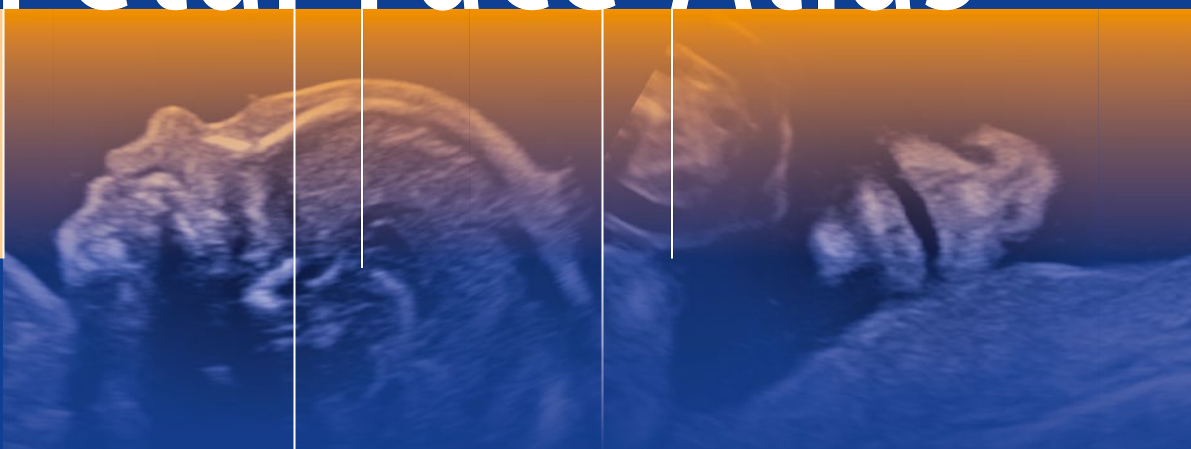
Normal and Abnormal Fetal Face Atlas



Ultrasonographic Features

Jean-Marc Levaillant

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Jean-Marc Levaillant
Center of women and fetal imaging
Créteil
France

Jean-Philippe Bault
Center of fetal imaging Ambroise Paré
Les Mureaux
France

Bernard Benoit
Princess Grace Hospital
Monaco

Gérard Couly
Department of maxillo-facial surgery
Necker Hospital
Paris
France

Translation from the French language edition 'La face foetale normale et pathologique: aspects échographiques' by Jean-Marc Levaillant, Jean Philippe Bault, Bernard Benoit, Gérard Couly © Sauramps Médical, Paris, 2013; ISBN: 978-2840238690

ISBN 978-3-319-43768-2 ISBN 978-3-319-43769-9 (eBook)
DOI 10.1007/978-3-319-43769-9

Library of Congress Control Number: 2017933711

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The registered company is Springer International Publishing AG
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Foreword

In the field of obstetrics and fetal medicine, there are a few invariables in clinical use of ultrasounds. The operator must have a clear representation of the structures he or she is analyzing and the possible anomalies, which supposes knowledge of basic physiology, anatomy, and embryology. He or she must be able to use the spectrum of technical solutions available, know what to expect from them, and use them in the most up-to-date fashion, such as not generating images that are aesthetically pleasing but lack information. He or she should take a systematic approach, standardizing imaging via an analysis through the planes of symmetry; the idea is to be able to reproduce the examination and to avoid creating nice-looking but fallacious images. When it is pertinent, use of objective quantitative criteria should be privileged.

Communication should also be taken into consideration—a communication based on transmitting information rather than producing flashy images. It is important to understand the expectations of the various partners involved—geneticists, pediatricians, surgeons, and parents—and be able to respond to their specific questions.

Sharing experience is an important part of our practice. Teaching involves disseminating knowledge and showing targeted images; it also involves transmitting how to obtain these images via a multitude of approaches: words, screen captures, hands-on tutoring, and live demonstrations.

This list must also include (self-) assessment, which is at the heart of any responsible practice.

This atlas of normal and abnormal fetal face ultrasound imagery presented by Jean-Marc Levaillant, Jean-Philippe Bault, Bernard Benoit, and Gérard Couly fits this approach. It provides a clear, didactic approach with a review of the basic embryology and anatomy needed to understand the images, along with up-to-date clinical knowledge and the links between clinical practice and the images under both normal and various pathological situations. It also includes a collection of biometric curves. Beyond the images themselves, readers will appreciate the authors' explanations of the optimization and utilization of the various ultrasound imagery modes, as well as how to obtain the various shots and corresponding images.

