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## Contents

<table>
<thead>
<tr>
<th>Title</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Profunda Femoris Artery and its Branching Pattern and Variations</td>
<td>03</td>
</tr>
<tr>
<td>Eswari AK, Hemanth Kommuru, Satyalakshimi V, Swayam Jothi S.</td>
<td></td>
</tr>
<tr>
<td>Department of Anatomy, Shri Sathya Sai Medical College &amp; Research Institute, Ammapettai, Nellikuppam - 603 108, Tamil Nadu, India.</td>
<td></td>
</tr>
<tr>
<td>Phenytoin Induced Up Regulation of DEFB1 Gene in Albino Rat Testis Gene Microarray Analysis</td>
<td>09</td>
</tr>
<tr>
<td>Rajkumar R*, Vatsala Venkatesan†, Sriram Thanigai‡</td>
<td></td>
</tr>
<tr>
<td>*Department of Anatomy, Mahatma Gandhi Postgraduate Institute of Dental Sciences, Puducherry – 605 006, India.</td>
<td></td>
</tr>
<tr>
<td>†Department of Anatomy, Sri Balaji Medical College &amp; Hospitals, Chennai – 600 044, Tamil Nadu, India.</td>
<td></td>
</tr>
<tr>
<td>‡Department of Orthopedics, SRM.Medical college and Research Centre, Kattankulathur, 603 203, Tamil Nadu, India.</td>
<td></td>
</tr>
<tr>
<td>Functional Topography of Human Face</td>
<td>12</td>
</tr>
<tr>
<td>Balaji Karuppaiah, Perumal Saraswathi</td>
<td></td>
</tr>
<tr>
<td>Department of Anatomy, Saveetha Medical College &amp; Hospital, Thandalam, Chennai - 602 105, Tamil Nadu, India.</td>
<td></td>
</tr>
<tr>
<td>INIENCEPHALY-A Case Report</td>
<td>22</td>
</tr>
<tr>
<td>Jacinta Antony, Swayam Jothi D, Sai Suchitra</td>
<td></td>
</tr>
<tr>
<td>Department of Anatomy, Sree Balaji Medical College &amp; Hospital, Chromepet, Chennai – 600 044, Tamilnadu, India.</td>
<td></td>
</tr>
<tr>
<td>Variations in the Venous Patterns of the Head and Neck</td>
<td>24</td>
</tr>
<tr>
<td>Sree Lekha D, Sai Sucheethra D, Swayam Jothi Dorai Raj S, Uday Kumar P.</td>
<td></td>
</tr>
<tr>
<td>Department of Anatomy, Guntur Medical College, Guntur - 522 004, Andra Pradesh, India.</td>
<td></td>
</tr>
<tr>
<td>High Origin of Radial Artery – A Case Report</td>
<td>27</td>
</tr>
<tr>
<td>Sai Sucheethra D, Sree Lekha D, Swayam Jothi Dorai Raj S, Rajeswara Rao N.</td>
<td></td>
</tr>
<tr>
<td>Department of Anatomy, Guntur Medical College, Guntur - 522 004, Andra Pradesh, India.</td>
<td></td>
</tr>
<tr>
<td>Study of Holoprosencephaly</td>
<td>29</td>
</tr>
<tr>
<td>Melani Rajendran S†, Venkatasai PM‡</td>
<td></td>
</tr>
<tr>
<td>†Dept. of Anatomy, Matha Dental College and Hospital, Chennai – 600 069, Tamil Nadu, India,</td>
<td></td>
</tr>
<tr>
<td>¶Dept. of Radiology and Imaging Sciences, Sri Ramachandra Medical College and Research Institute, Chennai - 600 116, Tamil Nadu, India.</td>
<td></td>
</tr>
<tr>
<td>Instructions to Contributors</td>
<td>35</td>
</tr>
</tbody>
</table>
Profunda Femoris Artery and its Branching Pattern and Variations

Eswari AK, Hemanth Kommuru, Satyalakshimi V, Swayam Jothi S.

Department of Anatomy, Shri Sathya Sai Medical College & Research Institute, Ammapettai, Nellikuppam - 603 108, Tamil Nadu, India.

Key Words: Profunda femoris artery, Variations, Medial circumflex artery, Lateral circumflex artery & Perforators

Abstract: Profunda femoris artery is the main artery of the posterior compartment of thigh. 40 adult specimens and 10 foetal specimens were dissected and the level of origin of Profunda femoris artery in adult cadavers varied from 2 cms to 9 cms from midpoint of inguinal ligament and 0.8 cms to 1 cms in fetuses. In the adult in 4 specimens Lateral circumflex femoral artery was arising from Femoral artery, and in 5 specimens Medial circumflex was arising from Femoral artery & out of 10 fetus specimens in 8 specimens Medial & Lateral circumflex femoral arteries were arising from the Femoral artery and in two specimens from the Profunda femoris artery. All the Perforating arteries were arising from Profunda femoris except in 1 adult specimen where the second perforator was arising from Femoral artery. Profunda femoris artery is an important large branch of Femoral artery taking part in the longitudinal anastomoses at the back of the thigh. Thorough knowledge about the normal course and the variations of Arteria profunda femoris is essential for the vascular and orthopaedic surgeons and hence a detailed study of this artery was undertaken.

Profunda femoris artery (deep femoral artery) is an important large branch of femoral artery 3.5 cm distal to the taking part in the longitudinal anastomoses at the back of the thigh. Since superficial femoral artery occlusion is more common, surgical exposure of the Profunda femoris artery is often necessary in vascular reconstructive procedures. Management of groin sepsis involving the Femoral artery requires removal of infected tissue or prosthetic material and restoration of blood flow in many cases through the Profunda femoris artery.

The knowledge about the normal Profunda femoris artery and its variations are very important for the vascular surgeon according to which he can modify the surgical procedure in a more satisfactory way. This will help him to prevent most of the common post operative complications. A thorough knowledge about the normal course and its variations were essential.

Hence a detailed study of the profunda femoris artery and its variations in the branching pattern was undertaken.

Materials and Methods

40 thigh specimens from adult human cadavers and 10 thigh specimens from dead born fetuses were made use of. Conventional dissection method was used for the study.
Observations

Observations were made under the following headings:

i. The level of origin of profunda femoris artery from the midpoint of inguinal ligament.

ii. Relation of origin of profunda femoris artery to femoral artery

iii. Branches

The distance of origin of the profunda femoris artery from the midpoint of the inguinal ligament ranged from 0.2 to 9 cm with an average 3.88 cm in adult cadavers (Table 1). In 36 specimens the distance of the origin of the profunda femoris artery from the midpoint of the inguinal ligament was less than 3.88 cm & in four specimens it was more than 3.88 cm (Table 2)(Fig. 1).

