NINE MONTHS that made you

The development of a new human being

From the moment of fertilisation right up until birth, the development of a new human being occurs through a complex set of coordinated biological processes working intensively to model the body's systems and establish their functioning. This poster charts this process, explaining what is happening in the key stages of a baby's development. It moves from the two-celled embryo that is created following the moment of fertilisation when sperm and egg fuse on Day 1, and leads you through to the 38-week foetus waiting to be born.

DAYS 1-2

TWO-CELLED EMBRYO Genetic information provided by the fusion of the sperm and egg is copied and the developing embryo divides to form two distinct cells, each with a copy of the combined set of genes. This happens at approximately **24–30** hours after fertilisation.

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DAY 3 MORULA

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The cells of the embryo keep replicating their combined set of genetic information and then dividing so that new cells are born. By the time the embryo is around three days old it contains roughly 12–16 cells and is called a 'morula'.

DAY 4 **BLASTOCYST**

The embryo finishes working its way through the Fallopian tube and begins its entry into the uterus. It forms into a fluid-filled sac with a bunch of cells located at one end. It is now called a 'blastocyst'

WEEKS 1–2 IMPLANTATION

At the end of Week 1 and the start of Week 2, the blastocyst cells hatch out of the fluid-filled sac and embed into the wall of the uterus, negotiating with the mother's immune system to prevent attack and instead assist it in the embedding process.

WEEK 3 APPROX SIZE 0.2 cm the lungs

Cells have organised themselves into three layers and start to fold. The outer layer will make skin and the nervous system. The middle layer will make the heart and circulatory system and the inner layer will make

NEEK 9 APPROX SIZE 5 cm

Arms and legs have now grown much longer. The fingernails are developing and fingerprints start to form when the layers of the skin buckle, forming folds that are seen as unique whorls, swirls and grooves.



The developing embryo has been continuing to fold its layers of cells, creating clearly defined grooves and folds. The most prominent of these are the neural fold and groove as the nervous system starts to take shape

APPROX SIZE 6 cm

As we have marked the moment of fertilisation as Day 1, this week corresponds to the end of the first trimester of pregnancy. The foetus is developing its genitals and the liver starts to make red blood cells



EYES

There are two different cell types within the human eye that are capable of detecting light. These are known as rods and cones These cells, located in the retina, contain a protein pigment that alters in response to light. This signal is then converted into an electrical signal and transmitted via the sensory nerve projections of these cells through to other cells in the retina. The other retinal cells then transfer the information to the brain for perceptual processing, where an overall image is formed.



The brain is a complex organ involved in sensory, notor and cognitive (thought) processes. There are many different cell types within the brain including neurons (which transmit information in the form of electrical and chemical impulses), glia (supporting neurons) and astrocytes. In order for the brain to achieve its functions these cell types must communicate with each other by forming a dense network of connections and by releasing and receiving chemical and electrical signals.

EARS

-----Our ears create our sense of balance and detect sounds. Sound detection occurs via fine hairs called stereocilia located on cells that form the organ of corti in the inner ear. Hairs vibrate in response to pressure generated by sound waves entering the ear. This vibration is then transmitted via sensory nerves on to the brain. Our sense of balance is also generated by the inner ear, again by hair cells, but these are located in the semicircular bony canals of the ear and in the nearby otolithic organs. The hairs move in response to the force created by the head turning and transmit this information as electrical and chemical signals to the brain for further processing.

limb buds appear. APPROX SIZE 0.8 cm

The tiny heart can be heard beating. The body takes on a distinctive curvature as the neural tube closes. Facial features such as the eyes start to develop and

The central nervous system is busy generating **WEEK 12** neurons and connections between them to support movement as the baby becomes much more active during the second trimester of pregnancy. At this APPROX SIZE 8.7 cm stage the sex of the baby is visible.

HEART

The heart consists of muscle that contracts and relaxes to pump blood around the body. The signal to contract and relax is electrically generated by the movement of calcium ions. The heart pumps red blood cells around the body delivering oxygen for cellular respiration, the process by which cells use energy in order to perform their function. The blood is also one location where some of the cells of the immune system are located, including white blood cells, responsible for detecting invading pathogens and mounting a response to defend the body. The heart therefore ensures adequate immune protection of the body via the circulation of blood.