In this respect, this atlas is another step in the development of the French School of Gynecological and Obstetrical Ultrasonography, which includes some major developments, including pioneering developments in 3D ultrasonography in collaboration with Philips Research Laboratories, followed by the first industrial developments with KretzTechnic; training in 2D and 3D ultrasound imagery at the Saint-Denis Ultrasound School, with trainers from all over France and let by the tireless and ubiquitous applied engineer Bernard Meyer; and journal publications on fetal face ultrasound imagery.

Daniel Rotten
Department of Gynecology and Obstetric
Delafontaine Hospital
Saint-Denis
France

Acknowledgements

We would like to thank our colleagues who contributed to the imagery found in this atlas:

Caroline Alby
Bettna Bessières
Christian Bisch
Joseph Bonan
Rabi Chaoui
Sophie Couderc
Laurent Guibaud
Soraya Kabar
Brigitte Leroy
Anne Elodie Millischer
Daniel Moeglin
Marc Molho
Edwin Quarello
Ahmed Sadji
Karima Sedikhi

Contents

1 Introduction Craniofacial Epigenesis in Vertebrates

The facial bones, as they appear in a fetal ultrasound image, result from a continuous process of development, regulated and sequenced by genetic and molecular embryology, which progressively constructs the face like epigenesis.

The face, like the entire head, is "prefigured" by prescribed regions that are genetically determined during the gastrula and neurula stages. Via typological deformations, the two-dimensional neural surface transforms symmetrically and acquires a third dimension.

When the dorsal region of the neural grove closes into a tube, the "fourth layer" of migrating neural crest cells invade the future embryonic cephalic pole and provide the cellular filling for the facial prominences.

The face develops thanks to the five founding prominences or swellings of the ectoderm, which grow volumetrically and then fuse by contact and are no longer recognizable as having existed once the face is totally formed. These prominences are:

- The frontonasal prominence, which is median, single, symmetrical and different from the other four as it is formed by two left and right halves.
- The maxillary prominences, right and left.
- The mandibular prominences, right and left (first branchial arches).

1.1 The Key Stages in Craniofacial Development

The prescribed regions of the face and brain are differentiated as early as the gastrula and neurula stages.

Gastrulation lays out the embryonic body plan for all vertebrates with bilateral symmetry via the formation of the notochord and the mesoderm interposed between the endoderm and the ectoderm (Fig. 1.1) (this beginning explanation of epigenesis or the phenomenology of development by stages replaced the eighteenth century concept of preformation that stemmed from the building of the first microscope, when Leeuwenhoek, its inventor, identified the spermatozoid in 1677).

The future regions of the face are situated in the rostral regions of the gastrula.

Neurulation is a theater of morphogenetic operations that will differentiate the prescribed regions of the head (Fig. 1.2). This is the stage during which the central nervous system, the facial mass and the neck truly begin their morphogenesis.

The neural plate is the two-dimensional cellular surface whose fate mapping demonstrates the closeness and unity of the regions of the brain, the neuro-sensorial receptors, and the face, genetically "printed" on the neural plate.

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J.-M. Levaillant et al., *Normal and Abnormal Fetal Face Atlas*, DOI 10.1007/978-3-319-43769-9_1

