional survey of selected health facilities from 138 medical officers (MO's) working in government health sector and 316 MO's in private health sector from the related departments of biochemistry, community medicine, pediatrics, general medicine, ENT and obstetrics & gynecology, 94 accredited social health activist (ASHAs) workers, 86 multipurpose health assistant-female (MPHA-F), 34 multipurpose health assistant-male (MPHA-M), 14 multipurpose health supervisors-female (MPHS-F), 10 multipurpose health supervisors-male (MPHS-M), 6 multipurpose health extension officer/ community health officer (MPHEO/CHO), 10 public health nurse (PHN), 45 lab technicians (LT's) working in government health sector and 254 in private health sector, 409 nursing staff (NS) working in government health sector and 995 in private health sector, 91 sub-centers (SC's), 15 primary health centers (PHC's), 4 community health centers (CHC's), 1 district government hospital (DGH), 3 referral hospitals (RH's). From the side of private health institutions 25 corporate hospitals (CH's), 3 medical colleges (MC's) and 25 diagnostic laboratories (DL's) was conducted.

Qualitative and quantitative research methodologies were used to assess the management practices on genetic disorders at the selected health facilities. Data collected from each of the selected health institutes through a combination of self-administered questionnaires and interviews. The study tools were facility checklist; record review for certain services in the study period; human resource availability, infrastructure and equipment, service utilization, referrals, and semi-structured interview schedule for service providers. Where ever indispensable, the responses to the questions were confirmed by inspection and observation of the facilities. The follow-up of the Indian Public Health Standards is examined at each level of the facility. The Institutional Ethical approval and permission were obtained from both Andhra University and Andhra medical college to carry out the survey. Along with this, permission was also sought from the district directors of various health care units. All of them agreed to the inclusion of their facilities for data collection. Throughout the survey, privacy and confidentiality were maintained. SPSS Software Version 19 was used for data entry and analysis (SPSS South Asia Pvt. Ltd, Bangalore, Karnataka).

Results

In the present study, the existing manpower as per Indian



public health standards (IPHS) at different government health centers were observed (Table 1). At the selected sub-centers there are about 88.6% (94) of ASHA workers, 94.50% (86) MPHA-F, 37.63% (34) MPHA-M. In the selected 15 PHC's, there is a working strength of 100% MO's, 93% (14) MPHS-F, 66.33% (10) of MPHS-M, 53% (8) LT's, and 42% NS. MPHEO posts were sanctioned for only 6 PHC's out of the 15 PHCs and all of them were filled and only 10 PHC's have PHN. Further, only 12 PHC's have existing Pharmacist (PC) positions. As per IPHS, CHC's are to be filled with MO's from different professional areas, such as 1 block health officer, 1 general medicine, 1 general surgeon, 1 obstetrician & gynecologist, and 1 pediatrics. In the present study, 81% of MO posts were filled at selected CHC's. From this, the posts of gynecologists were filled up to cent percent of the sanctioned posts. However, general medicine and child specialist posts were filled only at 50%. In all the CHC's the ANM and PHN posts were completely filled, NS including Grade I & II are filled up to 46%, 41% of LT's, and 66% of PC posts were filled. The genetic disease management in Visakhapatnam district government hospital namely King

George Hospital (KGH) was assessed. In this hospital on behalf of manpower gynecologists and general medicine posts were filled up to 86%, and biochemistry and pediatrics posts were filled only up to 66% of the sanctioned posts. ENT and ophthalmology posts were filled at a satisfactory level and community medicine posts were filled with 4 numbers. The supporting staff that is, NS and LT's posts was filled up to 65% and 63% respectively. In the selected referral hospitals like Government ENT Hospital (GEH), Regional eye hospital (REH), and Government Mental Health Hospital (GMHH), the staff position was assessed. The GEH was filled with 20 numbers of ENT specialist posts and 1 pathologist which accounts for 83% and 33% of sanctioned posts. The REH was filled with 77% of ophthalmology specialist posts and the GMHH was filled with 73% of psychiatry specialist posts. On average, 87% of NS and 47% of LT's were filled from the total sanctioned posts in selected RH's. In addition to this survey was also performed in 25 corporate or private hospitals, 3 private medical colleges, and 25 diagnostic laboratories. The services offered by the selected private hospitals are varied based on their specialization in particular procedures. As

Table 1: Availability of manpower in District Government health centers for management of genetic diseases.

