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EVALUATION IMPACT IN PATTERN OF DISEASE RECURRANCES AND TREATMENT IN GENETIC ENDOCRINE LOPSTIC SURGERY IN DIFFERNTIATED CLAUSES

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Abstract:

Aim: *This study aims to formulate a specific arrangement for carrying out the clinical genetic testing in a Molecular diagnostic laboratory with the ultimate goal of researching aggressions in genetic testing in endocrine surgery with opportunities for precision surgery.*

Technique: *The study had included about nine MEN-1 individuals. In the research laboratory, the individuals were grouped according to their type of disorder, age, and number. Mutations were firstly identified for each group. There were about 56 individuals tested, and then parathyroidectomy was carried out in the patients with hyperthyroidism. Every patient in the research got proper and detailed genetic consultation.*

Results: *The genetic testing was carried out in 56 patients at risk. Patients were divided into three respective groups. Group, I contained about 25 patients with a risk of 50 percent. These were younger than 30 years. Group II included about 20 patients, with a chance of 50 percent. These were aged 30 or older.*

On the other hand, Group III contained about 11 patients with a risk of 25 percent. There was a positive genetic test obtained in about seven patients. Six belonged to group 1, and once belonged to group 2. Hypercalcemia was diagnosed in these patients. Moreover, parathyroidectomy was also carried out in these patients. One patient was also having the symptoms of the pancreatic tail mass lesion and pancreatic polypeptide level.

Conclusion: *Mostly, endocrine disorders have an inherited origin. Also, most are syndromic. Therefore, the surgeons must have a sound knowledge of the disease, its causes, and its genetic background. In this way, they will be able to save their time, cost, and carry out excellent results.*

Keywords: *Genetic Testing, Endocrine Surgery, Precision Surgery*

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INTRODUCTION:

The progress in genetics has enabled us to understand the underlying pathogenesis, the diagnosis, and the treatment of various diseases. In 2003, geneticists sequenced the entire human genome to formulate a gene map of the human. This cost about 3 billion dollars and 13 years of continuous hard work to complete it. This cost and hard work paved the way for "precision medicine. " Precision medicine involves disease management, which depends on the variability of the patient's lifestyle and environment (1).

Moreover, precision surgery is carried out with the accuracy and precision of senior surgeons. These precision surgeries are widely used during many endocrine operations, such as parathyroidectomy, pancreatectomy, and various others(2)(3). In this study, we conducted a systematic program on a wide-ranging number of MEN 1 families for carrying out clinical genetic testing. This will ultimately enable researchers to study the aggressions in genetic testing in endocrine surgery with opportunities for precision surgery. The major part of this research is based on the translation of molecular genetic results into an approved Molecular Diagnostic Laboratory. Afterward, these structured genetic therapy systems will be implemented on the selected patients. For this purpose, the biochemical screening will be done on genetically positive patients. In this way, we will get a first idea about the presence of a tumor and surgical interventions(4)(5).

METHODOLOGY:

For this research, we considered nine Multiple Endocrine Neoplasia 1, comprising a total of 921 individuals. Out of these, 63 people were affected, 411 were at 50% genetic risk. The Multiple Endocrine Neoplasia Program followed this. The 411 individuals with 50% were older than thirty years. They have no specific clinical signs of this disease, and therefore it is not feasible for them to be affected and or reside away from the location of the experimentation.

Consequently, they will have significant issues complying with all the visits needed for genetic counseling, consent taking, taking a blood sample, and then reporting the results. To carry out this translational analysis and the research, we have well-defined the entire sample population into subcategories that are most closely followed by our ongoing research program. In the research laboratory, the genetic mutations were initially identified for each individual taken for the sample. Initially, DNA was selected and then isolated from the peripheral blood specimens. It was collected in special ACD vacutainer tubes. Optimum temperature is maintained for experimenting. For this purpose, the particular Pure gene system is used.

Furthermore, PCR is meant to amplify DNA fragments with the primers and the Taq PCR kit. Moreover, Before direct sequencing of the DNA, Big Dye chemistry is used to extract the nucleotides and the unincorporated primers. These products of PCR were processed by a specialized via Wizard columns. Afterward, the sequence of DNA reactions was analyzed. Furthermore, SacI was used for the digestion of 10 µg of genomic DNA by SacI, and the electrophoresis method was used to isolate 1 percent agarose. Afterward, these fragments were transferred to nylon membranes and then hybridized(6)(7).

