ACCSCIENCE PUBLISHING

Gene & Protein in Disease

PERSPECTIVE ARTICLE

Constructing human genetic disease database in Bangladesh

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Abstract

In the advent of technological revolution, genetic interaction has become a crucial aspect in the understanding of any diseases. Information on individual genetic variation is now used in translational genomics to implement precise diagnosis and personalized medicine. Finding causal genes for genetic diseases or exploring interaction of genes in diseases with genetic predisposition is the first step to getting insights into such diseases. The human genome project made a paradigm shift in thinking, especially in the developed countries affected by non-communicable diseases. Some cutting-edge technologies, including gene therapy and genome editing, hold the promise in better diagnosis and treatment of common to rare genetic diseases. Scientific communities are trying hard to accumulate all the genetic disease information from publicly available platforms. A genetic disease database of a country serves as a depository. Many developed and a few developing countries have already developed genetic disease databases, which could benefit early diagnosis and proper patient management. Unfortunately, Bangladesh is lagging behind in this aspect. It is imperative to develop genetic disease database in Bangladesh because of its large population of patients with genetic disease. In this review, we discuss the reasons for constructing a genetic disease database and how this database can help to fight against challenges arising from the genetic diseases in Bangladesh.

Keywords: Genetic disease; Database; Bangladesh

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Citation: Akter F, Araf Y, Anwar S, *et al*., 2022, Constructing human genetic disease database in Bangladesh. *Gene Protein Dis*, 1(1): 78.
https://doi.org/10.36922/gpd.v1i1.78

Received: April 28, 2022
Accepted: June 22, 2022
Published Online: June 29, 2022

1. Introduction

Genetic disease or disorder is caused by the mutation(s) in one or more genes or anomalies in the chromosome[1]. The precise diagnosis, control, and management of a genetic disease depend on the identification of causal variants in the genome[2]. The breakthrough discovery of the human genome project allows scientists to understand the disease better. With

the advent of technological revolution and gold standard Sanger sequencing, massively parallel DNA sequencing has revolutionized the identification of novel human genetic variations. As a result, 20,442 coding genes and more than 15,000 monogenic diseases have been discovered[3]. Multi-omics and translational research also make precision medicine and gene therapy possible for many diseases[4].

On the other hand, the incidence of non-communicable diseases, including cardiovascular diseases, diabetes, and cancers with genetic predisposition, has increased alarmingly worldwide. Genetic variants in chronic disease provide a better understanding for screening, diagnosis, and precise early treatment[5]. Pharmacogenomics using genome-based tools allow for administration of precise drug doses to eliminate toxicity and improve efficacy[6]. Thus, genetic information has become a key component for efficiently diagnosing and managing any diseases.

Genetic diseases are emerging at an alarming rate in both developed and developing countries. According to the World Health Organization, about 2 – 3% of people suffered from genetic diseases globally by birth, and over 70% of these disorders are preventable[7]. It has been reported that approximately 65% of people have some health problems resulting from genetic mutations, one in 50 people is affected by a single-gene mutation, and around one in 263 people is affected by chromosomal abnormalities[8]. Remarkably, the prevalence of genetic diseases varies between populations. Thus, the population- or country-specific mutation spectrum of genetic diseases is important as a guide for controlling and properly managing genetic diseases. Unfortunately, genetic research remains neglected in low-income and lower-middle-income countries, including Bangladesh. Most developed countries and a few low-income and lower-middle-income countries have their own genetic disease databases, but a genetic disease database is currently unavailable in Bangladesh.

In this paper, we discuss the reasons for constructing a genetic disease database and review the status quo of genetic disease research in Bangladesh, which can aid in the management of genetic diseases.