In the dead born fetuses the origin of the Profunda femoris artery from the midpoint of the inguinal ligament ranged from 0.80 to 1.00cms with an average of 0.94 cm in length (Table 3) (Fig. 2). In four specimens origin of Profunda femoris from the midpoint of the inguinal ligament was less than 0.94 cm and in 6 specimens it was more than 0.94 cm. 38(95%) out of 40 adult specimens had the normal posterolateral origin (Fig. 3) and two (5%) of them had a more lateral origin (Fig. 4) (Table 4) from the femoral artery.

In all the 10 fetus specimens the profunda femoris artery was arising from the lateral side of the Femoral artery.

Regarding the branches, in 35(87.5%) out of 40 specimens of adult cadavers the Medial circumflex femoral artery was arising from the posteromedial side of the profunda femoris artery (Fig. 5). In 5(12.5%) specimens it was arising directly from the Femoral artery (Fig. 6) (Table 5). In 36(90%) out of 40 specimens Lateral circumflex femoral artery was arising from the profunda femoris artery (Fig. 7). In four (10%) specimens it was arising from Femoral artery (Fig. 8) (Table 6).

| Table 1 |

| TABLE 1 |

<p>| DISTANCE OF ORIGIN OF PROFUNDA FEMORIS ARTERY FROM THE MIDPOINT OF INGUINAL LIGAMENT |</p>
<table>
<thead>
<tr>
<th>Spec. No</th>
<th>Distance in cm</th>
<th>Spec. No</th>
<th>Distance in cm</th>
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<tbody>
<tr>
<td>1</td>
<td>9</td>
<td>2</td>
<td>21</td>
</tr>
<tr>
<td>2</td>
<td>3.8</td>
<td>22</td>
<td>3.8</td>
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<td>20</td>
<td>3.5</td>
<td>40</td>
<td>3.5</td>
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Average length: 3.88 cm

| Table 2 |

| TABLE 2 |

<table>
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<th>VARIABLE</th>
<th>SPECIMENS IN NUMBER</th>
<th>PERCENTAGE</th>
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<tr>
<td>Less than 3.88 cms</td>
<td>36</td>
<td>90%</td>
</tr>
<tr>
<td>More than 3.88 cms</td>
<td>04</td>
<td>10%</td>
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| Table 3 |

| TABLE 3 |

<table>
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<tr>
<th>Fetal cadaveric present study</th>
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<tr>
<td>Distance of origin of profunda femoris artery from the inguinal ligament</td>
</tr>
<tr>
<td>Spec. No.</td>
</tr>
<tr>
<td>-----------</td>
</tr>
<tr>
<td>1</td>
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<tr>
<td>2</td>
</tr>
<tr>
<td>3</td>
</tr>
<tr>
<td>4</td>
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<tr>
<td>5</td>
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In 34 (85%) specimens the Lateral circumflex femoral artery gave three branches - ascending, transverse & descending (Fig. 7, 8). In 6 (15%) specimens four branches were arising – 1 ascending, 2 transverse and 1 descending (Table 7) (Fig. 10).

Table 4

<table>
<thead>
<tr>
<th>Variable</th>
<th>Specimen in number</th>
<th>Percentage</th>
</tr>
</thead>
<tbody>
<tr>
<td>Less than 0.94 cm</td>
<td>4</td>
<td>40%</td>
</tr>
<tr>
<td>More than 0.94 cm</td>
<td>6</td>
<td>60%</td>
</tr>
</tbody>
</table>

Average length: 0.94 cm

Table 5

<table>
<thead>
<tr>
<th>Origin of medial circumflex femoral artery</th>
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<tbody>
<tr>
<td>From profunda femoris artery</td>
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<tr>
<td>From femoral artery</td>
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</tbody>
</table>

All the perforating arteries were arising from profunda femoris (Fig. 11) except in 1 adult specimen where the second perforator was arising from Femoral artery (Fig. 12) (Table 8).

Table 6

<table>
<thead>
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<th>Origin of lateral circumflex femoral artery</th>
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<tbody>
<tr>
<td>From profunda femoris artery</td>
</tr>
<tr>
<td>From femoral artery</td>
</tr>
</tbody>
</table>

Discussion

A number of scientists have worked on this area. According to them the origin ranged from 1 to 9.7 cm. In the present study the distance varied from 2 to 9 cm with an average distance of 3.88 cm. The present finding in the South Indian cadavers coincided with that of others (Susan, 2005; Wood Jones, 1953; Boilean Grant, 1958; Gene et al. 1995; Sinnatamby, 1999).

According to Hollinshed (1957), Gene (1995), the origin of profunda femoris artery was posterolateral in 95% cases so also in the present study. The more lateral origin seen in 5% of cases coincides with that of Susan (2005) and Sinnatamby (1999).

Medial circumflex femoral artery was arising from the profunda femoris artery in 84.5% cases and in 12.5% from the Femoral artery and these findings were similar to that of Gene (1995).

According to Hollinshed, Lateral circumflex was arising from the profunda femoris in 90% of the cases and in 10% from the Femoral artery and the present study reveals the same percentage of incidence. The Lateral circumflex artery giving more than three branches in 15% of the specimens was not reported by the earlier workers.

In 95% of the specimens three perforators were seen arising from the profunda agreeing with the findings of many authors. In 5% of the specimens, the 1st and 3rd perforators were arising from the profunda femoris and the 2nd arising from the Femoral artery. This was rare and was not documented so far.
Fig. 1 Right side long distance of origin of profunda femoris in adult cadavers (More than average distance- 3.88cm)

(PFA - profunda femoris artery, FA - femoral artery)

Fig. 2 In fetus origin of profunda femoris from femoral artery was close to the midpoint of inguinal ligament

(FA - Femoral artery, FV - Femoral vein, PF - Profunda femoris artery)

Fig. 3 Right side - Normal posterolateral origin of PFA from Femoral artery. IL - inguinal ligament

(FA - Femoral artery, MIP - Mid inguinal point, PF - Profunda femoris artery)

Fig. 4 Right side - Lateral origin of PFA from Femoral artery

(MIP - Mid inguinal point, FA - Femoral artery, PFA - Profunda femoris artery)

Fig. 5 Left side MCFA from PFA; PFA - Profunda femoris artery.

(MCFA - Medial circumflex femoral artery)

Fig. 6 Right side MCFA from FA directly (cut end seen)

(MCFA - Medial circumflex femoral artery, FA - Femoral artery)
Fig. 7 Left side LCFA – from PFA With 3 branches

Fig. 8 Right side LCFA – from femoral artery with 3 Branches

(FFA - Profunda femoris artery, LCFA - Lateral circumflex femoral artery)

(FCA - Lateral circumflex femoral artery, FA - Femoral artery)

Fig. 9 In fetus on the right side LCFA and MCFA from the femoral artery

Fig. 10 Right side 4 branches from LCFA

(LCFA - Lateral circumflex femoral artery, FA - Femoral artery, MCFA - Medial circumflex femoral artery)

(LCFA - Lateral circumflex femoral artery, FA - Femoral artery)

Fig. 11 Right side PFA with 3 perforators – Normal

Fig. 12 3-Perforators coming from the profunda femoris & second perforator was coming from the femoral artery.