S.No	Name of the health center	Personnel	As per IPHS standards total sanctioned positions	Existing pattern % as per IPHS
	Sub-Centers (N=91)	ASHA	106 (Total sanctioned positions under selected sub centers)	94(88.67%)
		MPHA(F)	91(1 at each Sub-center)	86(94.50%)
		MPHA(M)	91(1 at each Sub-center)	34(37.36%)
2	Primary Health Centers (N=15)	Medical officer	15(1 at each PHC)	15(100%)
		MPHS(F)	15(1 at each PHC)	14(93.33%)
		MPHS(M)	15(1 at each PHC)	10(66.66%)
		MPHEO/CHO	15(1 at each PHC)	6(40%)
		PHN	15(1 at each PHC)	10(66.66%)
		LAB Technician GR-II	15(1 at each PHC)	8(53.33%)
		Staff Nurse	45(3 at each PHC)	19(42.22%)
		Pharmacist GR-II	15(1 at each PHC)	12(80%)
3	Community Health Centers (N=4)	Medical staff (Including block health officer (1), obstetrician & gynaecologist (1), general surgeon (1), physician (1) and paediatrics (1) for each CHC)	16(4 at each CHC)	13(81.25%)
		ANM	4(1 at each CHC)	4(100%)
		PHN	4(1 at each CHC)	4(100%)
		LAB Technician	12(3 at each CHC)	5(41.66%)
		Nursing staff (Grade I & II, Head Nurse, Staff Nurse)	60(15 at each CHC)	28(46.66%)
		Pharmacist	12(3 at each CHC)	8(66.66%)
4	District Government Hospital (N=1)	Gynecologists	15	13(86%)
		General medicine	15	13(86%)
		Pediatrics	15	10(66%)
		Biochemistry	3	2(66%)
		ENT	5	3(60%)
		Ophthalmology	6	5
		Community medicine	4	4
		Nursing Staff	450	294 (65%)
		Laboratory Technicians(LTS)	36	23(63.88%)
5	District referral hospitals (N=3) I) Government ENT hospital II) Government regional eye hospital III) Government mental health hospital	ENT (MS.ENT)	24	20 (83%)
		Pathology (MBBS, MD)	3	1(33%)
		Ophthalmology (MS,DO)	22	17(77%)
		Psychiatry (MD)	19	14(73%)
		Total Nursing staff	89	68 (87%)
		Total Laboratory Technicians(LTS)	19	9(47%)

IPHS: Indian Public Health Standards.



shown in table 2, the existing pattern of the doctors, nursing staff, and LT's in all the selected private health care institutions was observed. Of the 25 DLs, only 12 have qualified directors. Nine of the directors are MD in pathology and three are MD in medical genetics or having similar postgraduate degrees. Of all the DLs, only 9 have qualified lab technicians.

Table 3, explain the capacity assessment of district health institutions in genetic disease management. As part of this, case studies were presented to MO's and gynecologists to assess their knowledge and practice on maternal genetic disorders management in selected health care centers. It was found that only 50% of MO's in district government hospitals and CHC's are aware of multifactorial, monogenetic, and other chromosomal anomalies. Among these MO's, the majority of

gynecologists reported that they were trained for placental biopsy, chorionic villus sampling, and other Biochemical parameters. Merely 32% of CH's and 36% of DL's provide these services on monitoring maternal genetic disorders. 33% of PHC's and 25% of SC's have knowledge of maternal genetic disorders. The majority of centers perform well at monitoring genetic birth defects. Management of these defects was provided by 75% of DH's and CHC's, but only at 32% of CH's and 36% of DL's. Knowledge acquired staff regarding genetic birth defects management is available at 53% of PHCs and 50% of SCs. Services regarding maintenance of family health history checklist during pregnancy and maternal death review (MDR) with identified genetic defect deaths related to genetic disorders were only available at DGH, 36% of CH's and 32%

Table 2: Availability of manpower in selected district private health centers for management of genetic diseases.

S.No	Type of the heal care center	Personnel	Existing pattern
	District corporate hospitals (N=25)	Gynecologists	26
		General medicine	29
		Pediatrics	22
		Biochemistry	8
		ENT	21
		Nursing Staff	725
		Laboratory Technicians(LTS)	82
2	Medical colleges (N=3)	Biochemistry	20
		Obstetrics & Gynecology	40
		Pediatrics	34
		General Medicine	52
		ENT	16
		Ophthalmology	15
		Community medicine	21
		Nursing Staff	270
3	Selected diagnostic laboratories (N=25)	Availability of Doctors/Medical officers related to genetic disorders	12
		Number of laboratory technicians (LTS)	150

Table 3: Capacity assessment of District health institutions in genetic disease management.