2. Status of genetic disease research in Bangladesh

Bangladesh is a highly dense country with a population of 166 million people, of which 90% are Muslim, and consanguineous marriage is very prevalent in the country[9]. Inhabitants of Bangladesh are suffering from many infectious and non-infectious diseases. Among non-infectious diseases, genetic diseases and congenital malformations are quite common. Since there is neither systematic genetic testing nor well-developed expertise for counseling nationwide, most genetic diseases in Bangladesh remain undiagnosed and have become a significant health burden[10]. Through extrapolating from the worldwide prevalence of genetic diseases, it is presumed that there are many patients with genetic diseases in Bangladesh. A comprehensive literature review revealed that more than 60 case reports on genetic diseases have been published, summarized in Table 1. Only nine of the available case reports demonstrated the utilization of genetic testing (possibly done from abroad) to confirm the presence of a specific disorder. Whole-genome sequencing has been done for only four Bangladeshi individuals, revealing over 11,500 variants responsible for different diseases and 17 genetic diseases[11]. Another whole-exome sequencing study discovered the presence of pathogenic variants in other genes associated with an extremely rare genetic diseases in five unrelated patients[12].

Table 1. A detailed summary of published case reports on genetic diseases in Bangladesh.

Genetic diseases	Inheritance mode	Diagnosis procedure	Publishing year of the first case report	Reference
Achondroplasia	Autosomal dominant	(1) Medical history collection (2) Physical examination	2020	[13]
Attention-deficit hyperactivity disorder (ADHD)	Autosomal dominant	(1) Interview (2) Identification of serum level of lead (3) Significant increase of plasma ammonia and lactate	2013	[14,15]
Adrenoleukodystrophy	X-linked	(1) Medical history collection (2) Physical examination (3) Biochemical findings (4) MRI and MRS of brain (5) USG of abdomen	2010	[16-18]
Bart's syndrome	X-linked	(1) Medical history collection (2) Physical examination (3) Skin biopsy	2012	[19]

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Table 1. (*Continued*).

Genetic diseases	Inheritance mode	Diagnosis procedure	Publishing year of the first case report	Reference
Bardet-Biedl syndrome	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) Biochemical findings (4) Organ function tests (5) Genital examination (6) Hormone analysis (7) X-ray (8) EKG or ECG	2013	[20-24]
Catecholaminergic polymorphic ventricular tachycardia type 3	Autosomal recessive	(1) Medical history collection (2) Genetic testing	2021	[25]
Cutis laxa syndrome	Heterogeneous	(1) Medical history collection (2) Physical examination (3) Skin biopsy (4) Chest X-ray and echocardiogram	2010	[26,27]
Color blindness	X-linked recessive	(1) Medical history collection (2) Ishihara's test	2009	[28,29]
Cerebral creatine deficiency syndrome	Autosomal recessive or X-linked	(1) Medical history collection (2) MRI of the brain (3) EEG (4) Genetic testing	2021	[30]
Crouzon Syndrome	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) X-ray examination (4) MRI of the brain (5) CT scan of brain and skull	2020	[31]
Congenital hypotrichosis simplex	Heterogeneous	(1) Medical history collection (2) Physical examination (3) Scalp biopsy	2018	[32]
Cockayne syndrome	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) CT scan of the brain (4) EEG (5) Biochemical tests (6) X-ray (7) Genetic testing	2010	[33,34]
Crohn's disease	Single-gene mutation	Not mentioned	2010	[35]
Christmas disease	X-linked recessive	(1) Medical history collection (2) Physical examination (3) Nervous system examination (4) Coagulation factor test (5) Erythrocyte sedimentation rate (6) Hemoglobin test (7) Platelet count blood test	2009	[36]

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Table 1. (*Continued*).

Genetic diseases	Inheritance mode	Diagnosis procedure	Publishing year of the first case report	Reference
Cystic fibrosis	Single-gene mutation	(1) Answering a questionnaire (2) Medical history collection (3) X-ray (4) Biochemical test (5) Mantoux test (6) ECG (7) Echocardiography (8) Primary Immunodeficiency panel (9) Saccharine test (10) FOB (11) Stool microscopy (12) Sweat chloride test (13) Mutation analysis	2004	[37,38]
Down syndrome	Chromosomal disorder	(1) Physical examination (2) Biochemical test (3) USG (4) Karyotype test	2016	[39,40]
Darier's disease/keratosis follicularis	Autosomal dominant	(1) Medical history collection (2) Biochemical test (3) Histological examination (4) Biopsy	2020	[41]
Duchenne muscular dystrophy	X-linked recessive	(1) Medical history collection (2) Physical examination	2009	[42]
Epidermodysplasia verruciformis	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) Biopsy (4) X-ray	2011	[43]
Edward syndrome	Chromosomal disorder	(1) Medical history collection (2) Transabdominal ultrasound (3) Physical examination (4) Chromosome analysis	2012	[44,45]
Escobar syndrome	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) Evaluation by various specialized doctors (4) Spine radiography	2015	[46]
Ellis-van Creveld syndrome	Autosomal recessive	(1) Medical history collection (2) Biochemical test (3) X-ray	2016	[47]
Fraser syndrome	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) X-ray (4) USG (5) Blood tests (6) Biopsy (7) Karyotype test	2014	[48,49]

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Table 1. (*Continued*).