(PFA - Profunda femoris artery, 1, 2, 3 - Perforators)

(PFA - Profunda femoris artery, FA, Femoral artery)
Conclusion

The profunda femoris artery is the artery of the posterior compartment and the knowledge about its variation is immaterial for orthopedic and vascular surgeons. In addition to the variations reported by the earlier authors, the present study revealed four branches arising from Lateral circumflex artery and the 2nd perforating artery arising from the Femoral artery.

References


Phenytoin Induced Up Regulation of DEFB1 Gene in Albino Rat Testis Gene Microarray Analysis

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‡Department of Anatomy, Sri Balaji Medical College & Hospitals, Chennai – 600 044, Tamil Nadu, India.
¥Department of Orthopedics, SRM Medical College and Research Centre, Kattankulathur 603 203, Tamil Nadu, India.

Key Words: micro array analyses – gene expression – LPL gene – Phenytoin effects

Abstract: Phenytoin is widely used against all types of partial and tonic-clonic seizures. Phenytoin may alter the release and action of different hormones which may contribute to sexual dysfunction. The present study is aimed at the effect of phenytoin induced differential regulation of DEFB1 gene in albino rat testis. The Albino rat were divided into two groups, control and test. The test group were given 150mgs/kg of phenytoin orally and equal amount of normal saline was given for control group. After 45 days with the rat under deep anesthesia, the testis were removed from the scrotum and stored in liquid nitrogen. The stored specimens of testis of control and tests group were subjected for cDNA microarray analysis. This study showed the differential expression of gene DEFB1 compared to the control group.

Phenytoin is effective against all types of partial and tonic clonic seizures. Its toxic effects are associated with chronic medication. Tania et al reported depression of cellular or humoral immunity or both, in a significant number of phenytoin treated patients. They found that phenytoin therapy was associated with failure of antibody response to Salmonella typhi antigen. Phenytoin is a major cause of antiepileptic drug hypersensitivity syndrome (AHS), which is rare but potentially fatal complication. Ito et al., reported agranulocytosis following phenytoin induced hypersensitivity syndrome. Kim et al., reported a case of 40 year old man who had drug rash with Eosinophilia and systemic symptoms (DRESS) syndrome associated with anticonvulsant drugs which are rare but potentially life threatening disease that occurs in response to arene oxide producing anticonvulsant drugs like phenytoin and carbamazepine. Human Beta-defsin DEFB1 is the most important antimicrobial peptide. It is coded by DEFB1 gene. It is the only innate immunity gene that shows long-term balanced selection and heterozygote advantage. It is capable of differentially regulate gene expression upon inflammatory or microbial stimuli.
Materials and Methods

Animal treatment and sample collection

Male adult albino rats were segregated into control and test groups. The test groups were treated with phenytoin 150 mg/kg body weight/day orally for 45 days. Similarly control groups were given equal amount of normal saline. In life study protocols, including animal housing, dosage, sacrifice and tissue harvesting were as per IAEC guidelines. After 45 days the tissue samples from test and control collected in Rnase free tubes and snap frozen in liquid nitrogen. Frozen tissues were stored in RNA later at -70°C until processed for RNA extraction.

RNA Isolation and DNA Microarray Hybridization And Analysis

RNA was extracted from the testis preserved in RNA later using Qiagen’s RNeasy minikit Cat#74104 and checked for purity and concentration. The extracted mRNA labeled with Agilent’s Quick-Amp labeling kit (p/n5190-0442) Hybridized with Agilent’s in situ Hybridization kit5188-5242 and scanned using high throughput Agilent scanner with “Surescan” technology.

Comprehensive Data Analysis

Data analysis includes automated feature extraction using Agilent feature extraction Software, Normalisation and statistical analysis and pathway and gene ontology analysis using Agilent’s Genespring GXv10==10.0 Biological interpretation of significant gene using Genotypics Bio interpreter Tool with literature curated information.

Observations

Phenytoin induced 2.41 folds up regulated DEFB1 gene expression was observed in phenytoin treated group when compared with untreated control group.

Discussion

Suresh yenugu et al., study of Systematic suites to identify and characterize novel antimicrobial proteins and peptides revealed that the majority of defensins are expressed predominantly in the male reproductive tract. Ito et al., reported agranulocytosis following phenytoin induced hypersensitivity syndrome. Subbannan Gujral reported necrotising lymphadenitis associated with the phenytoin induced hypersensitivity syndrome. Kim et al., reported drug hypersensitivity to previously tolerated phenytoin carbamazepine induced DRESS. All these reports confirm that phenytoin can cause hypersensitivity, necrotizing lymphadenits, drug rash with eosinophilia and systemic symptoms (DRESS) syndrome. Prado-montes de oca E found that DEFB1 is capable of upregulation upon inflammatory or microbial stimuli. The present study also confirms the 2.41 folds up regulated expression of DEFB1 which is due to hypersensitivity induced by phenytoin toxicity.

References


Hamadeh HK, Bushel PR, Supriya J, Bennet DO, Leping Li, Raymond SJ, Barrett C, Paules RS,

Functional Topography of Human Face

Balaji Karuppaiah, Perumal Saraswathi.

Department of Anatomy, Saveetha Medical College & Hospital, Thandalam, Chennai - 602 105, Tamil Nadu, India.

Key Words: Functional magnetic resonance imaging (fMRI), Blood Oxygen Level Dependent (BOLD) Technique, Echo planar imaging (EPI)

Abstract: Over yonks, motor cortex were studied by stimulatory method, with the advent of functional Magnetic Resonance Imaging, a new road map is created in studying motor cortex of human brain by non-invasive technique. In this study functional topography of human face is constructed using Blood Oxygen Level Dependent Technique. Instructions were given to the normal individuals to perform various paradigms involving different parts of the body and the corresponding cortex of the brain are represented as glowing areas. The amount of motor cortex involved is directly proportional to the density of innervations and not the area of the body surface.

Functional magnetic resonance imaging (fMRI) visualizes the active process in brain functions. It shows good correlation of neuronal function by shift in blood oxygenation following neuronal activity using Blood Oxygen Level Dependent (BOLD) Technique. Using non-invasive fMRI method, it is possible to localize functional brain activation in normal individuals with an accuracy of spatial and temporal resolution. Though numerous technical challenges remained, fMRI was increasingly becoming a key method for understanding the topographical organization of human brain (Cohen et al. 1994). The aim of the present study was to find the somatotopic representation of face in the cerebral hemisphere.

