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Congenital Disorder in the Fetal Development of a Live Birth using Rule Based Inference

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ABSTRACT

Congenital disorder is a condition existing at or before birth regardless of cause. Of these diseases, those characterized by structural deformities are termed "congenital anomalies" and involve defects in a developing fetus. Birth defects vary widely in cause and symptoms. Any substance that causes birth defects is known as a teratogen. Some disorders can be detected before birth through prenatal diagnosis It is caused due to some genetically disorder, environmental or drug exposure. The aim of the paper is to find the proportion of the population of congenital disorder. The inference engine helps in finding the new conclusion referencing to knowledge base and conclude using inference engine. The proposed system looks after the infant and the maternal information and draws some conclusion about the occurrence of the congenital disorder. This type of experiments should be conducted all over the world so that the death dates of infants can be reduced and proper medications and care can be taken in order to prevent disorders in prior.

KEYWORDS: *Congenital Disorder, Association rules, data mining, expert system*

I. INTRODUCTION

A genetic disorder is a genetic problem caused by one or more abnormalities in the genome, especially a condition that is present from birth (congenital). Most genetic disorders are quite rare and affect one person in every several thousands or millions.

Genetic disorders may be hereditary, passed down from the parents' genes. In other genetic disorders, defects may be caused by new mutations or changes to the DNA. In such cases, the defect will only be passed down if it occurs in the germ line. The same disease, such as some forms of cancer, may be caused by an inherited genetic condition in some people, by new mutations in other people, and mainly by environmental causes in other people. Whether, when and to what extent a person with the genetic defect or abnormality will actually suffer from the disease is almost always affected by the environmental factors and events in the person's development.

There is huge amount of information in the datasets at the health care organizations that can be extracted using data mining techniques. The main aim of this study is to extract the data and patterns from these large datasets and bring it into a form that is useful for medical practitioners for better diagnosis.

The main objective of the project is to prediction of congenital malformation in live births. Medical Data mining is finding significant useful pattern from a large bio-medical dataset. Data mining utilizes this information and patterns to build stronger predictive model. Any medical reason for the congenital malformations in the newborns at the time of birth. The only factors that were taken into consideration in these studies were only the social factors that could cause of these anomalies. The prototype expert system was testing using the testing dataset to obtain the accuracy of the system.

Section II of the paper gives a brief overview of work done by researchers regarding congenital malformations. Exsisting system is explained in section III. Proposed framework for the inference engine is explained in section IV and section V inference engine is explained in detailed .

II. Literature Survey

Prevalence of Congenital Malformations

EUROCAT (European Surveillance of Congenital Anomalies) is the network of population-based registers of congenital anomaly in Europe, with a common protocol and data quality review, covering 1.5 million annual births in 22 countries. EUROCAT recorded a total prevalence of major congenital anomalies of 23.9 per 1,000 births for 2003-2007. 80% were livebirths. 2.5% of livebirths with congenital anomaly died in the first week of life. 2.0% were stillbirths or fetal deaths from 20 weeks gestation. 17.6% of all cases were terminations of pregnancy following prenatal diagnosis (TOPFA). Thus, congenital anomalies overwhelmingly concern children surviving the early neonatal period, who have important medical, social or educational needs. The prevalence of chromosomal anomalies was 3.6 per 1,000 births, contributing 28% of stillbirths/fetal deaths from 20 weeks gestation with congenital anomaly, and 48% of all TOPFA. Congenital heart defects (CHD) were the most common non-chromosomal subgroup, at 6.5 per 1,000 births, followed by limb defects (3.8 per 1,000), anomalies of urinary system (3.1 per 1,000) and nervous system defects (2.3 per 1,000). In 2004, perinatal mortality associated with congenital anomaly was 0.93 per 1,000 births, and TOPFA 4.4 per 1,000 births, with considerable country variation. Primary prevention of congenital anomalies in the population based on controlling environmental risk factors is a crucial policy priority, including preconceptional care and whole population approaches.

III. Existing System

Our system may be viewed as consisting of three basic components: a user-interface, a set of rules in knowledgebase and an interpreter for the rules an inference engine. User interface is required to do a little more than capturing user inputs. It is also required to transform user-input in form suitable for inference-engine to work with. It is also required to transform inference-engine output in a language suitable for user fto understand or deduce.

IV. Proposed System

Data Mining Framework

A compact biomedical data framework based on preprocessed data gathered from Questionnaires.

Three Important phases:

1) Preparing the data
2) Pre-processing the data
3) Mining of medical data

Preparing the data:

This research is based on real time data in the form of questionnaires Hospital.

Stages in data preparation are:

- Adding or assuming the data
- fetching the important data necessary mining
- Changing the data in more unified format

Pre-processing the data:

Structured data undergoes various transformation in this step. Data cleaning, Removal of duplicates, discretization done to bring data in to a form that can be used by mining algorithms. Data Conversion is accomplished in this stage. Denormalizing in above way yields 38 regular attributes. This data is then entered in to our SQL database based upon the binned attributes in the form of binomial values. The values entered is 1 or 0 so that it can be used by mining algorithms

Mining of medical data

Association Rules: To discover associations between the discrete data that might be helpful in establishing inferences about other attributes. Dataset consists of continuous numerical and categorical attributes so discretize by binning and rehashing technique is used to convert that into binomial.