Genetic diseases	Inheritance mode	Diagnosis procedure	Publishing year of the first case report	Reference
Familial chylomicronemia syndrome	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) Different blood component tests (4) Coagulation factor test (5) Serum examination (6) Investigation of lipid profile (7) USG (8) Ophthalmoscopy (9) Hemoglobin test (10) Blood sugar analysis (11) Total white blood cell count (12) Investigation of liver and renal function	2015	[50,51]
Familial hypophosphatemic rickets	X-linked dominant	(1) Medical history collection (2) Physical examination (3) X-ray (4) Biochemical examination	2010	[52]
Fanconi anemia	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) Biochemical test (4) USG of abdomen (5) X-ray (6) Karyotype test	2017	[53]
Familial hypercholesterolemia	Autosomal codominant monogenic disease	(1) Medical history collection (2) Physical examination (3) Examination of the cardiovascular system (4) Biochemical test (5) X-ray (6) ECG (7) Echocardiogram	2012	[54,55]
Gaucher disease	Autosomal recessive	(1) Medical history collection (2) Biochemical test (3) X-ray (4) USG (5) Bone marrow examination (6) Leucocyte acid beta-glucocerebrosidase assay (7) Abdominal examination (8) Serum antinuclear antibody and Coombs test (9) Immunochromatographic test strip (10) Endoscopy	2009	[56-58]
Glucose-6-phosphate dehydrogenase deficiency	X-linked recessive	(1) Physical examination (2) Hemoglobin test (3) Hematocrit test (4) Reticulocyte count (5) Chest X-ray (6) Blood smear test (7) Bone marrow examination (8) Cold agglutinins test (9) Coombs test (10) G6PD assay	2017	[59]

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Table 1. (*Continued*).

Genetic diseases	Inheritance mode	Diagnosis procedure	Publishing year of the first case report	Reference
Huntington's disease	Autosomal dominant	(1) Medical history collection (2) Analysis of blood parameters (3) MRI (4) Neurological examination (5) Genetic testing	2016	[60-62]
Hemophilia	X-linked recessive	(1) Medical history collection (2) Physical examination (3) Coagulation screening test (4) X-ray (5) Biochemical tests	2006	[36,63,64]
Hajdu-Cheney syndrome	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) X-ray	2012	[65]
Hutchinson-Gilford progeria syndrome	Autosomal dominant	(1) Medical history collection (2) Biochemical test (3) Radiographic findings	2017	[66]
Hereditary spastic paraplegia	Autosomal dominant/ recessive/X-linked	(1) Medical history collection (2) Physical examination (3) MRI (4) CT scan	2013	[67]
Inborn errors of metabolism	Autosomal dominant/ recessive/X-linked	(1) Medical history collection (2) Screening of amino acid, organic acid, and fatty acid metabolism disorders by tandem mass spectrometry	2018	[68]
Kartagener's syndrome	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) X-ray (4) Electrocardiogram (5) High-resolution computed tomography	2015	[69,70]
Keratosis follicularis spinulosa decalvans	X-linked recessive	(1) Medical history collection (2) Physical examination (3) Scalp biopsy	2017	[71]
Klinefelter syndrome	Chromosomal disorder	(1) Karyotype test	2018	[72]
Larsen syndrome	Autosomal dominant/ recessive	(1) Medical history collection (2) Physical examination (3) Hematology test (4) Radiological Survey	2015	[73]
Lysosomal storage disorders	Autosomal recessive/X-linked	(1) Clinical evaluation (2) Analysis of urinary metabolites (3) Bone marrow study (4) Liver biopsy	2019	[74]
Marfan syndrome	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) Echocardiography (4) USG (5) Analysis of complete blood count, urine, serum creatinine, and random blood sugar, as well as anti-streptolysin O test (6) Chest radiography	2012	[75-77]

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Table 1. (*Continued*).