Materials and Methods

The present study examined the activation maps of face in normal adults. Institutional Review Board (IRB) and Ethical Committee (EC) approved the study. Each subject (patient) gave written informed consent. fMRI data were collected from 6 healthy volunteers aged above 25 yrs. Using Siemens 1.5 T (70mm wide bore MRI), fMRIs were acquired in active and rest states by performing special paradigms such as lip movement, tongue movement, swallowing and facial expression. The images were transferred to workstation for post processing and 3D reconstruction of motor homunculus. This will be useful to teach neuroanatomy and to locate primary motor areas more easily during surgery without functional deficit to the motor area. This technique maps the physiological or metabolic consequences of altered electrical activity in the brain and can be repeated in patients and normal individuals because of its non-ionizing nature.
**Echo planar imaging (EPI)**

It is a fast magnetic resonance technique, which is used to sequentially acquire brain images every few seconds (TR=2000ms-TE=4000ms) during several minutes of data acquisition. Its minimum acquisition time makes it ideal for fMRI acquisition.

**fMRI Paradigms**

fMRI detects the difference in electrical activity of brain neural signaling by difference in magnetic property between oxyhemoglobin and deoxyhemoglobin using a special technique known as Blood Oxygen Level Dependent (BOLD) Technique (Thulborn et al. 1982). During fMRI image acquisition, the subject as asked to perform several tasks such as swallowing, tongue movement with closed lip, lip movement, facial expression and visual -blinking movement. Each of these tasks was repeated several times and was separated by rest periods. The combinations of these tasks and resting states were known as fMRI paradigms.

**Blood Oxygen Level Dependent (BOLD) Technique**

The detection of functional areas of the brain was based on blood oxygen level dependent technique which created a difference between oxy and deoxyhemoglobin in each area of the brain when special task was performed. The biophysical basis of BOLD technique in fMRI was based on paramagnetic deoxyhemoglobin and diamagnetic oxyhemoglobin in an fMRI sequence. When neurons were stimulated during a task in an MRI scanner, it lead to local increase in energy and oxygen consumption in functional areas, and the hemodynamic changes transmitted via neurovascular coupling were measured using fMRI.

Paramagnetic deoxyhemoglobin produced local field in homogeneities in a magnetic field whereas the diamagnetic oxyhemoglobin did not interfere with the magnetic field (Ogawa and Kwong 1992).

When neurons were stimulated, there was an increase in local oxygen consumption that resulted in an initial decrease of oxyhemoglobin and increase in deoxyhemoglobin in the functional areas. To provide the active neurons with oxygenated blood, perfusion in capillaries and draining veins was enhanced with local oxyhemoglobin in several seconds. As a result, the initial decrease of local oxyhemoglobin was equalized and then over compensated. The progressive washout of deoxy hemoglobin by oxyhemoglobin caused a reduction in homogeneity in local field and increased BOLD signal in an f-MR image.

![Blood Oxygen Level Dependent (BOLD) Technique Diagram](image)

**Study Design and Data Acquisition**

During functional imaging, the images were acquired in a few seconds at quicker rate, when a subject performs a task that shifted the brain activity between active state and rest state. This could be explained by a simple BOXCAR design. This study design was simple and the average time consumed was efficient approach for comparing brain responses in different states (active state and rest state). An alternate approach of event-related paradigm was performed in the task.
between active and rest states, when the scanning was in progress.

Similar five sets of images were acquired in active state (30 sec) and rest state (30 sec) each, when the scan was in progress using boxcar study design. The signal time course in each voxel of the slices and the time course of different tasks were correlated. This could identify voxels in brain that showed statistically significant changes associated with brain functions. This statistical map (z-score) was superimposed on a high resolution anatomical image by using co-registration technique for proper identification of the precise anatomic location of origin of the signal. Since the procedure was cumbersome, it was carried out in a post processing work station of Siemens.

f- MRI Mapping of Eloquent Cortex

fMRI could obtain data preoperatively and non-invasively together with high sensitivity for visualizing brain lesions. It could distinguish and define the relation between the margin of a lesion and any adjacent functionally significant brain tissues. FMRI had the potential to predict possible deficit in motor and sensory perceptual functions and also to localize the motor homunculus in relation to the lesions in the brain. The usefulness of fMRI was to select the subject for surgery, tailoring surgical resection and in predicting the postsurgical outcome.

Observations

Swallowing movement in coronal sections

The functional activation of swallowing movement was seen at the motor area 4 on the superolateral surface of the cerebral hemisphere (Fig. 1)

The activation glow corresponded to the 3rd ventricle just above the lateral sulcus as in coronal sections. Functional glow was not seen on the right side at this level. Less functional activation specs of glow were on the superolateral surface of the cerebrum above the one mentioned on the left side. A small spec of glow was on the right side corresponding to the level of lateral ventricle.

Swallowing movement in axial sections

Similar functional activation was on the superolateral surface of the cerebral hemisphere on the motor area 4, which corresponded to 3rd ventricle as in axial section (Fig. 2). Functional glow was not on the right side at this level.

Swallowing movement in sagittal sections

Sagittal sections showed functional activation glow on the superolateral surface of motor area 4 in front of Rolandiuc sulcus, 5 cm above the external auditory meatus (Fig. 3). A speck of activation glow was in motor speech area 44, 45 and two specs of activation glow in the premotor area 6, 8.

3 Dimensional representation of swallowing area

3Dimensional representation of image on axial plane showed functional activation in the superolateral surface of the cerebral hemisphere on the motor area 4 with reference axial, coronal and sagittal planes in it (Fig. 4) This had been brought together to represent the swallowing area in somatomotor area 4.

Tongue movement in coronal sections

The functional activation of tongue movement was bilaterally on the superolateral surface of the cerebral hemisphere at the motor area 4, which corresponded to the level of lateral ventricle, fornix and septum pellucidum as
in coronal section (Fig. 5). The activation glow was more on the left side with stoop activation on the right side.

**Tongue movement in axial sections**

The functional activation of tongue movement was seen bilaterally on the superolateral surface of the cerebral hemisphere at the motor area 4, which corresponded to the level of body of lateral ventricle, fornix, and septum pellucidum as in axial section (Fig. 6).

**Tongue movement in sagittal sections**

Functional activation was at the motor area 4 on the superolateral surface of the cerebral hemisphere, 5 cm above the external auditory meatus (Fig. 7).

**3Dimensional representation of tongue movement**

3Dimensional representation of functional activation of tongue movement
was in coronal section (Fig. 8), at the motor area 4 on the superolateral surface of the cerebral hemisphere with reference axial, coronal and sagittal planes. The functional activation was bilaterally at motor area 4 on the superolateral surface just above the swallowing area.