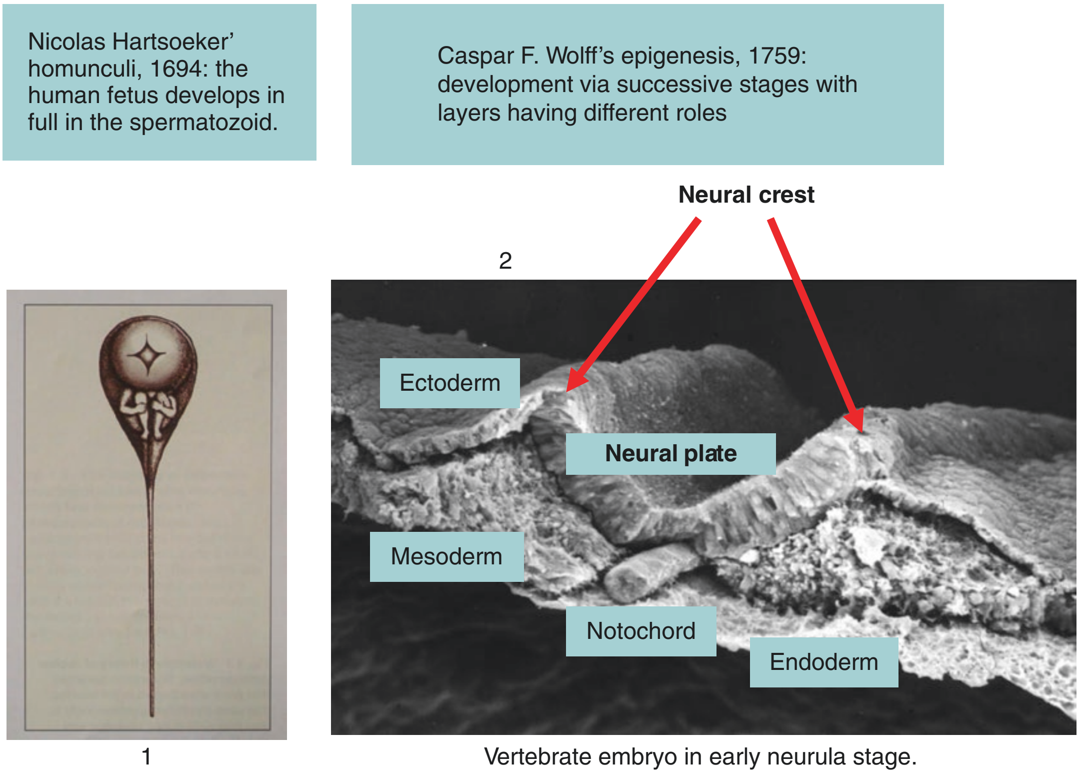

Fig. 1.1 Preformationism vs. epigenesis. *1* Homunculus. *2* End of gastrula phase, beginning of neurula stage

During neurulation, the topological transformation of the neural plate continues. The face is in the neurula and "grows" in front of it (Fig. 1.3).

The neural plate lengthens and folds, forming a groove with edges rising up and fusing into a tube (Figs. 1.4 and 1.5).

Closure of the neural tube's rostral opening represents an exceptional topological originality (Fig. 1.6): the anterior-most parts of the neural plate roll up forward, completing a half-circle rotation (180°), flipping these regions into a ventral position (it is an elegant and original solution to the problem of closing a tube that is open in the front (Figs. 1.7 and 1.8).

The lateral edges of the neural tube come together and fuse in the middle. As a result, the adenohypophysis (Fig. 1.9), initially the anterior-most neuro-ectodermic region, moves under the diencephalon and thus becomes the posterior-most region, above the future stomodeum. The two olfactory placodes sprout nerves and the olfactory apparatus, initially located above and on either side of the telencephalon and the adenohypophysis, approach the median line due to the median fusing of the neural ridges and position themselves ventrally under the tele-dicephalon, or the anterior brain (Fig. 1.10).

This is how the future nasal cavities and the rhinencephalon are linked for the sense of smell.

Jointly, the retina surfaces on the neural plate "blister" outwards and becomes the optic cups and then the optic vesicles (see Fig. 1.7).

The dorsal closure of the neural tube by molecular fusion triggers the migration of cephalic neural crest cells (CNCC) **(see** Figs. 1.4, 1.11, 1.12, 1.13, 1.14 and 1.15).