Genetic diseases	Inheritance mode	Diagnosis procedure	Publishing year of the first case report	Reference
Mitochondrial encephalomyopathy, lactic acidosis, and stroke like syndrome (MELAS)	Mitochondrial inheritance	(1) Medical history collection (2) Neurological examination (3) Funduscopic examination (4) Blood lactate test (5) USG (6) EEG (7) CT scan (8) ECG (9) Muscle biopsy	2009	[78]
Myotonic muscle disorders	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) Routine nerve conduction study (4) EMG	2008	[79]
Neurocutaneous syndromes	Autosomal dominant	Examinations of: (1) Major features: Skin, brain, and eye lesions Tumors in the heart, lungs, and kidneys (2) Minor features: Bone cysts, rectal polyps, rectal polyps, dental enamel pits, gingival fibromas, non-renal hamartomas, achromatic retinal patches, confetti skin lesions, and multiple renal cysts	2019	[80]
Noonan's Syndrome	Autosomal dominant	(1) Medical history collection (2) Blood tests (3) Routine urine test (4) IgE test (5) Echocardiogram	2009	[81]
Osteogenesis imperfecta	Autosomal dominant	(1) Medical history collection (2) X-ray	2014	[82]
Osteopetrosis	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) Blood test (4) X-ray (5) USG	1996	[83]
Pachydermoperiostosis	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) Biochemical test (4) X-ray (5) NCV test (6) MRI of the brain (7) Examination of the musculoskeletal system (8) Radiography of the limbs (9) Skin biopsy	2012	[84-86]
Peutz-Jeghers' syndrome	Autosomal dominant	(1) Medical history collection (2) Gastrointestinal examination (3) Blood test (4) OBT (5) USG (6) Upper gastrointestinal endoscopy (7) Barium meal test	2010	[87-89]
Parkinson's disease	Autosomal dominant	(1) Medical history collection (2) Physical examination	2020	[90]

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Table 1. (*Continued*).

Genetic diseases	Inheritance mode	Diagnosis procedure	Publishing year of the first case report	Reference
Porphyria cutanea tarda	Autosomal dominant	(1) Medical history collection (2) General examination (3) Routine Blood test	2013	[91]
Poland's syndrome	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) Radiological examination (4) X-ray (5) USG (6) Cardiovascular system examination (7) Alimentary system and nervous system examination (8) Echocardiography	2015	[92,93]
Retinitis pigmentosa	X-linked recessive	(1) Medical history collection (2) Neurological examination (3) Fundoscopy	2017	[62]
Spinal muscular atrophy type 3	Autosomal recessive	(1) Medical history collection (2) Nerve examination (3) Biochemical test (4) EMG (5) Muscle biopsy	2019	[94]
Thalassemia	Autosomal recessive	(1) Hemoglobin electrophoresis (2) Complete blood count analysis (3) Bilirubin analysis	2005	[95-97]
Touraine-Solente-Gole syndrome	Autosomal recessive/ dominant	(1) Physical examination (2) X-ray	2012	[98]
Turner syndrome	Autosomal dominant	(1) Physical examination (2) Karyotype test	1984	[99,100]
Treacher Collins syndrome	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) Biochemical test (4) X-ray (5) Eye examination	2008	[101]
Thrombophilia	Autosomal dominant/ recessive/X-linked	(1) Physical examination (2) CT angiography (3) Biochemical tests (4) Chest radiography (5) Echocardiography (6) Blood coagulation test	2012	[102]
Ulcerative colitis	Multifactorial disorder	(1) Medical history collection (2) Physical examination (3) Stool microscopy (4) Hemoglobin test (5) Serum albumin test (6) Blood test (7) Chest X-ray (8) USG (9) Ileocolonoscopy (10) Colonic biopsy	2013	[103]
Von Hippel-Lindau disease	Autosomal dominant	(1) Medical history collection (2) USG (3) Biochemical test (4) Antibody test (5) Gastrointestinal tract endoscopy (6) CT scan (7) MRI (8) Retinal examination	2012	[104]

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Table 1. (*Continued*).