LIMBS

Our bones give our body structure and strength. The skull protects the delicate brain while the bones of our limbs are used by our muscles and tendons to pull against (as levers) to help us to move. Our joints contain sensory receptor cells capable of telling our brain where our joints are at all times. Peripheral nerves carry this information from the joints, via the spinal cord and on to the brain which in turn can generate motor signals, carried back to the body by peripheral nerves, telling our muscles to move.

Tiny webbed fingers and toes start to become visible WEEK 6 as the limb buds develop. The lenses of the eye start to form and the lungs begin to develop. APPROX SIZE 1.3 cm

WEEK 24 APPROX SIZE 36 cm

The baby practices breathing movements ready for birth even though there is no air to fill its **lungs**. It is now responsive to touch and sounds from outside the womb, and will move in response to them.





The lungs perform the process of respiration, taking in oxygen and delivering it into the blood supply so that it is ready to be used by cells for cellular respiration. The lungs also receive the output product of cellular respiration (carbon dioxide) from the blood, ready to exhale and expel this from the body. Carbon dioxide is toxic so it is vital that it is not allowed to build up inside the body. Likewise, without a constant supply of oxygen, the cells of the body will start to die.

The Open University

THE BIOLOGY OF THE HUMAN BODY

BBC

The human body is a biological marvel, consisting of a number of highly specialised organs and other tissues. The cells that make up these organs and tissues all started out as unspecialised cells created in the two-celled embryo but they will have differentiated into their specialised forms during the process of development. Each of our organs and other tissues performs very specific functions that are important for our survival. All must therefore be correctly formed during the period of development between fertilisation and birth so that they can perform as required. Here you can get a brief insight into the functioning of some of our organs and tissues so that you can appreciate the level of sophistication of the human developmental process. On the reverse side of this poster you have the chance to explore human developmental biology at the cellular and molecular level by learning how our genes can affect our future health.

For more information visit www.open.edu/openlearn/countdowntolife

WEEK 7 APPROX SIZE 1.8 cm

Fingers start to form as the webbing between them gradually disappears through a process of programmed cellular destruction. **Ear** shapes are slowly forming on the side of the head.

WEEK 33 APPROX SIZE 50 cm

The heart and all the blood vessels are complete. However, the **lungs** are still developing, producing larger amounts of surfactant that will help the alveoli in the lungs to inflate after birth, taking in air.



WEEK 38 APPROX SIZE 52 cm

The baby is full term. All **bones** have hardened except the skull, which remains flexible so that the baby can be squeezed through the birth canal. The amniotic fluid that fills the lungs will drain away on the baby's first breath of air.



BBC

The interaction of genes and environment in the womb

A baby's genes are inherited from its father and mother. Genes contain the basic information that is used to shape the baby's characteristics. These include things like hair colour and eye colour, but they also include characteristics such as the baby's temperament and its susceptibility to illness.

The nine months that makes a baby can effectively be thought of as a period of intense programming. Genetic information is not fixed at the moment of fertilisation, where the sperm and egg fuse together and combine their genetic material creating the genetic basis for a new human being.

Genetic material, comes in the form of DNA. DNA stands for deoxyribonucleic acid, (pronounced dee-oxy-rybo-new-clayic acid). DNA can be found in the nucleus of cells, and it is organised into small sections called chromosomes



The DNA within each cell provides information that can be read and then used by the cell to build substances that will help the cell to function. These substances are called proteins which are absolutely essential for life because they can have a profound effect on the functioning of a baby's (and later a child's or an adult's) bodily processes.

During the period of development between the fertilisation of the egg and birth, our cells multiply, growing in number by replicating their DNA, and then dividing. At a particular stage in this process cells start to become 'specialised'. This means that they start to make proteins required for very specific bodily functions. For example, a cell that is destined to become a brain cell (a neuron) will make proteins that are essential for brain function allowing the cell to send and receive signals to and from other neurons. A cell that is destined to become a heart muscle cell will make proteins essential for heart function, allowing the heart to expand and contract for the purpose of pumping blood around the body. Of course, a whole human body has very many different specialised cells so DNA must contain a very complex set of

instructions

Maternal nutrition and **DNA methylation**

he term 'epigenome' refers to all of the genetic information located within the cells of an organism, including the markers that are placed onto the DNA to regulate transcription and translation of genes.



of growth is during development within the womb, nutrients are in great demand during this time period and it seems that the availability of some key nutrients can have lasting consequences for the epigenome.

