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Abilities of Health Care Centers towards Genetic Services: A Study from an Indian Coastal District

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KEYWORDS

District health system, genetic disorder, human resource, infrastructural amenities, management practices, public health

ABSTRACT:

Introduction: In India, the services for genetic diseases are mostly ignored in the community health system.

Aim: The present study targets to focus on the available amenities and management practices for treating genetic diseases in the district health system.

Materials and methods: The Visakhapatnam district in India was selected for the survey. A cross-sectional survey of health facilities related to genetic diseases in government and private health sectors was conducted. From the government health establishments, 597 Sub-centers (SCs), 82 Primary Health Centers (PHCs), 12 Community Health Centers (CHCs), 3 District Hospitals (DHs), 2 Area Hospitals (AHs), 1 Medical College (MC) and 3 Referral Hospitals (RHs) were studied. In respect of private health institutions, 80 Corporate Hospitals (CHs), 62 Diagnostic Laboratories (DLs) and 3 Medical Colleges (MCs) were selected for the survey. From these, the existing manpower and their disease management abilities were also examined. Quantitative and qualitative investigation methods were used to estimate the managing operations on genetic complaints at the selected health facilities.

Results: The findings indicate that satisfactory level of manpower was in place at more than 70% of the health centers, but none of the medical staff have gone through any operative training on genetic disease control and hence are lacking effective knowledge in this field. A few basic infrastructural and diagnostic amenities for managing genetic diseases were available at the District hospital of Visakhapatnam.

Conclusion: By the facility strengthening and advanced policy resolutions, it is possible to provide progressive disease management in the district health system.

1. Introduction

Presently, genetic diseases have become an intensifying public health issue in the world including India. Many communities in India still have a high prevalence of genetic diseases due to practices such as consanguineous marriages. To overcome this situation, it is necessary to maintain the family health history and early genetic testing, which will be helpful to prevent the diseases in advance.[1, 2]

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The public health system in India comprises of three major categories of health system; the primary health centers are for both the urban and rural population, district and taluka hospitals evolving as secondary stage hospitals, and bigger cities accommodating the tertiary level hospitals. Generally, trained doctors and well equipped laboratories are available in the tertiary level hospitals.[3] The budget allocation and health service deliverance are monitored by the central government under the department of Ministry of Health & Family Welfare. Due to shortage in budgetary allocations, there is a shortfall in service delivery and this affects a large section of the population. The health needs of majority of the patients are being met by private health services.[4] Most of these private health services are offered by super-specialty hospitals which are located in the urban areas and these are mostly accessed by the financially strong families. Further, the advanced diagnostic labs are also present only in few cities in the country.[5] As most of the people in India live in rural areas, a major sector of the patients need to travel to urban areas for laboratory genetic services, which is a costly affair.[6] In India, ICMR provides complete information related to medical genetics and genetic experiments to patients as well as individual clinicians through its website.[7]

The purpose of the present study is to analyze the genetic disease management capacity in Visakhapatnam district by studying the availability of existing manpower, usage of technologies and type of tests performed. By knowing the actual conditions, it will be helpful to improve finest strategies for strengthening the genetic services.

2. Methods

2.1 Study setting: The survey focuses on the medical genetic operations performed in the highly urbanized north coastal district of Visakhapatnam from 1st November, 2020 to 30st October, 2023. A descriptive cross sectional survey of institutional health amenities from highly urbanized coastal district of Visakhapatnam, Andhra Pradesh, India was conducted. The district has four divisions, namely, Anakapalli, Narsipatnam, Paderu and Visakhapatnam and all the government health centers in these divisions were selected for the present survey. They are 597 Sub Centers (SCs), 82 Primary Health Centers (PHCs), 12 Community Health Centers (CHCs), 3 Referral Hospitals (RHs), 3 District Hospital (DH), 4 Medical Colleges (MCs) (1 public and 3 private). In respect of private health sector, reputed health centers were selected duly covering all the divisions, which include 80 Corporate Hospitals (CHs) and 62 Diagnostic Laboratories (DLs), located at the division headquarters. From these government and private health sectors, all the manpower and institutional capacities which are related to genetic disease management were studied.

The ethical consent was acquired from both Andhra Medical College and Andhra University, Visakhapatnam to carry out the study. Along with this, approval was also sought for from the directors of various district health units and all of them approved and allowed for collecting the data from their respective health centers.