Genetic diseases	Inheritance mode	Diagnosis procedure	Publishing year of the first case report	Reference
von Willebrand disease	Autosomal dominant/ recessive	(1) Medical history collection (2) Biochemical tests (3) Endoscopy (4) Bone marrow study (5) von Willebrand factor antigen assay	2019	[105]
Wilson's disease	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) Limb examination (4) Nerve examination (5) Slit-lamp examination (6) ECG (7) CT scan (8) MRI of the brain	2015	[106,107]
Williams syndrome	Autosomal dominant	(1) Medical history collection (2) Physical examination (3) ECG (4) Chest X-ray (5) Cardiac characterization (6) STG aortography (7) Angiography (8) Blood test	2014	[108]
Xeroderma pigmentosum	Autosomal recessive	(1) Medical history collection (2) Physical examination (3) Biochemical test (4) X-ray (5) USG (6) ECG (7) MRI of the brain (8) Skin biopsy (9) Neurological examination	2005	[109-111]

Like many other countries, the prevalence of genetic diseases is progressively increasing in the Bangladeshi population. Out of all the genetic diseases, some are ubiquitous in Bangladesh, such as thalassemia, Down syndrome, and autism spectrum disorder. The most common genetic disease in Bangladesh is beta-thalassemia, and about 7% of the population are carriers[112]. Over 2000 children are born with thalassemia every year in Bangladesh[113]. A recent nationwide carrier detection program showed that the overall ratio of beta-thalassemia carrier is 2.24%, whereas the Hb-E trait carrier rate is 8.68%. Besides, the study revealed another concerning factor by screening participants with Hb-D trait, asymptomatic Hb-E disease, suspected Hb-E, beta-thalassemia, hereditary persistence of fetal hemoglobin, and alpha-thalassemia trait, who account for 11.8% carriers with abnormal hemoglobin genes[114]. Parkinson's disease is another matter of concern as the World Health Organization announced the death rate of Parkinson's disease, which is 0.07% of total deaths in Bangladesh in 2017. The number of deceased individuals is increasing day by day[115]. Surprisingly, the death rate jumped to 0.18% of total deaths in 2018[116]. Different studies have been performed to determine the prevalence rate of autism spectrum disorder in Bangladesh. A national survey in 2013 found that the frequency was 1.55/1000 and 0.68/1000 in different conditions, whereas another cross-sectional study in 2018 found that the prevalence was 0.75/1000 in rural areas[117]. The Autistic Children's Welfare Foundation presumed that the number of patients with autism spectrum disorder in Bangladesh could be around 300,000, which indicates the significance of taking required steps nationwide to prevent unfortunate consequences[118]. About 1.3 – 1.5 million cancer patients in Bangladesh and thousands of new patients are diagnosed yearly[119]. Genetic changes cause most cancers, and nearly 5 – 10% of all cancers are inherited from parents[119]. Another common genetic disease in this country is cystic fibrosis, caused by mutations in the CF transmembrane conductance regulator protein-coding gene. About one in 2500 babies is born with cystic fibrosis in the United Kingdom, whereas the ratio is 1:31,000 in Asia region[38]. Although many genetic diseases have already been reported in Bangladesh, their prevalence

data, regional distribution, and mutation profiling are still unknown[120]. In addition, the number of people diagnosed with genetic diseases, such as attention-deficit hyperactivity disorder, Down syndrome, Turner syndrome, and Klinefelter syndrome, increases[120]. Besides, nearly eight in 1000 live births in this country suffer from congenital heart defects[7].