**Lip movement in coronal sections**

Coronal section showed the functional activation on the superolateral surface of the cerebral hemisphere, just above the lateral sulcus at the level of 3rd ventricle. There was no glow at this level on the right side. There was stoop activation on the left hemisphere above the area of functional activation at the level of lateral ventricle as in coronal sections (Fig. 9). A few specs of diminishing activity were bilaterally on the right and left hemisphere, which corresponded to each other, above the level of lateral ventricle.

**Lip movement in axial sections**

Functional activation of lip movement was in motor area 4 on the superolateral surface of the cerebral hemisphere in axial sections (Fig. 10)

Activation was more on the left side. There was no activational glow on the right Side at the level of 3rd ventricle as in axial section (Fig. 10). A diminishing glow was on the right side corresponding to the lateral ventricle.

**Lip movement in sagittal sections**

Functional activation was above the lateral sulcus. The activation lay 5 cm above the tragus (Fig. 11), which corresponded to the somatomotor area of the cortex. A few diminishing glow was on the motor area above the functional activation.

**3Dimensional representation of lip movement**

3 Dimensional representation of lip movement on the motor cortex was in all the 3 axial, coronal and sagittal reference planes with central axial 3D image (Fig. 12), above tongue movement. The diminishing activity was noted in sagittal and coronal reference planes.

**Facial expression in coronal sections**

In coronal section, functional activation was on the superolateral surface at the level of body of corpus callosum (Fig. 13). There was no glow on the right Side at this level. A few diminishing activations were on the left side above the functional area of activation. There was a speck of glow above the level of corpus callosum on the right side.

**Facial expression in axial sections**

Functional activation of facial expression was on the superolateral surface of the cerebral hemisphere on the left side. There was neither similar nor small glow at this level on the right side of axial section. Little activation was in front of the above said glow on the right side.

Functional activation was at the area 17, 18, 19 on the right occipital lobe corresponding to the left hemisphere. The occipital lobe showed a few diminished activations in area 17 as in axial sections (Fig. 14).

**Facial expression in sagittal sections**

Functional glow was on the superolateral surface of the cerebral hemisphere above the circular sulcus of insula. There was also a little functional activation above, below and behind the above mentioned glow (Fig. 15).

**3Dimensional representation of facial expression**

3Dimensional representation of facial expression was in the coronal image with reference axial, sagittal and coronal planes. Coronal section revealed activation on the left side with diminishing activity on the right side visual area (Fig. 16).
Balaji and Saraswathi, Functional topography of human face
Discussion

Using Siemens 1.5 T, fMRI were acquired by performing special paradigms such as swallowing, tongue movement, lip movement and facial expression. The images were transferred to workstation for post processing and 3Dimensional reconstruction of motor homunculus of the face.

Swallowing movement

The functional activation of swallowing movement was at the motor area 4 on the superolateral surface of the cerebral hemisphere on the left side. The glow corresponded to the 3rd ventricle above the lateral sulcus in coronal, axial and sagittal sections. The functional glow well correlated with the electrical stimulation, which was a gold standard method. Less activated specs of functional glow were on the superolateral surface on both side cerebral hemispheres. It could have been due to slight movement of the muscles of the face.

Swallowing movement in sagittal section revealed the same findings as that of coronal and axial sections. A few specs of functional glow were on the motor speech area 44, 45 and premotor area 6 and 8. It could have been due to the coordinated movement of the swallowing area with premotor and motor speech area.

fMRI and electrical stimulation area were highly concordant (Stephani et al. 2000). These findings might enable the neurosurgeons to locate the primary motor area easily during surgery. High correspondence was between the somatotopic anatomy and function in Rolandoic sulcus (Stephani et al. 2000).

Tongue movement

Functional activation of the tongue movement was bilaterally at the motor area on the superolateral surface of the cerebral hemisphere, which corresponded to the level of lateral ventricle as in coronal section (Fig. 5). The activational glow was more on the left side because of dominant hemisphere. The stoop activation in the right side could have been due to possible bilateral cortical representation, tongue being a midline organ as in axial section (Fig. 6).

In sagittal section, functional activation was at the motor area on the superolateral surface of the cerebral hemisphere, 5cm above the external auditory meatus (Fig. 7).

3Dimensional observations showed bilateral representation of tongue movement clearly in all the 3 reference planes (Fig. 8).

Anatomical central location of global maximum intensity for each individual might vary, that depended on...
individuals task complexity, paradigm design, data analysis techniques or a combination of them, which formed the basis of MR imaging and the functions were related to anatomical foci (Vincent et al. 2006).

**Lip movement**

Functional activation was on the superolateral surface of the left dominant cerebral hemisphere, just above the lateral sulcus at the level of 3rd ventricle as in coronal section (Fig. 9). Stoop activation was above the area on the left side. This could have been due to group activity of the muscles of facial expression.

A few specs of diminishing activity bilaterally corresponded to each other on the right and left hemispheres above the level of lateral ventricle. This showed the bilateral cortical representation of the upper part of the face in the facial nerve nucleus. It also correlated with clinical cases of supranuclear injury of facial nerve. Only lower half of the face was paralyzed and upper half of the face was spared.

In addition to functional glow at the motor area in sagittal section lip movement, a few diminishing glow was above and in front of lip area. It could have been due to group action of the muscle of facial expression.

**Facial expression:**

Functional activation of facial expression was on the superolateral surface of the left hemisphere at the level of trunk of corpus callosum (Fig. 13). No glow was on the left side. There were a few diminishing glows on both the hemisphere at this level. The diminishing glow was bilateral and indicated bilateral cortical representation of upper half of the face. Functional activation on the right side also indicated the bilateral cortical representation.

Functional activation was on the right calcarine sulcus area 17 & 18 on the left hemisphere and a few diminishing activity in area 17 of right hemisphere in axial section (Fig. 14). This could have been due to the blink reflex during facial paradigm.

Functional glow was also on the superolateral surface above the circular sulcus of insula in sagittal section. There was also a little functional activation above, below and behind the functional area of the left hemisphere. This could have been due to group activity of the muscles. All this diminishing activity was only in the pre-Rolandic area. This could have been due to stimulation of cerebral cortex in response to cerebral activity.

High functioning individual with autistic disorder differed from normal individual in the activity of cerebellum, limbic and temporal lobe cortical region of the brain, when processing facial expression (Critchly et al. 2002). They did not activate the cortical face area when explicitly (consciously) or the lt. amygdala or the lt. cerebellum, when implicitly (unconsciously) processing the emotional facial expression. This could be due to Neuro developmental in origin.