2.1.1 Inclusion criteria

The various criteria considered for this survey are; institutional facilities such as infrastructural facilities and existing diagnostic services, role of the doctors and other associated health workers in the management of genetic diseases.

2.1.2 Exclusion criteria

Health workers who are not cooperative (or) are not willing to participate in the study and unpopular corporate hospitals and diagnostic institutes are excluded from the survey.

2.2 Data collection and analysis

Quantitative and qualitative research procedures were used to estimate the management operations on genetic diseases at the selected health centers. Information was taken from each of the selected health canters through interviews and self-administered questionnaires. The survey implements through verification of facility list, record analysis for certain services in the present period; availability of human resources, equipment and infrastructure, utilization of certain services and conducting a small interview program for facility providers. Where ever necessary, the replies to the questions were confirmed by direct examination of the facilities. The Indian Public Health Standards (IPHS) component has been adopted as standard at every level. Confidentiality and privacy were maintained throughout the survey. The collected data was scrutinized using SPSS version 16 using expository statistics.

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3. Results

3.1 Manpower analysis

In the present study, the manpower existing at different government health centers for management of genetic diseases was observed with reference to Indian public health standards (IPHS) (Table-1). Accredited Social Health Activist (ASHA) workers are filed up to 91.02% against the sanctioned posts among the sub centers in the surveyed area. The Multipurpose Health Assistant (MPHA) female posts are at satisfactory level, but the deficiency in MPHA male posts is up to 73.05%. Multi-Purpose Health Supervisor (MPHS) female posts are at satisfactory level (90.24%) and however, there is a deficiency of 39.24 % in MPHS male posts.

In respect of the PHCs, each center has a sanctioned post of one MO and all these positions are completely filed. As per health standards, each PHC shall have minimum one Nursing Staff (NS). As verified in the PHCs, the deficiency of NS and Lab Technician's (LTs) is 23.38% and 11.89% respectively. In the CHC's, MO's are to be filled from different specialization areas as per health standards. It was observed that only 61% of MO posts were filled at the selected CHCs. All the sanctioned posts of gynecologists were filled, but, the general medicine and child specialist posts were filled only up to 50%. There are three number of district hospitals in Visakhapatnam district located at Visakhapatnam city (King George Hospital-KGH), Anakapalli and Paderu. The manpower engaged in management of genetic diseases was assessed from these district hospitals and it was observed that the gynecologist and general medicine posts were filled up to 85%, and ENT and ophthalmology posts were filled up to satisfactory level. However, the biochemistry and pediatrics posts were filled only up to 36% as compared to the sanctioned posts.

In the selected three referral hospitals including the Government ENT Hospital (GEH), Regional eye hospital (REH), and Government Mental Health Hospital (GMHH), the available staff position and their genetic disease management capacities were analyzed. The GEH was filled with 20 numbers (83%) of ENT specialist posts and 1 number (33%) of pathologist post. The REH was filled with 77 % of ophthalmology specialist posts and the GMHH was filled with 73% of psychiatry specialist posts. In all the three referral hospitals, it was observed that there is a lack of expertise related to genetic disease management. On an 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 conducted in district private health centers comprising of 80 corporate hospitals, 3 medical colleges, and 62 diagnostic laboratories. The selected private hospitals offered services which are varied based on their specialization in particular procedures. As shown in table-2, the current pattern of the doctors, NS and LTs in all the selected corporate health care centers was observed. Of the 62 DLs, only 14 labs have specialized and experienced directors and most of these directors are MD in pathology (11) and the remaining (3) are MD in medical genetics. Of all the DLs, only 9 have qualified lab technicians. All the government health centers in this region generally refer their patients to private diagnostic labs at district headquarters for accurate diagnosis of genetic diseases.

3.2 Ability assessment of the health institutions

Graph-1, Table-3, explains the ability assessment of district health centers in genetic disorder management. As part of this, the knowledge of gynecologists and other related medical officers in maternal genetic disorder management was assessed from selected health centers. It was observed that, very less number of medical officers are dealing the monogenetic and multifactorial anomalies. Among these medical officers, especially gynecologists have reported that they were trained for different biochemical parameters, long term villus sampling and biopsy of placenta. Among the supporting staff like staff nurses and others, only few are playing a considerable role in genetic disorder management. Special expertise in genetic counseling is not available in any of the selected health centers. Pediatricians, obstetricians, hematologists, and other related medical specialists are providing genetic counseling to some extent in this area.