3. The reasons for constructing a human genetic disease database in Bangladesh

Along with the technological advancement in disease identification, distinguishing and reserving the genomic variation such as mutations in different parts of the genome are of great help to control and manage the adverse outcomes of any specific diseases. All genomic data must be deposited in a particular location. That is why different genetic databases have already been constructed, such as Online Mendelian Inheritance of Man (OMIM), Human Gene Mutation Database, and Locus-Specific Databases. Genetic databases gather the molecular genetic data, standardized clinical data, and data regarding associated factors of an individual for the interpretation of gene function, the identification of genes that are mostly present in a specific community, and the differentiation from other communities to estimate the underlying cause of genetic disease[121]. The broad spectrum of genetic diversity among the people of different countries is the fundamental cause of a diverse array of genetic diseases. Genetic databases help elucidate gene function, estimate the prevalence of genes in populations, distinguish between subtypes of diseases, trace how genes may predispose to or protect against illnesses, and improve medical intervention[121]. Therefore, a nation-specific genetic disease database should contain information about the diseases' frequency rates and provide clues on the required steps to prevent the spread of the diseases from generation to generation. On realizing the significance, many countries have introduced national databases containing mutation details. However, Asian countries were lagging behind in constructing genetic databases compared to the European countries. To encourage genetic data deposition from Asian countries, the Genome Asia 100K Project has been introduced to assemble population-specific variation data and extend the genome-wide association studies. The GAsP database has already taken DNA sequences from India, Malaysia, Korea, Pakistan, China, and many other Asian countries from their genetic disease databases[122]. Despite having published case reports on more than 60 genetic diseases, Bangladesh still has not constructed a genetic disease database to collate genetic disease and mutation details. Therefore, the prevalence data and the mutation profiles of most genetic diseases are still unknown, making the diagnosis procedure burdensome[120,123].

Moreover, the curriculum for a primary medical degree (MBBS) does not cover adequate knowledge in genetics, so medical professionals in rural areas may have limited knowledge and access to the new technologies for detecting a genetic disease[10]. A genetic disease database should also contain all the possible symptoms of the diseases. The information regarding the possible diagnosis procedures should be added as well. The health-care system faces different problems while managing inherited diseases because of the complexity and heterogeneity of the clinical data. Establishing a genetic disease database will allow extensive access to all the related features of the disease and genetic mutation data that will be beneficial for the precise diagnosis and treatment of patients with genetic diseases. These genetic disease databases serve as a platform to educate health-care professionals, scientists, patients, and the common people of Bangladesh about different genetic diseases and the available treatments.

Moreover, South Asia has the most ethnically diverse population and therefore significant genetic variability compared to the other parts of the world[113]. Comparing the clinical features of South Asian people with those in the databases of other countries can lead to a false-positive or -negative result because of incompatible mutation patterns. Thus, having a population-specific genetic diseases database with specific mutations can ease the analysis of genetic differences between ethnic groups, trace population diversity, and disease susceptibility. Furthermore, the database will lend a helping hand to the research groups working on human genetic diversity, medical and evolutionary history of ethnic groups, genetic disease diagnosis, and treatment by supplying data while designing study and during result interpretation. Understanding genotype-phenotype correlation, developing molecular diagnostic tests, analyzing mutation, and genetic counseling will be easier with the help of a genetic disease database because all relevant information are in one place. In addition, the database would be helpful for other researchers to analyze specific Asian mutations. Pharmacogeneticists will find the database highly advantageous after utilizing it to understand gene-related variabilities in drug responsiveness and metabolism, thereby facilitating drug screening in accordance with genetic susceptibility before prescribing.

4. Genetic disease database of different Asian countries

The diversity of genetic diseases in different populations depends on geographical location, reproductive practices, and environmental factors. Thus, many neighboring Asian countries have already established their own national genetic disease databases, which are summarized in Table 2.

Table 2. Asian countries and regions having individual genetic disease database.

