GENETIC DISORDERS AND ITS PREVENTION
–AN AYURVEDIC PERSPECTIVE

1Dr Girija Devi M, 2Dr Shyny Thankachan, 3Dr Dayana H, 4Dr Bhagavan G Kulkarni

1Associate professor and Head of the Department, 2PhD Scholar, 3Assistant Professor, 4Professor & H.O.D
1Department of Kriya Shareera,
1Pankajakasthuri Ayurveda Medical College and PG Centre, Kattakkada, Trivandrum, India. Pin: 695 572.

Abstract

Genetics is one of the most interesting topics of research in medical science. This science of heredity has got an elaborate description in the text books of Ayurveda. Modern genetics deals with the combination of maternal and paternal genes as the responsible factors for an offspring. Genetics explains how the gametes combine with each other, and how a specific gene gets stimulated. But books in Ayurveda deals much more than this. Ayurveda has explained the role of different factors favouring this development such as -Matruja (maternal), Pitruja (paternal), Atmaja (obtained from soul), Satvaja (obtained from mind), Rasaja (obtained from nourishment), Satymaja (obtained from habituation) which explains that the child born to a couple is not merely a blend of parental and maternal factors alone, but an amalgamation of many more factors. Ayurveda is conceptually well equipped to explain that the constitutional makes up of a progeny can be decided by scientific methods in line with the present-day Genetic Engineering concepts. Proper following of Rtukalaniyamas (~Do’s and don’ts at the time of Rtukala), avoiding Garbhopaghatarakabhavas (~Factors hindering the growth of fetus), following of Masanumasika Garbhini Paricharya (~Month wise regimens for the pregnant women) are few of them. Ayurveda advises pre-conceptional Shodhana upachara and proper diet for parents to protect the progeny from congenital disorders. The above said ideas can be implemented in fields like infertility management, preventing habitual abortions and certain hereditary diseases.

Current study is a conceptual work regarding the effect of Aharavihara ~(Lifestyle) of mother before and after conception in terms of Beejadosha and modifications in the current day life style to give birth to a healthy offspring.

Key words: Genetics, Genetic disorders, Beeja, Beejadosha, Lifestyle modifications.

INTRODUCTION

Science of Ayurveda has a potential to become leader in developments of future modern medicine. We are in frantic search of the most interesting aspect of genetics in our system of medicine. It is a branch of science that deals with the studies of factors responsible for transmission of similarities and dissimilarities from parents to offspring. Ancient Ayurvedic text books describe “Powerful actions of present life will even overcome the effects of actions of the previous lives.”[1] These words make us search for the seeds of genetics in Ayurveda which needs a deep insight through literatures. This science of heredity has got an elaborate description in the textbooks of Ayurveda. Concept of Beeja, Maturaharavihara, different factors affecting the offspring have always been an enigma to the researchers of Ayurveda and modern medicine. Data reveals that 3-5% of all births result in congenital malformations. 20-30% of all infant deaths are due to genetic disorders and 30-50% of all post-neonatal deaths are due to congenital malformations. As a whole 60% of the birth defects do not have any known cause. Chromosomal defects account for about 6% of the all-birth defects. Multifactorial inheritance (Genetics & environmental) accounts for approximately 20% of congenital anomalies. Teratogens cause about 4-5% of all birth defect.[2] This is an indication that the unknown in this field is far greater than the known.

The current study was undertaken keeping in mind the dearth of link between congenital disorders and their aetiology. Since Ayurveda has rich descriptions in the concept of Beejadosha and Maturaharavihara the present study aims at contributing holistically in this respect.
CONCEPT OF BEEJA & BEEJA DOSHA

Vedas, Brahmaṇa Grantas, Upanishads and Smritis gives us the reference of planning of healthy progeny starts even before marriage. The girl coming from a family suffering from congenital diseases was considered as unfit for marriage because she can be a carrier if not suffering from disease. In Charaka Samhita there are references about Beeja. The anomaly of body parts depends upon the affected portion of Beeja, Beejabhaga, Beejabhagavayava. If parents have certain diseases by the imbalance of vata, pitta or kapha then it is reflected in the Beejabhagavayava, and hence can cause illness of the offspring. Charaka says that an organic abnormality cannot occur without abnormality of the corresponding portion of Beeja. For example if portion of Beeja of a person suffering from Kushta has defect in the region which is responsible for skin formation it may produce Kushta in the born child, but if that portion is normal then child will be healthy. Same will happen in child whose father is blind. Chakrapani has also explained the role of Beeja,Beejabhaga,Beejabhagavayava in detail.