3.3 Existing genetic services

Table-4 reveals the capacity assessment of genetic testing facilities from selected health centers in disease management. Basic equipment for managing genetic diseases was not readily available in most of the health centers, particularly in the PHC level. DHs and all the selected medical colleges are having basic Biochemical genetic services. However, majority corporate hospitals

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(76%) in this region did not have the basic biochemical units. Regarding the DLs, 32 % (8 out of 25) of DLs are having advanced biochemical genetics units, but 72% of DLs does not offer the molecular genetics services. Very less number of DLs that are offering cytogenetic examination are located in the Visakhapatnam district. In general, these DLs manage the referred genetic disorder cases. SRL diagnostic lab, Quantum specialty diagnostics, Metropolis, Vimta lab, Access Path Lab and Vijaya diagnostic centers are offering diagnosis for monogenic diseases. β-thalassemia, sickle-cell disease, cystic fibrosis, hemophilia, and fragile X syndrome are most common disorders observed in this area.

4. Discussion

In the present investigation, the evolving enormous need for extensive genetic disease facilities have been identified. In the past similar findings on high genetic encumbrance in India were made by Venugopal et al., and it was suggested that the public sectors should be prepared for addressing all the requirements of patients for management of genetic diseases.[8]

4.1 Amenities of government health sector at genetic disease management

4.1.1 Infrastructure

The results of this study indicate that in the public health sector, especially up to the PHC level, only simple serum, blood and urine analysis tests are available and there is no availability of even basic services for detection of genetic disorders. Previous research by Koteswara Rao Pagolu and T Raghava Rao, (2021) support the present findings that the problem of genetic disorders is increasing in India due to rare diagnosis and management conditions and therefore existing primary health care centers should be upgraded with these services.[9] Basic diagnostic facilities related to genetic disorders like hemophilia, thalassemia, sickle-cell anemia, and other primary immune deficiencies are available for needy patients in CHCs and government district hospitals. In Visakhapatnam district, screening for hemoglobinopathy in pregnant women and children is conducted in KGH on priority. Previous studies have supported these findings that the district government hospitals have the above mentioned testing capabilities.[10] However, as advanced diagnostic facilities for examination of cytogenetic or molecular genetics analysis, are not available here, the patients are being referred to private diagnostic laboratories in Visakhapatnam city for testing. It was demonstrated by universal screening panel investigations conducted in 2020 that for advanced detection of genetic disorders, referral cases were sent to private diagnostic laboratories.[11]

4.1.2 Manpower

In the present study, it was observed that many service providers are ineffective in managing genetic diseases due to lack of in-service training. For advanced management of genetic diseases in this region, there is an immediate need for imparting the MOs and supporting staff with specialized training and skills. Previously, Graf and Frank have also made similar findings that due to lack of proper training of service personal, the prevalence of genetic burden is high.[12]

4.2 Corporate hospital facilities for genetic diseases

An important observation from the district corporate hospitals is that very few hospitals are offering genetic services and further, they have a limited number of genetic experts. About 76% of hospitals do not have investigation facilities like advanced biochemical genetics units to detect genetic diseases. Cytogenetic or molecular genetics units are not available in all the selected corporate hospitals. 85% of prenatal diagnosis and termination of pregnancy services are handled by gynecology departments, and the neonatal hearing loss treatment services are managed by general medicine units. Earlier reports by Abhiruchi galhotra and Preeti Sahu (2019) are also supporting these observations that the accessibility of hearing screening is generally available to newborns at corporate hospitals with advanced general medicine units.[13] In respect of human resources, many corporate hospitals do not have expert doctors, nurses and lab technicians for the management of genetic diseases. Previous reports by Kathleen et al in 2010 also support these findings.[14]

4.3 Abilities of teaching health institutes for genetic diseases

All the selected medical colleges have relevant care departments for management of genetic diseases and also have satisfactory level of manpower engaged in performing proper role in genetic disease management. Regarding the facilities provided by these medical

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colleges, biochemical genetic units with polymerase chain reaction (PCR) and electrophoresis equipment with good condition are present in all the medical colleges, but there are no facilities for molecular and cytogenetic genetic analysis. All colleges have good management in respect of prenatal diagnosis and termination of pregnancy as per need, and also regarding newborn hearing screening. In the selected medical colleges, hereditary disorders have been included as a syllabus in the biochemistry, obstetrics & gynecology and pediatric departments of the four-and-a-half-year medical degree program. It was observed that the guidelines of the Medical Council of India are being followed in the curriculum of the selected medical colleges. The above findings are supported by Shagun Aggarwal & Shubha R Phadke in the year 2015 stating that MCI has taken steps to upgrade genetics curriculum at all stages of medical courses.[15] From this survey we observed that most of the medical personnel fail to treat genetic disorders because of inadequate training in identifying genetic risk factors and this is considered as a major obstacle in the management of genetic diseases. To address such hurdles, a national policy for the treatment of rare diseases was launched in 2021 by the Ministry of Health and Family Welfare, Government of India, which is aiding in genetic disease management.[16] Earlier, ICMR also launched common data registry for genetic diseases, which is helpful for the needy patients and health care providers.[17]