The primary donor (provider) of methyl for the purpose of DNA tion is a substance called S-Adenosylmethionine (or SAM for short). This substance is generated in a metabolic process called 'one carbon metabolism'. The one carbon metabolic process gets its name from the fact that the entire process concerns the movement of a carbon atom between several different molecules.

One carbon metabolism relies on several key nutrients including vitamins B2, B6, B12, Folate, Choline, Betaine and Methionine. One carbon metabolism is a complex metabolic process that takes place in the body's cells but the stage that is most relevant to DNA methylation is the stage where methy from SAM is attached to DNA in the presence of an enzyme called DNA methyltransferase. An enzyme is a protein made by cells that is involved in assisting reactions involving other organic substances. Usually enzymes speed up reactions between substances (this can be quite significant – even in the order of one million times faster!). Essentially this means that some very complex chemical reactions are able to take

place within the body at body temperature (37°C in humans) Most scientific research studying the effect of maternal nutrition on epigenetic mechanisms in offspring has been done on animals. In 2014, however, a study in humans has

also shown that factors affecting availability of nutrients

NINE MONTHS that made YOU

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One type of marker is methyl made from carbon and hydrogen atoms. The main source of methyl groups that are used in the marking of DNA is nutrients. Nutrients are found in our food sources and they are used by the body to support processes essential for life and growth. Given that an intensive period



Every human being is therefore a product of their genes – but they are not a fixed product, whereby the genetic information created by the fusion of the sperm and the egg is maintained in a constant state. During a baby's time in the womb, genes can be chemically altered by events happening outside the womb, and this will affect how a baby is 'made' during development. vents happening within the environment, to the mother, have the power to switch genes on or off. Switching a gene on or off affects the proteins that get made. A gene that is switched on is able to make its protein while a gene that is switched off cannot do this. The lack of an essential protein could have a profound effect on the health of an individual, even threatening survival, while the production of some proteins when certain genes are switched on can have more positive consequences.

The rapidly growing area of science discovering more about how genes can be turned on and off according to events happening in the environment is known as 'epigenetics'. It is showing us that what we experience in our lives today may have very little to do with what happened to us yesterday, or last week. Instead, it may have much more to do with how our genes were programmed during our time in the womb.

DID YOU KNOW?

That there is currently no universally accepted theory of how the organic building blocks of life, such as amino acids, originated on Earth.

WANT TO LEARN MORE?

basis of human life, and how this impacts on health, we recommend our qualifications in Health Sciences and Natural Sciences (Biology). If you would like to try a short taster module in Science to explore whether distance learning is for you, why not try our Access module Y03: nce, technology and maths, which provides excellent academic preparation for our Health Sciences and Natural Sciences (Biology) qualifications, should you decide to continue with your Open University studies.

f you are interested in learning more about the genetic

involved in the one-carbon metabolic process in a mother, such as the effect of the local weather on crops, can have an effect on DNA methylation in her offspring. More importantly, the effect of maternal nutrition on DNA methylation can occur based on the mother's pre-conception nutrient levels. The study was done in rural Gambia where the local population

are reliant on home grown/tended food sources. Therefore the weather can greatly impact on the availability of nutrients in the diet of this population. The study confirmed that whether it was the 'rainy' or 'dry' season did affect the levels of onecarbon nutrients in women in the region, with some being higher and some being lower depending on which season it was. Furthermore, the level of these nutrients was linked to the level of DNA methylation in specific genes within the babies of mothers in this region. What was not investigated, however, was what the functional effects of this methylation were, since this would require what is known as a longitudinal study where

the babies that took part in the study would be monitored throughout childhood and on into adulthood to determine any impact on their bodily processes. Such a study would obviously be quite invasive for the people concerned and this is therefore an example where ethical considerations might limit the ability of science to gain answers to the burning questions that



DID YOU KNOW?

researchers have.