Congenital anomalies are explained by Susrutha under the heading of Adiblapravritta and Jannabalarpravritta Vyadhis. It is evident from Sushruta Samhita that Adiblapravrutta diseases are a groups of illnesses which are attributed to defects inherent in either the Shukra (the male reproductive element) or Shonita (the female reproductive element). Ex are Kushta, Arshas, Meha etc. There are four factors responsible in the formation of Garbha. They are Matruja, Pitruja, Aharaja and Atmakarmaja. The subtle form of four Mahabhutas viz. Prithvi, Jala, Agni and Vayu are present in each factors. A progeny will have characters according to the predominance of Mahabhutas as well as the deeds done by parents as well as him during the previous life also owing to the influence of diet. Mana is also influenced by previous life. According to Ayurveda, there are 6 factors which are individually responsible for Garbhotpatti These are as Matruja, Pitruja, Atmaja, Satvaja, Rasaaja, Satymaja. As a whole source of genetic material can be concluded as four Bhutas and four Bhavas [Matruja, Pitruja, Rasaaja and Atmaja]. Satmyata factor can be explained as a preconceptional factor. Satwa explained as emerging way of mental material for which Jeevatma is responsible.

In brief congenital disorders can be explained under Beeja Dosha- (Involving Beeja as a whole, and also Beejabhagavayava), Atmakarmaja Dosha (personal deeds of previous birth), Ashaya Dosha (Abnormality in the uterus), Kala Dosha(Environmental factor), Maturaharavihara Dosha(Errors in diet and regimen of mother).

A BIRDVIEW ON GENETIC DISEASES

Gregor Johann Mendel is often called the father of genetics. From his experiments, Mendel concluded that certain factors must produce certain traits which are carried from parents to offspring. Today, Mendel’s factors are called genes. Congenital anatomic anomalies, birth defects and congenital malformations are terms currently used to describe developmental disorders present at birth. Birth defects are the leading cause of infant mortality and may be structural, functional, metabolic, behavioural or hereditary.

Genetic disorders: Single gene disorders may be autosomal (not sex linked) or X-linked (related to the sex chromosome). Another common classification for genetic disorders is dominant and recessive. Some common genetic disorder classifications are a) autosomal dominant disorder inherited from one parent who is affected b) autosomal recessive disorder when both parents carry the trait and the offspring receives the non-functional gene from each parent c) an X-linked recessive disorder which typically affect males.

Chromosome disorder: A chromosomal disorder may be inherited or may be sporadic, meaning there is no family history. The most common types of chromosomal disorders can be classified as numerical and structural. Numerical includes non-disjunction resulting in a cell with an imbalance of chromosomes ex; Trisomy 21, Trisomy18 etc. In structural chromosome breakage followed by reconstitution in an abnormal combination which can have the influence of environmental factors to a great extent. Ex: Deletion. (Ex: -Cri du Chat syndrome in which a portion of chromosome 5 is deleted), Inversion (Chromosome that has broken and the broken piece turns upside down and reattaches itself. Inversions may or may not cause birth defects depending on their exact structure) and translocation. (A rearrangement of a chromosomal segment from one location to another). Another classification explained in modern text books of embryology is Malformation, Disruption, Deformation, Dysplasia.
ROLE OF DIET, HABIT, MEDICINE ON GENETIC DISORDERS

Diet: Diet plays a major role in human health especially in pregnancy. Mother’s diet has a major role in determining health of offspring. Early nutritional changes in mother effect epigenetic modifications which can pass to offspring and also down across the generations.[15] The early development can have long term effects. High fat diet, vitamin & folate deficiencies, trigger epigenetic changes in the fetus and child born will be having obese may be associated with cardiovascular disorders.[15] Low protein diet prior to conception and during early days of fetal development can increase the chances of HTN, Arterial disorders and metabolic disorders.[15] Ayurvedic texts have much illustrative description of method of fetal nourishment. Fetus gets its nourishment from Rasa supplied by mother by the process of Upasneha and Upasweda.[16]

Environmental Causes: Other forms of prenatal brain damage have an environmental basis Ex; teratogens. Susceptibility to the teratogen depends on the dose and timing of the exposure. If exposure occurs close to conception, it generally has an all-or-none effect. At the other extreme, late in the pregnancy, the teratogen may affect the size of the fetus or may precipitate premature birth but will not cause a birth defect. The most damage is likely to be done by teratogens in the first trimester, when the body organs are forming. X-rays and UV radiation exposure, chemicals (mutation agents) exposure are other examples.

Substance Abuse: The drug most commonly associated with fetal malformations is alcohol. Approximately one third of women with alcoholism give birth to babies who have a spectrum of impairments called fetal alcohol syndrome[17] which may include mental retardation or learning and emotional disabilities, deformed limbs, a small head, and congenital heart defects. Cocaine and methamphetamine use during pregnancy have been associated with shortened limbs and intestinal malformations, presumably because of constriction of fetal blood vessels. Cigarette smoking has not been proven to cause fetal malformations but is associated with an increased occurrence of prematurity and low birth weight.[17]

Maternal Illness: Illness in a pregnant woman, especially certain viral infections, affect the fetus, causing brain damage and related disabilities. Ex TORCH infections, varicella, rubella, and herpes. Unlike the specific viral infections just mentioned, other acute viral and bacterial infections such as influenza, strep throat, and urinary tract infections do not damage the fetus. Certain chronic maternal illnesses like diabetes and seizure disorders place the fetus at risk of abnormalities, including spina bifida, heart problems, and malformations of the legs. Later in pregnancy there is an increased risk of toxaemia (described in the next section) and premature birth. With expectant mothers who have seizure disorders, the drugs used to treat the seizures rather than the seizures themselves appear to cause fetal malformations.