5. Limitations

Present study was mainly questionnaire-based investigation. However, actual cases were not directly examined and this is the major limitation of the study. For extracting realistic information, we created hypothetical situations in case studies. Also the assessment of the diagnostic infrastructure efficiency was not monitored physically.

6. Conclusion

From the studied area it is observed that in the Visakhapatnam district health system, the services for advanced management of genetic diseases were not available in the rural area and very limited health centers offer the services in the Visakhapatnam city.

Due to lack of clinical diagnostic capacity and lack of trained personnel in primary health care especially in rural areas, patients with genetic diseases are suffering a lot. So, in the grassroots hospitals for genetic disorder management, it is imperative to provide adequate manpower and infrastructure for speedy and effective service delivery.

In the study region, general medicine and pediatricians are handling the patients with genetic diseases due to shortage of geneticists. However, these doctors are having lower knowledge about rare diseases when compared to their respective fields. Hence, to reduce the access gap faced by rare disease patients, it is necessary to deploy geneticists even from the primary health care level. Also, to achieve the goal of reducing the prevalence of genetic disorders in public health system, the appointment of genetic counselors is essential. For successful implementation of genetic disease prevention programs, it is very important to develop national policies to strengthen the genetic services and also to educate the patients and general public for early diagnosis.

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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 under selected sub centers)	Existing pattern % as per IPHS
1	**Sub-Centers (N=597)**	ASHA	769	700 (91.02 %)
		MPHA(F)	597 (1 at each Sub-center)	540 (90.45%)
		MPHA (M)	386	282 (73.05 %)
2	**Primary Health Centers (N=82)**	Medical officer	82	82 (100%)
		MPHS(F)	123	111 (90.24%)
		MPHS(M)	105	68 (60.76%)

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S.No	Name of the health center	Personnel	As per IPHS standards total sanctioned positions under selected sub centers)	Existing pattern % as per IPHS
		MPHEO/ CHO	38	32 (84.21 %)
		PHN	44	39 (88.63%)
		LAB Technician (GR-II)	101	89 (88.11%)
		Staff Nurse	154	118 (76.62%)
		Pharmacist GR-II	77	61 (79.22 %)
3	**Community Health Centers (N=12)**	Medical staff (Including block health officer (1), obstetrician & gynecologist (1), general surgeon (1), physician (1) and pediatrics (1) for each CHC	79	61 (77.21 %)
		Paramedical staff	64	36(56.25 %)
		LAB Technician	39	22 (56.41%)
		Nursing staff (Grade I & II, Head Nurse, Staff Nurse)	106	88 (83.01%)
4	**District Hospitals (Government) (N=3)**	Gynecologists	44	37 (84.09%)
		General medicine	39	33 (84.61%)
		Pediatrics	25	16 (64%)
		Biochemistry	15	9 (64.28%)
		ENT	26	18 (69.23%)
		Ophthalmology	25	19 (76%)
		Community medicine	11	6 (54.54%)
		Nursing Staff	352	299 (84.94%)
		Laboratory Technicians (LTS)	41	34 (82.92%)
5	**Area Hospitals (N=2)**	Gynecology	14	5 (35.71%)
		General medicine	12	4 (33.33%)
		ENT	11	4 (36.36%)
		Ophthalmology	10	5 (50%)

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S.No	Name of the health center	Personnel	As per IPHS standards total sanctioned positions under selected sub centers)	Existing pattern % as per IPHS
		Pediatrics	12	5 (41.66%)
		Neonatology	4	2 (50%)
		Nursing Staff	37	28 (75.65%)
		Laboratory Technicians (LTS)	9	7 (77.77%)
6	**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)	24	12 (50%)

(IPHS): Indian Public Health Standards; ASHA-Accredited social health activist; MPHA(F)-Multipurpose health assistant-female; MPHA(M)-Multipurpose health assistant-male; MPHS(F)- Multipurpose health supervisors-female; MPHS(M)-Multipurpose health supervisors-male; MPHEO/CHO- Multipurpose health extension officer/ community health officer; PHN-public health nurse (PHN).