That laboratory experiments have shown that organic molecules like amino acids can be synthesised from the small inorganic molecules that were thought to be present on early Earth.

WANT TO LEARN MORE?

If you would like to learn more about nutrition and its effect on health we recommend our Level 1 module SDK1 which forms the first stage in our undergraduate degree in

Health Sciences. SDK100 provides a gentle introduction to the areas of science that are relevant to human health, including nutrition. In particular the module is focused on understanding the methods that science can use to answer

important questions in health, and on understanding why, sometimes, the answers we can gain from our methods may be frustratingly limited.

Reading genes to make essential life proteins

Almost all cells in the human body contain genes. Genes are located within the nucleus of cells and they are used to make proteins, amongst other things. The proteins made from genetic information are essential for life since they are involved in the functioning of bodily processes.



Proteins are said to be 'coded for' by genes, which means that the instructions for making a particular protein can be found in parts of genes contained within the nuclei of the body's cells.

The entire collection of genes that contains the coding information for a whole human being is known as the human genome and every cell in the human body, except red blood cells, contains this entire set of instructions within its nucleus. However, as the body's cells are specialised, each one will only need to make some of these proteins rather than all of the proteins that are coded for by the human genome.

A helpful analogy for thinking about the human genome is to think of it as a recipe book. This recipe book contains a recipe (a gene) for every dish (a protein) in the human body. In each cell in the body a different set of recipes (genes) is used and thus a different menu of dishes (proteins) can be produced by that cell. Therefore, there has to be a mechanism for turning genes on and off so that only the proteins that are needed by the specialised cell are made rather than all the proteins coded for in the human genome.

One mechanism for turning genes on and off is the process of methylation. But in order to understand how this turns genes on and off you first need to know a little about how information from genes is read for the purposes of making proteins, Within the nucleus of a cell, where genetic information can be

found, the part of the DNA that makes up a particular gene coding for a specific protein is copied to create a specialised molecule. This molecule is called RNA (ribonucleic acid, pronounced rybo-new-clay-ik acid). The process of copying the genetic information is called 'transcription' because the RNA is transcribing the instructions contained within the DNA for making a protein. Since DNA exists in a double helix structure

Maternal stress and the developing brain

When scientists use the term 'stress' they are referring to a state of heightened awareness and a readiness to act, typically caused by something that has happened in the external environment. It is the body's way of responding to pressure.

Stress has an evolutionary significance since parts of the body's stress response have evolved to help us to deal with threats where we might need to run away from a predator to survive or to stav and fight

The feeling of 'being stressed' is known to be regulated by a collection of structures within the brain known as the hypothalamic pituitary adrenal (HPA) axis. This term is a collective term for all the structures involved in this response including the hypothalamus (an area deep within the brain), the pituitary gland and the adrenal gland. The structures that form the 'HPA axis' all release hormones to communicate with each other and the rest of the body during a stress response. Hormones are proteins that have the ability to enter the blood circulation, so they can have wide-ranging effects throughout the body since they have the ability to easily move around it.



Excessive and prolonged activation of the HPA axis of a mother under conditions of extreme stress during pregnancy can have effects on the developing brain of the baby. This effect can occur via epigenetic mechanisms. However, the good news is that these effects can also be compensated for after birth since the epigenome is constantly in a state of flux. It is not simply modelled during the womb and then fixed from birth onwards

Stress during pregnancy can affect methylation of the gene that codes for a very important protein involved in the stress response. This protein acts as a 'receptor' for the stress hormone called cortisol in the brain of the developing baby. Cortisol is a form of hormone called a glucocorticoid. Receptors for cortisol are proteins that are shaped in such a way that they can join together with the cortisol that is released by structures in the HPA axis. As cortisol is a glucocorticoid the receptors for it are called glucocorticoid receptors (or GRs for short)

where two strands of molecules are tightly coiled together, copying the genetic information within DNA during the process of transcription involves unwinding and separating these two strands of DNA. Once the two strands are separated RNA then uses the information in one of the strands to help it to form into a copy of the matching strand. This is effectively the process of 'transcription'. Once the RNA has successfully transcribed the genetic instruction it is then known as 'messenger RNA'. This is because it is carrying the message needed for the cell to make a protein. Why is there a need for a messenger molecule like messenger RNA? It is because the genetic instructions are not in the