<table>
<thead>
<tr>
<th>DEFICIENCY IN DIET</th>
<th>GENETIC DISORDERS</th>
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<tbody>
<tr>
<td>Zinc deficiency</td>
<td>Down syndrome</td>
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<tr>
<td>Folic acid deficiency</td>
<td>Mitral valve proplase</td>
</tr>
<tr>
<td>Iron deficiency</td>
<td>Spinabifida, cleft palate, Anencephaly</td>
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<td>Low protein, low caloric diet, low vitamins in diet</td>
<td>Cystic fibrosis</td>
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<td>Excess copper accumulation</td>
<td>Wilsons disesse</td>
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<tr>
<td>Excess protein phenylalanine</td>
<td>Phenylketonurea</td>
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</tbody>
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Table 1
Medications: A number of medications have been associated with fetal malformations. The FDA guidelines divides drugs into 5 groups as follows. Anticancer drugs, massive doses of orally administered vitamin A, acne medication Accutane (isotretinoin) and the psoriasis drug Tegison (etretinate), antiepileptic drug Dilantin (phenytoin) have resulted in face and brain malformations in infants. A pregnant woman should take as few medications as possible during pregnancy, especially during the first trimester.

### Table 2

<table>
<thead>
<tr>
<th>MEDICINES</th>
<th>RISK FACTOR</th>
<th>Ex</th>
</tr>
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<tbody>
<tr>
<td>Group A</td>
<td>No foetal risk</td>
<td>Vitamin supplements</td>
</tr>
<tr>
<td>Group B</td>
<td>Not confirmed yet (Risk in animals)</td>
<td>Pencilin,Heparin,Insulin</td>
</tr>
<tr>
<td>Group C</td>
<td>Adverse foetal risk</td>
<td>Thalidomide</td>
</tr>
<tr>
<td>Group D</td>
<td>Can be used only in emergency</td>
<td>Anticonvulsants Antidepressants +/- benzodiazepines</td>
</tr>
<tr>
<td>Group E</td>
<td>Proved in no pregnant women</td>
<td>Anticancer drugs</td>
</tr>
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</table>

**PREVENTION**

The ultimate aim of conception is to get a better progeny. The planning for better progeny should be started before marriage. The whole ancient literature provides importance to the age factor including qualities of girl, specific mode of life, dietetic regimen, and various Samskaras to achieve a better progeny.

The excellence of Rtu, Kshetra, Ambu and Beeja always give excellent offspring. These four factors have to be protected from external and internal damages. Here excellence of Stree and Pumbeeja has to be emphasised rather than their mere presence.

Rtu kala niyamas are explained elaborately in the text books of Ayurveda. The utility of all restrictions is difficult to be explained in the present scenario because those reflects the social customs of that period, but definitely the physical and psychological status of women in the preconception stage have a clear impact on the to be born fetus.

Charaka Samhita elaborately explained Garbhopaghatakarabhavas, the harmful diet as well as activities of mother causing congenital anomalies. Since the fetus is fully dependant on mother any harmful things done by pregnant women will cause disruption to the fetus too. The suppression of desires may influence the psychology of mother and fetus as well.

Masanumasika Garbhiniparicharya should be followed as explained by the experts. During 1st trimester use of cold and sweet diet and milk besides the Madhurarasa Dravyas should be followed to prevent dehydration caused by vomiting and supply required nutrition. Same way month wise diet and regimen should be followed. Medications without consulting with the experts should be avoided.

The concept of prevention of Vikruta Garbha is the speciality of Ayurveda as it was well understood by the ancient scholars even thousands of years ago. The sperm and ovum which is having genetic components which are not visible where descend of Jeeva takes place. That’s why before taking decision about conception both male and female should undergo Panchakarma line of treatment for the prevention of transmission of congenital disorders to their offspring.

Based on contemporary understanding of concept of prenatal care we can summarise the following regimens to bing with the above said classical literature. Initially the lady has to undergo routine physical examination to verify that her body is healthy, immunization/vaccinations are up to date, review any prescription medications she is taking for safety during pregnancy, test for sexually transmitted diseases and seek treatment if already infected. A vitamin supplement recommended prior to conception. Folic acid supplements should be taken for...
three months prior to the pregnancy and through the first trimester. Prenatal testing can be done to reduce the risk of congenital anomalies and genetic counselling is recommended in case of high-risk cases.

CONCLUSION

Pregnancy should be by choice not by chance. Garbhakarabhavas are nothing but the carriers of organogenesis and other traits to the fetus. These traits are similar to traits carried by chromosomes /genes as per contemporary concepts. The normal transmitted traits through any of the Garbhakara Bhavas can be modified by preventive/curative measures unless they are permanent or serious. So antenatal care is having a major role in prevention of these type of abnormalities for which Ayurveda contributes certain diet and regimens from the time the couples plan for the conception till the time of delivery.

LIST OF REFERENCES

4. Ibid.Chapter1.p.2-3
22. Ibid.Chapter 8, Verse 31-32.