V. System Architecture

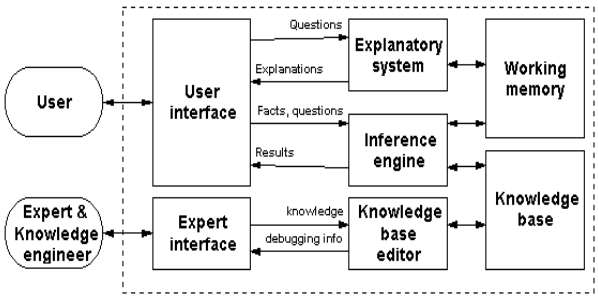

Fig1: System architecture

The figure shows the interaction of the user with the system. The System here has working memory and the knowledge base.

Knowledge base stores all the details and the attributes and the details about the user. Working memory keep track of the information given by the patient and explanatory system explains why the working memory is particularly selecting the particular attribute.

Rule Based Inference Engine

Like typical expert system our designed expert system will also be composed of three primary components: an inference engine, the knowledge base and the IO interface. The primary purpose of our expert-system is the validation of production-rules. It is required to be very problem specific; therefore, architectural complications owing to generalization of usage will be averted.

Our system may be viewed as consisting of three basic components: a user-interface, a set of rules in knowledgebase and an interpreter for the rules ___ an inference engine. User interface is required to do a little more than capturing user inputs. It is also required to transform user-input in form suitable for inference-engine to work with. It is also required to transform inference-engine output in a language suitable for user to understand or deduce

***User Interface*:**

User interface of our system is required to capture input from user against following attributes. User interface is also required to translate the input suitable for engine to interpret and apply on set of production rules.

Knowledgebase:

Knowledge base is for record keeping of the rules. It is a simple database which our engine will access for production rules. When a rule is discovered it is appended in knowledgebase

***Inference Engine*:**

Inference engine is the heart of the expert-system. It tracks the ordering of rules in which they are allowed to fire. For firing-table rules are ordered w.r.t. accuracy of the rule. Following this order in firing-tables will ensure that most specific rule fires first ___ greater precision strategy. If no rule qualifies firing then inference engine will regret prediction

A. Modelling Association Rules

Denormalizing fields yields 38 regular attributes. All attributes are recorded as either a one or zero (integer attribute) in the excel sheet. The data was entered in to the Microsoft SQL Server 2008 database table in the form of 1 and 0 for generating production rules.

The data was then analyzed using SQL queries to create the production rules.

B. Expert System

We have established our actual requirements and carried-out the knowledge acquisition. Now we are ready for constructing our expert-system components. Expert System is developed as a java desktop application and using the local database of net beans for storing the production rules.

Fig 2: Prototype of Expert System

The result of the prediction was matched with the result of the testing dataset provided by the doctors at. From the current record it was observed that the baby born to the women had congenital malformation thus validating our expert system predictions.

VI. RESULTS

The figure shows the system which predicted the congenital disorder percentage with the criteria given by the patient.

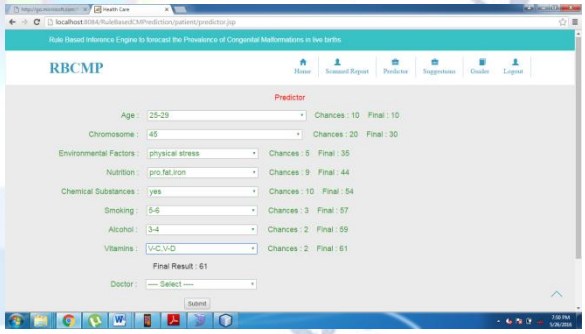

Fig 3: Prototype Expert System predicting malformation in newborn

The figure shows the scanned report generated and forwarded to the patient.

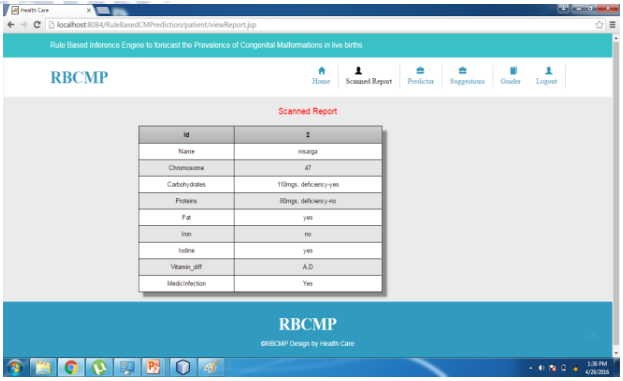

Fig 4: Scanned report

VII. Conclusion

On the detailed analysis of the association rules conclusions are drawn that would help in prediction of congenital malformation in live births. Some of the conclusions are stated below:

- Those females whose weight is greater than 65 and hemoglobin less than 10 will have 92% chances of having a child who will have a congenital malformation.
- Females having hemoglobin greater than 10 but belonging to a rural area will have 90% chances of having a child with congenital malformation
- It has been observed that there are 80% chances that those females whose parents are distantly related biologically (2nd cousin) will have a child who has some congenital malformation

Females having weight greater than 65 and level of sugar in blood not in the normal range will have chances of having a baby with congenital malformation

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