same location within a cell as the cellular machinery capable of building proteins. The DNA is contained within the nucleus of a cell. But proteins are made elsewhere within the cell in the cytosol (fluid) that surrounds the cell nucleus. Thus the nessenger RNA carries the message from the nucleus out into the cytosol where the protein can be built by specialised cellular machinery known as ribosomes that are capable of putting together the building blocks of the protein. These building blocks are known as amino acids and the process of reading the message contained within messenger RNA and using it to build a protein is known as 'translation'. Therefore, in order for genetic information to have any kind o functional effect within the body it must be both transcribed and translated. However, this is not the end of the story, since not all proteins are produced in the location where they will

eventually be needed to perform their function. Once protein synthesis is completed some proteins are simply released into the cytosol but some are delivered to their end location by a system of protein targeting. A clever method of using a signal sequence of amino acids within the protein to act like a postal address ensures that it is delivered to its required location.



DID YOU KNOW? That one theory suggests organic molecules arrived in debris from space. In 1969 a meteor that fell over Australia was shown to contain amino acids.





Within the HPA axis, the GRs for cortisol act as a form of feedback mechanism which helps to dampen down the stress response (the release of cortisol) and return the system to its pre-stress state, after a stressful situation. This dampening effect occurs because the joining of cortisol to its glucocorticoid receptors causes an overall inhibition of cortisol elease from the HPA axis.

The process of methylation turns the gene for cortisol receptors off in some cells, so the brain of a developing baby will have fewer of them, and so the stress response will be more likely to run unchecked. In a mild form this effect simply makes the baby, and later the child or adult, more stress-responsive. In its most extreme form, this kind of effect could also predispose the individual to health conditions like depression, which is linked to an overactive stress response and hyperactivity of the HPA axis

However, it should be remembered that parts of the epigenome remain adaptable throughout a person's life and it has been shown that a strong bond between mother and infant after birth can also positively affect the HPA axis and its functioning.

DID YOU KNOW?

That present day cells have a membrane that defines them. t is made up from small molecules with a water-loving ic) head and water-hating (hydrophobic) ta hese line up next to each other to form the membrane.

WANT TO LEARN MORE?

f you are interested in learning more about the links between brain functioning and mental health, including the epigenetic basis of mental health, we recommend module The science of the mind, which can be studied as a standalone module or as part of our Health Sciences or Natural Sciences (Biology) qualifications.

Turning genes on and off: the epigenome

Genes are short sections of DNA that can be copied into a molecule known as messenger RNA within a cell's nucleus so that the message contained within the RNA can then be translated outside of the cell's nucleus in order to produce proteins. The types of proteins that a cell makes determine its function within the body.

Proteins carry out essential functions in our bodies. For example, hormones are proteins. The antibodies produced by our immune systems are proteins. Digestive enzymes in our guts are proteins. The haemoglobin that transports oxygen around our body is also a protein. There are many more examples!

While the entire genetic code to make a human being is referred to as the human genome, the human epigenome can be thought of as all the chemical substances that are capable of interacting with DNA and marking particular genes so that these are either activated (switched on) or inactivated (switched off). The word 'epi' is Greek and it means 'on'. Therefore the word 'epigenome' guite literally means 'on the aenome

When a section of DNA (a gene) is inactivated, it cannot be transcribed by RNA – so the protein coded for by that particular gene would not be made by the cell. One process by which DNA can be marked and so turned on or off is the process of methylation. This refers to the attachment of chemicals called methyl groups to parts of the DNA. A methyl



Genetic mutations and human health





When a gene is present in a different form it is known as an allele (al-eel). Gene mutation is the fundamental source of heritable variation as long as the mutation arises in the cells that produce offspring. These cells are known as gametes and they are the sperm and the egg. Gametes are unusual in that they contain only one set of the genetic material of the parent All other cells in the parent's body contain two sets. But there is a reason for the gametes only having one set. Since gametes fuse during fertilisation to create a new individual, and that individual will require two sets of genetic information the gametes by default can only contain one set. If they did contain two sets the process of fertilisation would produce cells with four sets of genetic information!