Country/Region	Name of the database	Link	Reference
Bahrain	National Genome Center (Bio-Bank)	https://www.moh.gov.bh/	[124]
China	China National GeneBank	https://ngdc.cncb.ac.cn/databases	[125]
Dubai	Dubai Genomics	https://www.dha.gov.ae/en	[126]
India	Indian Genetic Disease Database (IGDD)	http://www.igdd.iicb.res.in/	[2]
Indonesia	Genomic Medicine Research Group Database (GMRGDB)	Not available	[127]
Iran	Iranian Human Mutation Gene Bank	www.IHMGB.com	[128]
Israel	Israeli National and Ethnic Mutation Database	http://server.goldenhelix.org/israeli	[129]
Japan	Medical Genomics Japan Variant Database (MGeND)	https://mgend.med.kyoto-u.ac.jp/	[130]
Korea	Korean Mutation Database	http://kmd.cdc.go.kr	[131]
Malaysia	Malaysian Node of the Human Variome Project database (MyHVPDb)	http://hvpmalaysia.kk.usm.my/	[132]
Pakistan	Pakistan Genetic Mutation Database	http://pakmutation.kust.edu.pk/	[133]
Qatar	Qatar Biobank	https://www.qatargenome.org.qa/	[134]
Singapore	The Singapore Human Mutation and Polymorphism Database	http://shmpd.bii.a-star.edu.sg/	[135]
Sri Lanka	Sri Lankan Genome Variation Database	http://www.hgucolombo.org/	[136]
Taiwan	Taiwan Biobank	https://www.twbiobank.org.tw/new_web_en/	[137]
Thailand	Thailand mutation and variation database (ThaiMUT)	http://gi.biotec.or.th/thaimut	[138]
Turkey	Turkish Human Mutation Database	http://hmut-tr.sourceforge.net/	[139]
Vietnam	Vietnamese Genome Research Project	https://www.vinmec.com/en/tin-tuc/hoat-dong-benh-vien/vinmec-announced-its-largest-vietnamese-genome-research-project/	[140]

5. Roadmap to generate a genetic disease database for Bangladesh

Data from authentic sources are a prerequisite for establishing a genetic disease database because the database should consist of details regarding specific genetic mutations, associated protein function alteration, and mutational statistics of different regions. To collect the data, databases such as PubMed, NCBI, OMIM, GeneCard, KEGG, and UniProt will be utilized. Details regarding specific genetic diseases that are scientifically accepted and observed can be obtained from PubMed and NCBI, whereas OMIM, GeneCard, KEGG, and UniProt give a clear dimension of human genes and associated genetic diseases depending on different traits, protein sequences, and functions, along with the correlation between genomic difference and phenotypic expression. Detailed information from all these authentic databases can display an approximate result for every individual when the person has a certain genetic mutation. Obtaining mutational analysis results beforehand will be beneficial in terms of taking adequate treatment and preventive measures to reduce the chance of disease development. Certain strategies must be considered when making the genetic disease database compatible for captive utilization. The entire procedure of constructing the database can be divided into two broad categories: Data collection and data curation. The overall plan is shown in Figure 1.

5.1. Data collection

The primary data can be obtained from peer-reviewed published articles in various sources, including PubMed and Google Scholar. Signs and symptoms can be added to the database for use by physicians, researchers, and patients. Taking consent from the correspondence before incorporating the data is necessary to determine the accuracy of data and eliminate any further inconvenience. In addition, the user submission option should always be kept open to include all new mutations, signs and symptoms, diagnoses, and treatment procedures and to update the database with helpful information about genetic diseases in Bangladesh. Other sources can be linked, including NCBI, OMIM, KEGG, and PDB, to expand the pieces of information in the database. Geographical