Using non-invasive fMRI, it was possible to localize functional brain activation in normal individual, with an accuracy of millimeters and temporal resolution of seconds. Though numerous technical challenges remained fMRI was increasingly becoming a key method for understanding the topographical organization of the human brain. Combination of the navigation system and fMRI was useful for preoperative design of the surgical strategy and tumor orientation during the operation, enabling aggressive surgery to be performed without functional deficit. (Morioka et al 2001).

fMRIi was useful during resection of tumor in the motor area and help the surgeon to avoid or minimize post operative...
functional deficit and was crucial preoperative decision step for 90% of brain tumors (Klink et al. 2012).

fMRI task might replace the invasive gold standard of electrical stimulation (Brockway et al 2004).

Conclusions

The amount of motor cortex involved was directly proportional to the density of innervations and not the area of body surface. fMRI could be used to learn functional neuroanatomy of brain by constructing motor homunculus. It could also be used to assess the motor outcome of patient with various neurological disorders. The diagnostic information of fMRI permitted functional preservation and safe treatment. Mapping motor homunculus -swallowing, tongue movement with closed lip, lip movement, facial expression using fMRI could identify the eloquent cortex and predict post operative deficit of specific functions during the pre surgical works. These findings might enable the neurosurgeons to locate primary motor area more easily during surgery. fMRI was a sensitive and specific method for mapping language and motor functions. BOLD technique was non--invasive and alternative to invasive stimulatory method. fMRI and DTI were used to assist in preservation of structure and functions of motor system, which promised to decrease the patient’s morbidity and to broaden the clinical applications of functional imaging. Local relationship of cerebral tumors and sub-cortical fiber tracts could be defined. FMRI passive paradigm was an alternative and complementary to active movement task in patient population. FMRI imaging enabled the selection of more aggressive therapeutic approach that might otherwise be considered as functional risks. In certain patients surgical time might be shortened, the extent of resection was increased and craniotomy size was decreased. Repeated electrical stimulation might weaken the neuronal activity. This disadvantage was overcome by non-invasive BOLD technique of fMRI. Though numerous technical challenges remained, fMRI was increasingly becoming a key method for understanding the topographical organization of human brain. This non-invasive fMRI might replace the invasive gold standard electrical stimulatory method.

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References


Case Report

INIENCEPHALY-A Case Report

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Key Words: Iniencephaly, inion, spina bifida, meningocoele

Abstract: A full term dead born foetus delivered by Caesarean Section from the Obstetrics and Gynaecology Department of the Government Kilpauk Medical College & Hospital was handed over to the Department of Anatomy for further study. The head of the foetus was retroflexed, a cystic swelling was present dorsally above the anal orifice and ventrally an omphalocoele.

Iniencephaly is a rare malformation which is incompatible for post-natal survival. It is more common in female sex than male and has been extensively described by Ballantyne (1904) and Gilmour (1941).

Case Report

The face of the foetus was well formed. The posterior hairline of the scalp merged with the skin of the thoracodorsal region thereby obliterating the nape of the neck. There was a cystic swelling above the anal orifice.

The scalp was opened by a coronal and a sagittal incision. The frontal and parietal bones were intact. The brain had autolysed. The squamous part of the occipital bone was absent. There was no foramen magnum. The base of the skull presented the cranial nerves in their respective foramina.

A mid sagittal section was made on the dorsal aspect encircling the cystic swelling. The laminae and spines in the lumbar vertebrae were absent-spina bifida. The vertebral canal contained the spinal cord and the swelling was a meningocoele. Omphalocoele was present ventrally.

Discussion:

The absence of the inion on the squamous part of the occipital bone which is absent is responsible for the title of this paper. This condition is normally associated with spina bifida or meningocoele.

Borderland and Morison in their text book on Foetal and Neonatal Pathology state that this condition cannot be distinguished from KLIPPER-FEIL SYNDROME. It has been reported as early as 1904.

Conclusion

Foetus with more than one anomaly is an interesting case for study. This requires study of each region in detail regarding its development and also the genetic reasons for such malformation. This condition is reported here because it had more anomalies than reported earlier. Past history and family history did not reveal any causative factor. The genetic abnormality leading to this condition could not be carried out.
Reference:


Variations in the Venous Patterns of the Head and Neck

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Key Words: venous pattern – venous drainage – head & neck

Abstract: In this study, variations in venous patterns of the head & neck studied in 30 human cadavers were reported.

Veins are known for their variations than arteries. The anterior facial vein is the chief vein of the face. Since the veins of the head & face are without valves the communication of the anterior facial vein with the ophthalmic vein and also with the infraorbital and buccinator veins are of special clinical importance. (Hollinshead 1961)

Materials and Methods

Venous drainage of face was observed in 30 cadavers. In an adult male cadaver during routine dissection the anterior facial vein was traced.

Observations

The variation found in one male cadaver was as follows. On the left side four deep facial veins were seen connecting the anterior facial veins with the pterygoid plexus of the veins (Fig. 1). The size of the anterior facial vein was average, joined a large anterior division of posterior facial vein to form a large common facial vein. The common facial vein divided into two – one drained into the internal jugular vein and the other division was large and continued as prominent, large anterior jugular vein (Fig. 2). The posterior division of retromandibular vein was small and it joined with the posterior auricular vein to form a thin external jugular vein and it soon divided into finer divisions and disappeared over the middle of the sternocleidomastoid muscle (Fig. 3). Beyond that existence of external jugular vein could not be made out in the posterior triangle.

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Fig. 1 On the left side four deep facial veins were seen connecting the anterior facial veins with the pterygoid plexus of the veins.

(AFV – Anterior Facial Vein, DFV – Deep Facial Vein)

On the right the anterior jugular vein was connected to the pterygoid plexus of veins by four deep facial veins (Fig. 4). The
Variations in venous patterns of head and neck were normal on the right side.

Fig. 2 Common facial vein divided into two – one drained into the internal jugular vein and the other division was large and continued as prominent, large anterior jugular vein

(CFV-Common facial vein, AJV-Anterior jugular vein, IJV- Internal jugular vein)

Fig. 3 External jugular vein and it soon divided into finer divisions and disappeared over the middle of the sternocleidomastoid muscle

(EJV- External jugular vein, IJV- Internal jugular vein)

Discussion

Because of the connections of the anterior facial vein it has often been emphasized that infections above the area of the mouth should not be touched from the point of view of attempting to evacuate them for instance. (Pratt 1937; Maes 1937). Roeder (1936) suggested ligation of veins as a prophylactic measure, in order to prevent spread of infection to the cavernous sinus. Maes (1937) regarded this as of dubious value because once the thrombus has reached the sinus, surgical treatment is of little use because of the development of meningitis. In the present case there were four deep facial veins connecting the anterior facial vein with the pterygoid plexus of veins which are connected to the cavernous sinus.