In order for a mother or father to pass on a mutation the allele must first be present in the DNA contained within the sperm and/or the egg. However, since the process of fertilisation joins the two donated sets of genetic material, the baby will receive either one mutated allele from the mother, one from the father, or one from each of the mother and father. This last scenario results in the baby having two matching mutated alleles. In this situation the individual is said to be homozygous for whatever characteristic the gene codes for. When the individual only receives one copy of a mutated allele they are said to be heterozygous. However, the story does not end there. An individual that receives only one mutated allele may not express the characteristic coded for by that gene because the allele may be recessive rather than dominant In other words, when the two sets of genes are different (an individual who is heterozygous), one allele will dominate (be switched on) and one will become recessive (be switched off) If the allele is a recessive one, two copies of it are required before the condition is expressed whereas if the allele is a dominant one, only one copy is required for the condition to be expressed (because the allele dominates).



group is made up of carbon and hydrogen (it is precisely one carbon atom joined to three hydrogen atoms). Methyl groups can be created from natural sources like food, but they can also be created from man-made sources like medicines and pesticides. This information should now make it a little easier to imagine how the external environment can come to influence how we are made from our genes, since the external environment can regulate DNA methylation by affecting the availability of methyl within the body.

Once they are joined onto DNA, methyl groups can sometimes be removed again. In fact, the process of DNA methylation as well as the removal of methyl groups (known as demethylation) is constant, not just occurring during the period of development in the womb. However, for some genes, DNA methylation can be stable. This means that once a section of DNA has methyl groups attached to it, these methyl groups can remain on the DNA even into adulthood.

For the sake of completeness, a known alternative means of marking DNA is something known as histone modification, where different chemical substances (not carbon and hydrogen in the form of methyl) can affect the way in which DNA is structured. Histones are proteins and they play an important role in the structure of DNA, since the DNA strands are tightly wound up around histone proteins with the proteins acting a little bit like spools. Their purpose is to enable very, very large molecules like DNA to be wound up into small chunks to form chromosomes.

The process of histone modification of DNA can make it harder or sometimes easier for RNA to transcribe the section of DNA that makes up a gene. If RNA cannot transcribe the information contained within DNA, then this information can never make it out of the cell nucleus, and it will therefore never be used to make the protein that it codes for.

DID YOU KNOW?

hat prior to the emergence of life on Earth it is thought that only gases like ammonia, hydrogen, carbon dioxide hydrogen sulphide and water vapour were present in





The mutated genes that are known to be related to health conditions (so called genetic disorders) have the same inheritance patterns and so some disorders are said to be dominant disorders' and some are 'recessive disorders'. An example of a dominant disorder is Huntington's disease which leads to the onset of degeneration of the nervous system in adult life. An example of a recessive disorder is cystic fibrosis. which is the commonest genetic disease of childhood and where life expectancy is often only 30–35 years of age.

Gene mutations, like epigenetic variations, will affect the proteins that are made within the body. Given that proteins are chains of amino acids it is entirely possible for these chains to be put together incorrectly so that the amino acids are out of order, or the wrong amino acid is inserted at a particular point within the chain that makes up the protein. The ability of a protein to function depends on its three-dimensional shape, which is determined by the amino acids it is made from and how these are pieced together. It is perhaps easy to see how a protein that functions as a receptor (such as a GR for cortisol) would be altered out of shape so that it no longer exactly fits around the substance that would join to it (the cortisol). The gene that is altered in Huntington's disease builds a protein that is too long. Unfortunately this longer-than-nomal protein is in some way toxic to the cells within the brain and the toxicity of the abnormal protein is directly related to the length of the protein. So the longer it is, the earlier the onset of cell death within the brain of an affected individual. There are many other ways that mutated genes can affect the body's functions. We have just given a few examples here

DID YOU KNOW?

That human and animal cells also have internal membranes that form around subcellular compartments called organelles. These have their own contents and functions which make every cell specialised.

WANT TO LEARN MORE?

If you would like to learn more about genes, proteins and cell function within the body we recommend module S Cellular and molecular biology which can be studied as a standalone module, or as part of our qualifications in Natural Sciences (Biology), and Health Sciences.