S.No		District Government hospital & referral hospitals (n = 4)	CHC (n = 4)	PHC (n = 15)	Sub-centers (n = 91)	District Corporate Hospitals (n = 25) (DCHs)	Private Diagnostic laboratories (n = 25) (PDLs)
% reporting good							
1	Maintenance Family Health History Checklist: During Pregnancy	1(25%)	0(0.0%)	7(46%)	39(42.85%)	9(36%)	8(32%)
2	Monitoring/Knowledge in Maternal Genetic Disorders	2(50%)	2(50%)	5(33%)	25(25%)	8(32%)	9(36%)
3	Monitoring in Genetic Birth Defects	3(75%)	3(75%)	8(53%)	46(50%)	8(32%)	9(36%)
4	Maternal Death Review (MDR) and identified genetic defect deaths related to genetic disorders.	1(25%)	0(0.0)	0	0	4(16%)	7(28%)
5	Update genetic history of the children	1(25%)	0(0.0%)	0(0.0%)	0(0.0%)	8(32%)	5(20%)
6	Record maintenance	4(100%)	2(50%)	12(80%)	48(45.05%)	8(32%)	8(32%)
7	Observed / follow up treatment genetic diseases from last one year	3(75%)	3(75%)	12(80%)	48(45.05%)	9(36%)	9(36%)
8	Availability of Doctors/Medical officers related to genetic disorders	3(75%)	4(100%)	15(100%)	0	9(36%)	8(32%)
9	Role of Doctors/Medical officers in genetic disease management	3(75%)	4(100%)	11(73%)	0	8(32%)	8(32%)
10	Role of Nuring staff in genetic disease management	3(75%)	3(75%)	10(66.66 %)	0	7(28%)	8(32%)
11	Role of Laboratory Technicians in genetic disease management	2(50%)	2(50%)	8(53%)	0	6(24%)	8(32%)
12	Genetic Counselors availability/role in disease management.	0	0	0	0	0	0



of DL's. It was observed that the majority of health centers were good at record maintenance and follow-up treatment of genetic diseases from the last year. Very few health institutes updated the genetic history of the children in the present study. A majority of NS in both DH's and CHC's play a good role in genetic disease management. Few health centers have qualified lab technicians with a good role in genetic disease management. Special expertise in genetic counseling is not available in any of the selected health centers. As a result, general pediatricians, hematologists, obstetricians, and medical specialists are providing genetic counseling in this area.

Table 4, reveals the capacity assessment of facilities providing genetic services from the selected health institutions in genetic disease management. Results of a survey performed in 25 DL's, 25 CH's, DH's, medical colleges (1 Government and 3 private), 4 CHC's and 15 PHC's were taken and analyzed the obtainable facilities for genetic disease management. DH's and all selected medical colleges have offered the basic Biochemical genetics units in the survey. 24% (6 out of 25) CH's are acquainted with basic biochemical units and 32% (8 out of 25) of DL's have advanced biochemical genetics units. 28% of DLs have (7 out of 25) molecular genetics units in the study. These laboratories carry out the study for DNA short lengths to identify alterations that caused a genetic disorder. About 6 (24%) DL's of cytogenetic laboratories were located in the Visakhapatnam district under the private sector. In general, these centers handle the referred genetic disorder cases and it shows that referral was most commonly for legal reasons. SRL diagnostic lab, Vimta lab, Quantum specialty diagnostics, Access Path Lab, Metropolis, and Vijaya diagnostic center are offering diagnosis for monogenic diseases. In general cystic fibrosis (CF), hemophilia A/B (F8/F9), β -thalassemia, sickle-cell disease (SCD), and fragile X syndrome (FRAXA) are obviously the most common disorders tested by the labs. Newborn screening is offered in 75% of DH's and all medical colleges; whereas only 32% of CH's and 36% of DL's are providing this facility in the present study. Early detection of genetic visual impairment and genetic hearing impairment are offered by about 75% of DH's and by all the selected medical colleges. Out of 25 DCHs and 25 diagnostic labs, 8 numbers each offer early detection of genetic visual impairment. Regarding early detection of genetic hearing impairment, only 9(36%) CH's and 8(32%) DLs offer the service.

Discussion and conclusion

In our study, the existence and demand for genetic disorder services of substantial enormity have been reported by service providers. In the past Venugopal, et al. reported on the high genetic burden in India [10]. For that reason, for meeting the unmet requirements of persons with genetic diseases, the public sectors need to be prepared.