The anterior jugular vein usually begins in the suprahoid region through the confluence of various and variable superficial veins or it may arise more laterally from the posterior or common facial veins or parotid veins. In this case it was arising from the common facial vein.
The external jugular vein, begins in the substance of the parotid gland where it is most often formed by the union of the posterior facial and the posterior auricular vein (Brown 1941) or quite variably by a single one or some communications of these and the common facial, internal maxillary or other veins in the parotid region. Close to the parotid gland it received a communication from the internal jugular vein. In the present case the size of the external jugular vein was smaller and faded away forming a plexus at the middle of the sternocleidomastoid muscle.

**Conclusion**

The case is presented for the clinical significance of variable location of the veins of the face. Since numbers of veins were leading from the danger area of the face to the pterygoid plexus of veins, the danger of spread of infection is much more and this is a precarious situation for cavernous thrombosis to the precipitated

**References**


Case Report

High Origin of Radial Artery – A Case Report

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Key Words: radial artery, anomalous origin

Abstract: An unusually high origin of radial artery observed during routine dissections was reported here for its rarity.

Superficial arteries of the arm are relatively common. McCormack (1953) found some types in 30.77% of 364 bodies in which both limbs were investigated. They were commonly unilateral, so the incidence among limbs was much less (18.53% of 750 limbs)

During routine dissection two brachial arteries were observed in the right arm of a male cadaver. The arteries were traced proximally and distally.

Case Report

The following observations were made – The largest branch of brachial artery – the arteria profunda brachii was arising from the third part of the axillary artery in common with subscapular artery (Fig. 1).

Two brachial arteries were seen (Fig. 2) one (superficial brachial artery) was found in continuity with the radial artery at the level of cubital fossa (Fig. 3). The other had usual course and divided into its terminal branches namely radial and ulnar arteries at the level of the tuberosity of the radius.

Discussion:

The primitive radial artery arises from the axial artery in the arm; where as the definitive radial artery arises from the axial artery at the level of the bend of the elbow. Subsequently the primitive radial artery joins with the definitive radial artery. At a later date the primary radial regresses
completely but occasionally it may persist so as to account for the high origin of the radial artery from the brachial artery in the arm.

Fig. 3 Deeply placed brachial artery divided into ulnar & radial arteries. Superficial brachial artery was communicating with radial artery

(SBA–superficial brachial artery communicating with the radial artery, R- Radial artery)

Trotter and her colleagues (1934) found the subscapular, both circumflex scapular, the profunda brachii and the superior ulnar collateral all share a common stem, the axillary artery then bifurcating into radial and ulnar arteries.

Origin of a superficial brachial artery in the arm formed 5.57 percent of the total number of vascular variations they observed in these limbs. High origin of radial artery formed 77% of all variations of the arteries of the arm in 12.14% it arose from the brachial artery 2.66% of limb has an anastamotic connections with the brachial artery (McCormack and Co workers, 1953)

In the present case the brachial artery was doubled. The high origin of radial artery was due to the persistence of the primitive radial artery. This anomaly could be explained with an embryological background. Further knowledge about the variation will be of use while reducing fracture of the humerus, in reparative surgery and during cardiac catheterization.

Reference:


Study of Holoprosencephaly

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Key Words: holoprosencephaly, alobar holoprosencephaly, semilobar holoprosencephaly, lobar holoprosencephaly, neural tube defect

Abstract: Holoprosencephaly is a rare congenital brain malformation that occurs during development due to incomplete separation of the cerebral hemispheres. It may be caused due to hereditary factors, chromosomal anomalies, environmental teratogenic factors. Four cases of holoprosencephaly were observed during a period of one year and their features are discussed.

Neonatal brain injury is very important since it predicts subsequent infant mortality and morbidity in the premature infants. The term holoprosencephaly is proposed by De Myer (1963). Holoprosencephaly is a disorder resulting from failure of septation, cleavage, or differentiation of the midline forebrain structures at various levels or to various degrees (De Myer, 1963). It is caused by a primary defect in patterning and induction of the basal forebrain during embryogenesis, causing the improper development of the brain and resulting in incomplete division of the cerebral hemispheres (Stashinkio, 2004). Holoprosencephaly most commonly affects the telencephalon and the diencephalon (De Myer, et al. 1963). According to the degrees of failed differentiation of the forebrain, the holoprosencephaly is classified into 4 categories: alobar holoprosencephaly, where the brain is not divided, semilobar holoprosencephaly in which the cerebral hemispheres are incompletely developed and lobar holoprosencephaly in which the brain has somewhat divided which is the least severe form. Syntelencephaly, or middle interhemispheric variant of holoprosencephaly (Armand Marie Leroi, 2003), in which the posterior parts of the frontal and parietal lobes are improperly separated, but the rostro-basal forebrain properly separates; it is also currently classified as a type of holoprosencephaly (Armand Marie Leroi, 2003).

Holoprosencephaly is associated with facial anomalies including cyclopia, proboscis, ethmocephaly or cebocephaly (Kinsman, et al. 2000), microcephaly, midface flattening, hypotelorism, flat nasal bridge and single maxillary central incisor, premaxilla agenesis, median cleft palate and cleft lip and other less-severe facial anomalies (Wallis and Merenke, 2000).

The aim of this study is to increase the understanding and awareness of genetic and clinical manifestations of holoprosencephaly. Therefore a study was carried out to find out the probabilities of occurrence of
holoprosencephaly among the neural tube defects for a period of one year. 40 cases of neural tube defects were found, among which 4 cases of holoprosencephaly were identified and reported.

Materials and Methods

About 1500 antenatal scans were done for a period of one year, in the I, II and III trimester; out of which, 40 central nervous system anomalies were found and in turn, 4 of them showed holoprosencephaly.

They were:
- Alobar prosencephaly – 2
- Semilobar prosencephaly – 1
- Lobar prosencephaly – 1

Autopsy was done with the concern of the parents. Autopsy findings were correlated with sonographic findings. CNS anomalies not compatible with life were terminated.

Observations

Alobar holoprosencephaly

The foetuses were 19 weeks & 20 weeks old respectively. They were found with alobar holoprosencephaly (Figs.1a &1b). The parents of both the foetuses were consanguineous and one of the parents suffered from infertility for 5 years. One of the fetuses had cyclops and with proboscis. Their brains showed no separation of cerebral hemispheres; absence of falx cerebri; a large single central ventricle; thin and fused thalami; small third ventricle and the midbrain with a small cerebral aqueduct.

Semilobar holoprosencephaly

The foetus was 22 week old with semilobar holoprosencephaly (Figs.2a & 2b). Its parents were non consanguineous. The foetus was found with median cleft lip. It had a small single central ventricle; fused frontal and parietal lobes bilaterally; interhemispheric fissure was present only posteriorly; Lack of cleavage of basal ganglia and thalami; absence of body of corpus callosum but genu and splenium were present.