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
1	**District corporate hospitals (N=80)**	Gynecologists	59
		General medicine	52
		Pediatrics	40
		Biochemistry	2
		ENT	32
		Endocrinology	1
		Ophthalmology	14
		Neonatology	9
		Neurology	4
		Internal medicine	1
		Dermatology	1

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S.No	Type of the heal care center	Personnel	Existing pattern
		General practice (MBBS only)	5
		Nursing Staff	525
		Laboratory Technicians(LTS)	155
2	**Medical colleges (N=4)**	Biochemistry	15
		Obstetrics & Gynecology	24
		Pediatrics	18
		General Medicine	16
		ENT	14
		Ophthalmology	17
		Community medicine	11
		Nursing Staff	142
		Laboratory Technicians(LTS)	38
3	**Selected diagnostic laboratories (N=62)**	Availability of Doctors/Medical officers related to genetic disorders	26
		Nursing Staff	54
		Number of laboratory technicians (LTS)	271

Table-3: Capacity assessment of the manpower in the District health institutions against genetic disease management.

S.No	Role of the manpower	DHs (n=3)	RHs (n=3)	Ahs (n=2)	CHCs (n=12)	PHCs (n=82)	CHs (n=80)	DLs (n=62)	MCs (n=4)
		% reporting good							
1	**Details of the Doctors: Present working number of posts**	140	52	25	61	82	220	26	115
	Exposure to genetic testing during graduate training (Poor/Good/Excellent)	140 (100%)	52 (100%)	2 (100%)	12 (100%)	82 (100%)	220 (100%)	26 (100%)	115 (100%)
	Good monitoring of Maternal Genetic Disorders	65 (46.42%)	24 (46.15%)	12 (48%)	29 (47.54%)	30 (36.58%)	115 (52.27%)	14 (53.84%)	71 (61.73%)
	High risk pregnancy : aware of genetic risk factors (Poor/Good/excellent)	64 (45.71%)	24 (46.15%)	12 (48%)	28 (45.20%)	35 (42.68%)	98 (44.54%)	14 (53.84%)	69 (60%)
	Update genetic history of the children	0	0	0	0	0	22 (10%)	12 (46.15%)	28 (24.34%)
	Monitoring/Knowledge in Genetic Birth Defects	65 (46.42%)	23 (44.23%)	13 (52%)	30 (49.18%)	35 (42.68%)	98 (44.54%)	14 (53.84%)	71 (61.73%)

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S.No	Role of the manpower	DHs (n=3)	RHs (n=3)	Ahs (n=2)	CHCs (n=12)	PHCs (n=82)	CHs (n=80)	DLs (n=62)	MCs (n=4)
		% reporting good							
	Follow up treatment genetic diseases from last one year	99 (70.71%)	32 (61.53%)	17 (68%)	48 (78.68%)	49 (59.75%)	129 (58.63%)	22 (84.61%)	78 (67.82%)
	Good maintenance of Birth defects record	91 (65%)	39 (75%)	17 (68%)	38 (62.29%)	47 (57.31%)	125 (56.81%)	24 (92.30%)	88 (76.52%)
2	**Total Nursing Staff**	299	68	28	88	118	525	54	142
	Good Role of Nursing staff in genetic disease management.	222 (74.74%)	35 (51.47%)	17 (60.71%)	32 (36.36%)	65 (55.08%)	298 (56.76%)	38 (70.37%)	88 (61.97%)
3	**Total Laboratory Technicians (LTS)**	34	12	7	22	89	155	271	38
	Role of Laboratory Technicians in genetic disease management. (Poor/ Good / Excellent).	26 (76.47%)	6 (50%)	4 (57.14%)	6 (27.27%)	38 (42.69%)	21 (13.54%)	68 (25.01 %)	14 (36.84%)
4	**Genetic Counselors availability/role in disease management.**	0	0	0	0	0	0	5 (8.06%)	0