The results of this study indicate that in the government health sector, basic services for identifying genetic disorders were not available particularly up to the PHCs level with the exception of basic blood, serum, and urine diagnostic tests. Previous studies also support these findings with Verma and Bijarnia that due to scarce diagnostic and management conditions, the trouble of genetic disorders is soaring in India and hence the existing primary health care services should be upgraded with these services. Basic genetic screening services, as well as carrier testing, are available for needy persons in District government hospitals namely 'KGH' and also in CHCs for hemophilia, thalassemia, sickle-cell anemia, and for some primary immune deficiencies. For screening hemoglobinopathies in pregnant women and children, a high significance was given in the 'KGH'. Previous studies also support these findings that the district government hospitals have the above testing capabilities [11,12]. For advanced detection of the genetic disorders, the cases were referred to diagnostic laboratories for examination of cytogenetic or molecular genetic analysis [13]. In the present study, it was noticed that many service providers have limited skills in diagnosing and managing genetic diseases due to a lack of in-service training. There is a necessity of imparting high-quality in-service training to the MOs and paramedical staff for facilitating them with specialized skills for advanced management of genetic diseases. Graf and Frank previously made similar findings [14].

As observed from the district corporate hospitals, genetic specialists are limited in number, and merely a small amounts of hospitals offering specific genetic services. 76% of hospitals do not have advanced investigation facilities for identification of the genetic diseases with biochemical genetics units. All the selected corporate hospitals did not have cytogenetic or molecular genetics units. 85% of gynecology departments maintained services of prenatal diagnosis and termination of pregnancy and similarly, general medicine units are

Table 4: Facilities providing genetic services in the District health institutions.

S.No	Health facility type/service	PHC (n = 15)	CHC(n = 4)	District Government & referral hospitals (n = 4)	District medical colleges		District Corporate Hospitals (n = 25)	Private Diagnostic laboratories (n = 25)
					Public (n = 1)	Private (n = 3)		
1	Biochemical Genetics Unit	0	0	1(25%)	1(100%)	3(100%)	6(24%)	8(32%)
2	Molecular Genetics Unit	0	0	0	0	0	0	7(28%)
3	Cytogenetics Unit	0	0	0	0	0	0	6(24%)
4	Newborn screening	0(0%)	0(0%)	3(75%)	1(100%)	3(100%)	8(32%)	9(36 %)
5	Early detection of genetic visual impairment	0(0%)	(0%)	3(75%)	1(100%)	3(100%)	8(32%)	8(32%)
6	Early detection of genetic hearing impairment	0(0%)	0(0%)	3(75%)	1(100%)	3(100%)	9(36%)	8(32%)



handling newborn hearing impairment treatment services. Previously Thomas mookken reported on significant issues for the implementation of a newborn screening program and how different model regional screening programs managed in India [15]. In the private sector, newborn screening is available in only a few hospitals (32%). As observed from human resources, the majority of corporate hospitals have a less number of trained doctors, nurses, and lab technicians for managing genetic diseases. Earlier reports by Mookken and Cariappa also supported these findings [16].

Selected medical colleges have the availability of care departments for genetic disease management and they have a satisfactory level of manpower such as staff nurses and LTs and they performed the proper roles in genetic disease management. The availability of biochemical genetic units which contain PCR and electrophoresis apparatus was present in all medical colleges and they are in good condition but they lack cytogenetic and molecular genetics units. All the colleges are good at newborn hearing screening, prenatal diagnosis, and termination of pregnancy. In the selected medical colleges, the syllabus on hereditary disorders is included in the subjects of biochemistry, obstetrics & gynecology, and pediatrics within the period of four and half year medical degree program. It was observed that the curriculum is following the guidelines of the medical council of India. Shagun Aggarwal & Shubha R Phadke, supported these studies in their previous findings [17].

From this survey what we have observed is that the majority of respondents of medical personnel have a lack of know-how in identifying genetic risk factors and treating genetic disorders due to inadequate training. This is a prime barrier in genetic disease management. To tackle such situations, recently national policy for the treatment of rare diseases is initiated by the Ministry of health and family welfare, the government of India in 2021 [18]. The launching of the Indian rare disease registry by ICMR is a step towards associating the healthcare providers and patients' distress from rare genetic diseases. Throughout the country, the registry is useful as a general data stockpile area [19].

The findings indicate that most of the services for the prevention of genetic diseases were available in limited health centers in the Visakhapatnam district health system. Hence there is a need for strengthening genetic diseases management services at the primary level in this region. For the better management of genetic diseases in the selected area, we are making some propositions. They are; educating the public on genetic risks, increasing the number of genetic testing units, improving the skills and expertise of the health care personnel by imparting necessary training in molecular genetics technology, and developing national policies for reinforcing genetic services.

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Authors' contributions

Substantial contributions to the design and interpretation of data for the work.

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