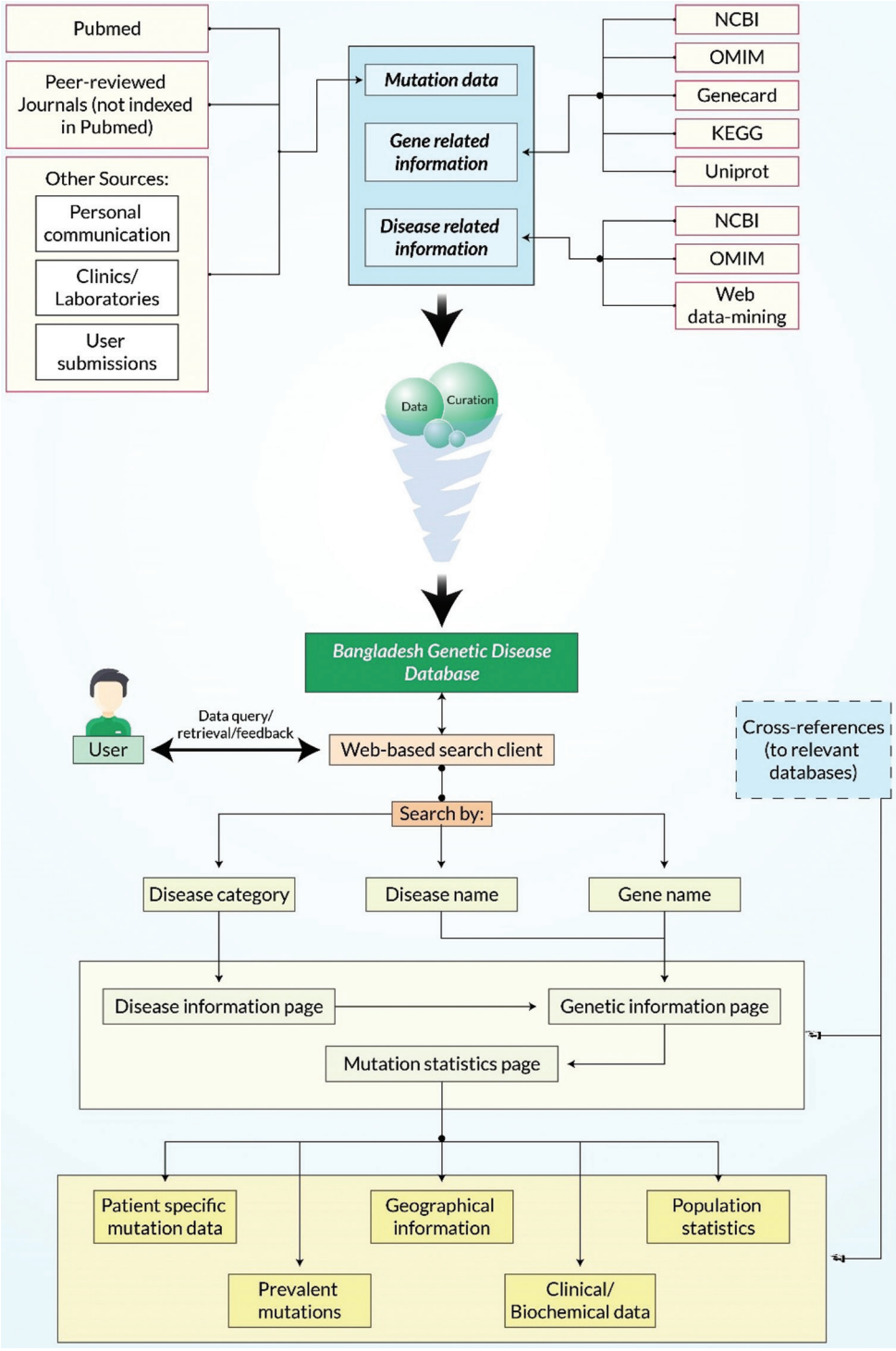

Figure 1. Schematic representation of the construction of proposed Bangladesh Genetic Disease Database.

information, which is vital to researchers to focus on the hotspot genetic disease in a specific community, should be incorporated. Next, the most crucial factor must be included in identifying the causal gene's location in the chromosome and the accession of nucleotide and protein. Moreover, patient-specific mutation data and symptoms, diagnosis, treatment, and management should also be included.

5.2. Data curation

The database should be updated regularly. Any uploaded mutation information will be cross-checked and filtered before incorporated into the database. Inserting data to a particular place are required, and *in vitro*, *in vivo*, or *in silico* characteristics of the variant will be helpful in this regard, along with differentiating its novelty from others. Mutation data errors will be corrected, modified, or updated whenever needed to maintain accuracy. Data without proper evidence will be removed. In addition, all the data need to be transferred in a single format as per Human Genome Variation Society's recommendation to make the format standardized.

6. Conclusion

The burden of non-communicable diseases increases dramatically throughout the world. Although the genetic data from Bangladesh are scanty, the worldwide prevalence rate of genetic diseases and the 60 published articles found in online sources indicate that Bangladeshi population could be affected by many genetic diseases. A comprehensive genetic disease database will be helpful to provide an insight into the current status of genetic diseases, research, diagnostics, awareness, treatment, and proper patient management. Moreover, a nation-specific genetic disease database would definitely help with strengthening the health-care system.

Acknowledgments

The authors acknowledge the members of the Community of Biotechnology, Dhaka, Bangladesh, for their support during the manuscript preparation.

Funding

The authors received no funding from external sources.

Conflict of interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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