Lobar holoprosencephaly

The foetus was 20 week old with lobar holoprosencephaly. The parents were non consanguineous. The foetus had median cleft lip and cleft palate. The brain was found with right and left cerebral hemispheres but the frontal lobes were fused and the lateral ventricles were separated (Figs.3a,b & 3c).

Discussion

The holoprosencephaly is a rare congenital malformation and one of the neural tube defects. It is classified into 4 categories: alobar holoprosencephaly, in which nondivision of the brain and with associated facial anomalies. The brain show absence of the interhemispheric fissure, falx cerebri, third ventricle and fused thalami and often absence of neurohypophysis and olfactory tracts (De Myer et al., 1963); semilobar holoprosencephaly in which cerebral hemispheres is incompletely developed with posterior partial formation of the interhemispheric fissure, with only a single ventricle, variant heterotopic gray matter; lobar holoprosencephaly is the least severe form in which the brain has somewhat divided. The brains are with the presence of an interhemispheric fissure but the cingulate gyrus and fused lateral ventricle and absence of septum pellucidum (De Myer et al., 1963) and syntelencephaly, or middle interhemispheric variant of holoprosencephaly (MIHV), in which the posterior frontal and the parietal lobes are not improperly separated, but the rostro-basal forebrain properly separates. Earlier it is not considered as a variant of HPE at all, but is currently classified as a type of holoprosencephaly (Armand Marie Leori, 2003).
Fig. 1a & b Alobar holoprosencephaly

Fig. 2a & b Semilobar holoprosencephaly (22\textsuperscript{nd} week of pregnancy)

Fig. 3a & b Lobar holoprosencephaly

Fig. 3c Lobar prosencephaly

The four cases of the holoprosencephaly observed in the present study also showed majority of the malformations reported above.

Apart from the facial anomalies, associated abnormalities include microcephaly, hydrocephalus, agenesis of the corpus callosum, posterior cranial fossa abnormality, cerebellar vermis aplasia, myelomeningocele, absence of an olfactory bulb, hypothalamic and brainstem dysfunction, increased muscle tone, sleep disturbances (Barr and Cohen, 1999), renal
dysplasia, renal cysts, cardiovascular malformations, intestinal abnormalities, omphalocele, gastro oesophageal reflux, choking etc, club foot, sirenomelia,(Chen et al. 1997), spina bifida, and endocrinopathies (pituitary gland dysplasia, growth hormone deficiency and diabetes insipidus (Traggiai and Stanhope, 2002), polydactyly (Verloes et al. 1991), inability to smell, developmental delay, intellectual impairment, seizures etc. (Bronshtein and Wiener, 1991).

Development

During the third week of embryonic life, the prechordal mesoderm migrates into the area prior to the notochord and affects midline facial development. Hence before 4 weeks of embryonic age, the varying degrees of loss or disruption in the development of prechordal mesoderm cause abnormal forebrain development, midfacial defects as well as fusion of lateral ventricles and the 3rd ventricle (Muenke and Beachy, 2000). Division of cerebrum into right and left halves normally occurs at the end of the 5th or the beginning of 6th week of gestation.

Birth prevalence rate

During early embryogenesis, the prevalence rate is about 1 in 250; the prevalence rate in live births ranges from 1: 14,736 to 1: 26,730 due to the high rate of spontaneous abortion, the prevalence rate in live births ranges from (Cohen,1989); Bullen et al. (2001) represented the total prevalence (including pregnancy termination) was 1.2 cases per 10,000 registered births, and the birth prevalence (affected live births and stillbirths at > 24 weeks' gestation) was 0.49 cases per 10,000 births. But according to Dubourg et al. 2007, holoprosencephaly is seen in 1 per 10,000 to 16,000 live births. In most cases of holoprosencephaly, if the malformations are so severe the babies die before birth (Tatori et al. 2005).

Etiology

Knowledge of the etiologies of holoprosencephaly is important for establishing the risk of recurrence. The etiology of holoprosencephaly indicates interactions with both genetic and environmental factors which include chromosomal anomalies, gene rearrangements, Mendelian mutations and teratogens.

Genetic factors

The majority of holoprosencephaly cases are autosomal dominant, autosomal recessive, or X-linked in inheritance. Almost 50% of all holoprosencephaly cases have cytogenetic abnormalities and approximately 18% -25% of patients of holoprosencephaly have a documented monogenic syndrome (Croen et al. 1996; Olsen et al. 1997). Approximately half of all infants or fetuses with holoprosencephaly also have chromosomal abnormalities, most often trisomy 13 (Cohen, 1989; Megahan et al. 1990; Muenke, 1994; Rasmussen et al. 1996; Ming et al. 1998; Bullen et al. 2001; Blaas et al. 2002).

Environmental factors

Environmental factors are mainly maternal diabetes mellitus which increase holoprosencephaly risk (Ramos-Arroyo et al. 1992; Muenke, 1994; Ming and Mueuke, 1998; Peebles, 1998; Croen, 2000; steroid alkaloids, thanol, and retinoic acid (Sulik et al. 1995). X-ray exposure, maternal alcohol consumption during early pregnancy (Cohen, 1989; 2002), steroid alkaloids, thanol and retinoic acid (Sulik et al. 1995). X-ray exposure, maternal alcohol consumption. Other factors which have teratogenic effects are maternal smoking, respiratory illness medications and salicylate-containing medications, estrogen/progesterin, anticonvulsants, weight reduction diets and low maternal weight, previous pregnancy loss and congenital infection with cytomegalovirus, rubella, and
toxoplasmosis (Croen et al. 1996). Women with early menarche are more likely to have holoprosencephaly (Croen et al. 2000).

**Conclusion**

Because of the short life span and ominous outcome in all patients with holoprosencephaly, genetic counseling and prenatal diagnosis is essential. The earliest gestational age at the time of diagnosis is 14 weeks (Bronshtein and Wiener, 1990). Prenatal ultrasound can detect the more severe forms of holoprosencephaly and associated defects such as hydrocephaly (Chervenak, 1985; Vintzileos et al. 1987; Peebles, 1998). Therefore prenatal diagnosis and with the consent of the parents, elective termination reduce the birth prevalence of holoprosencephaly (Croen, 1996; Forrester, 2000; Bullen, 2001; Blaas, 2002).

Knowledge of the different varieties of holoprosencephaly helps the clinician to arrive at an antenatal diagnosis and to decide about the fetal outcome.

**References**


Rasmussen SA, Moore CA, Khoury MJ, Cordero JF (1996) Descriptive epidemiology of holoprosencephaly and arhinencephaly in
Melani and Venkatasai, Holoprosencephaly


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