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

S.No	Health facility type/service	DHs (n=3)	RH (n=3)	AHs (n=2)	CHCs (n=12)	PHCs (n=82)	CHs (n=80)	DLs (n=62)	MCs (n=4)
1	**Biochemical Genetics Unit**	1 (33.33%)	0	1 (50%)	0	0	15 (18.75%)	12 (19.35%)	2 (50%)
2	**Molecular Genetics Unit**	0	0	0	0	0	0	12 (19.35%)	0
3	**Cytogenetics Unit**	0	0	0	0	0	0	10 (16.12%)	0
4	**Newborn screening**	3	1 (33.33%)	1 (50%)	4 (33.33%)	0	14 (17.5%)	14 (22.58%)	4 (100%)
5	**Early detection of genetic visual impairment**	3	1 (33.33%)	1 (50%)	4 (33.33%)	0	16 (20%)	14 (22.58%)	4 (100%)
6	**Early detection of genetic hearing impairment**	3	1 (33.33%)	1 (50%)	3 (12%)	0	11 (13.75%)	13 (20.96%)	4 (100%)

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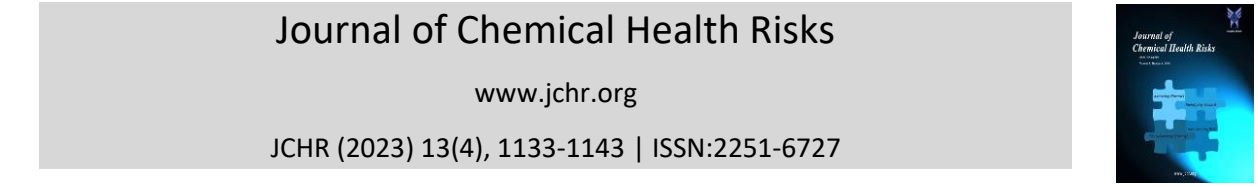
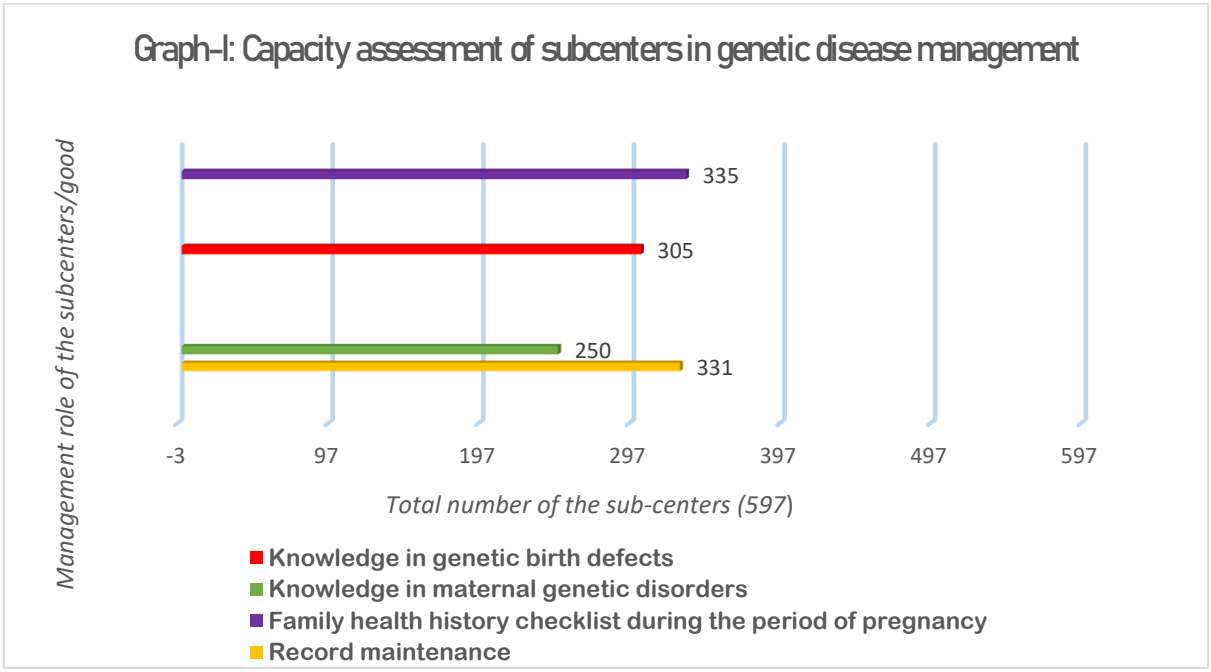


DH: District hospital; RHs: referral hospitals; CHCs-community health centers; PHCs- primary health centers; SCs- sub centers; CHs- corporate hospitals; DLs- diagnostic laboratories; MCs-Medical colleges



Graph-I: Capacity assessment of subcenters in genetic disease management