RESULTS:

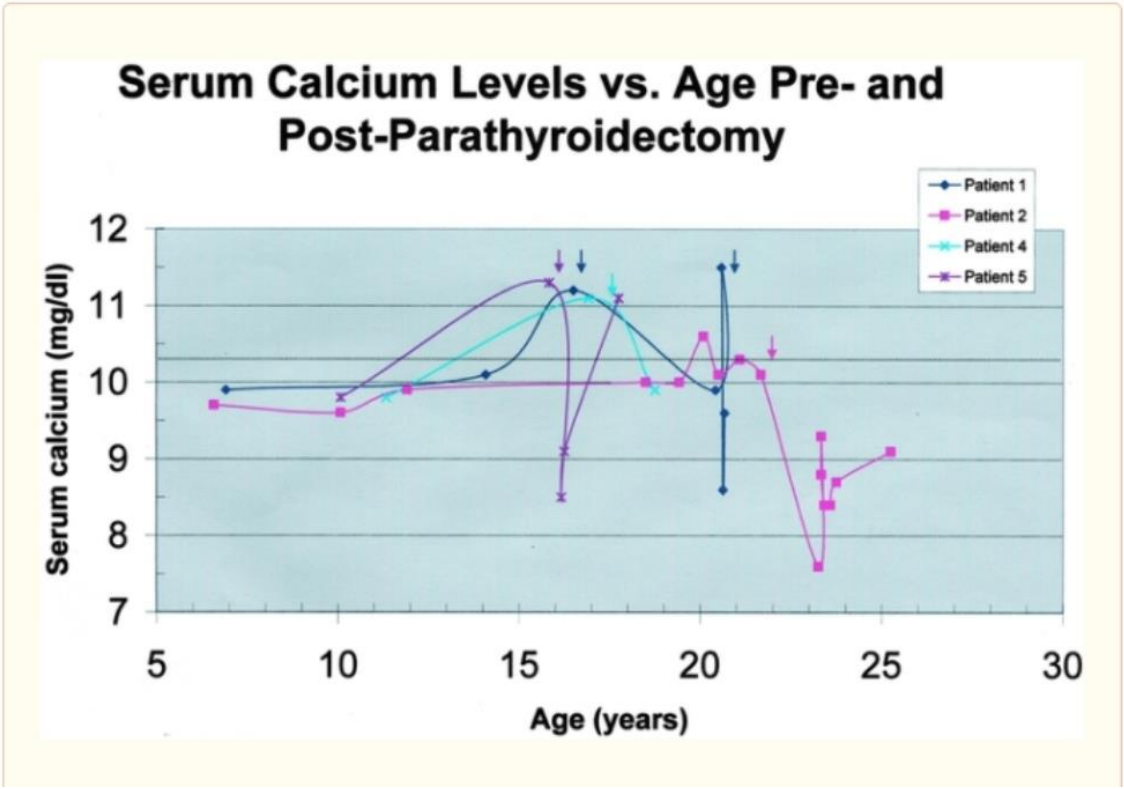
Genetic testing was carried out in fifty-six patients. These were divided into groups. Following was the data recorded:

GROUPS	NUMBER	AGE	RISK
Group 1	25	Younger than 30 years	50%
Group 2	20	Above 30 years	50%
Group 3	11	Random	25%

A total of seven patients had a positive genetic test with ages 12-42 years. Out of these, six belong to group 1, and 1 belong to group 2.

Patients tested positive	Group 1	Group 2	Ages
7	6	1	12-42 years

These patients had hypercalcemia present wither at the time of diagnosis or after the diagnosis that is during the period of surgery. A total of 4 patients underwent parathyroidectomy with the age of 16 years. On the other hand, one patient with a positive genetic test was not identified with hyperparathyroidism. A 15-year-old male who was asymptomatic and normocalcemic was recognized as having increased pancreatic tail mass lesion and pancreatic polypeptide level.



DISCUSSION:

Over the past few decades, innovation in genetics and molecular engineering has led to the development of many new diagnostic techniques and rapid advancement in the genomic and precision medicine. Also, these researches enabled geneticists to work on the application for its implementation. The classification and determination of numerous genetic problems, diseases, and susceptibility genes in endocrine surgical disorders are rapidly increasing and becoming relevant to patient care(8)(9). Most of the endocrine disorders are inherited and illustrates a considerable proportion of disease encountered by the surgeons. Therefore, genetic testing has become an integral part of the identification and treatment of patients with endocrine surgical disorders. They put emphasis mainly on the clinical use of these genetic testings and further its impacts on the surgical management of endocrine diseases (4).

CONCLUSION:

The innovations and development in genetics have paved the way for progress in this field. It introduced many ideas and treatments, as well. Doctors treat many diseases by taking help from the gene map and genetic testings. Furthermore, it enabled doctors to introduce precision medicines and precision surgeries. Genetic testing is done for figuring out and treating many endocrine disorders as well as their surgical treatment, such as parathyroidectomy, pancreatectomy, and other endocrine tumors. In this research, we experimented with the Molecular Diagnostic Laboratory to carry out genetic testing. Afterward, these structured genetic therapy systems will be implemented on the selected patients. For this purpose, the biochemical screening will be done on genetically positive patients. About seven patients were tested positive for genetic testing. These belong to the age group between 12 to 42 years.

Furthermore, they had hypercalcemia. One patient was also having pancreatic tail mass lesions and pancreatic polypeptide levels. Hence we can say that advancement in genomics has led to the introduction to the next-generation sequencing and targeted disease-specific gene sequencing, which further has made it possible to test susceptibility genes more rapidly, cost-effectively, and with precision. In conclusion, surgeons must have a sound knowledge of various diseases and genetic predisposition regarding endocrine disorders that are mostly encountered. This will allow precision surgery and is likely to provide betterment in future cases.

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