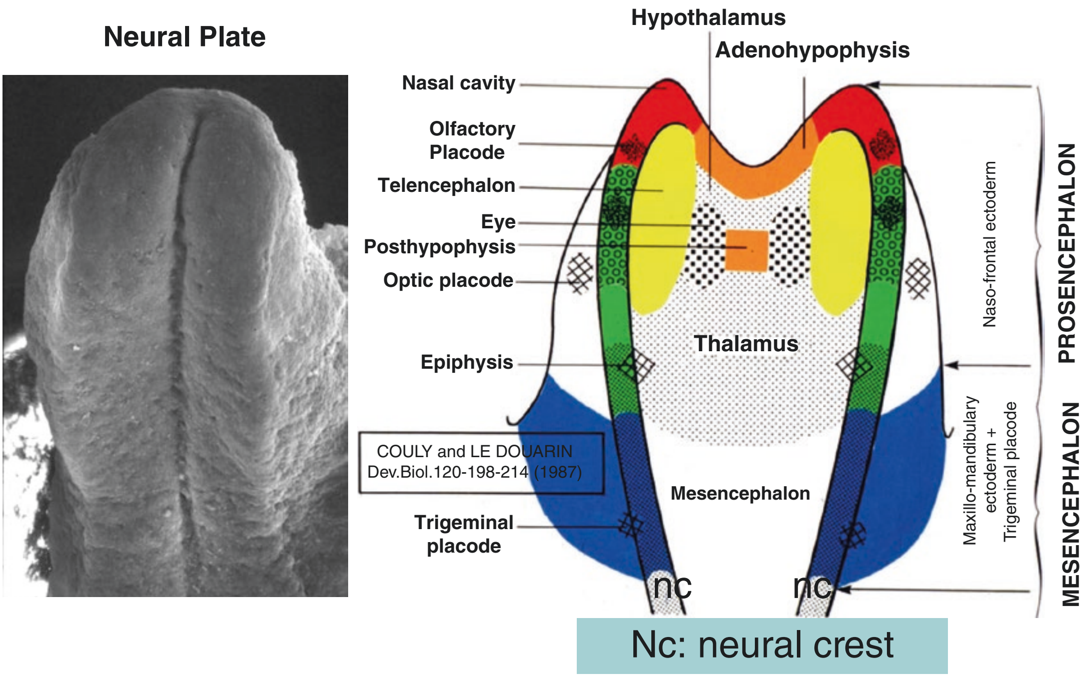

Fig. 1.2 Map of neural plate regions

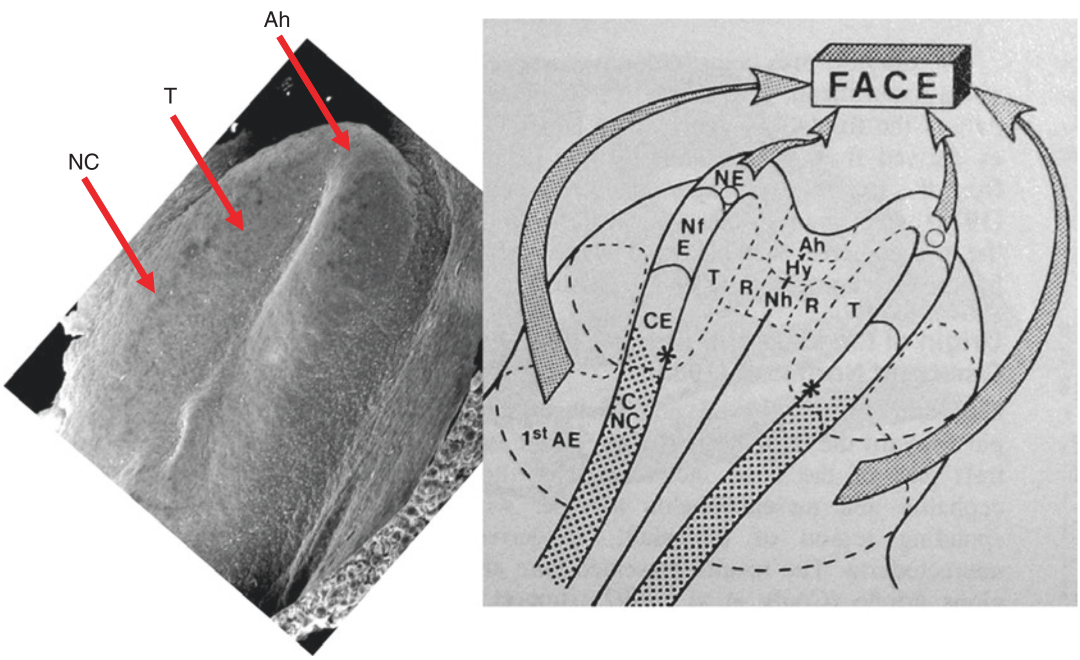

Fig. 1.3 The face grows in front of the neural plate

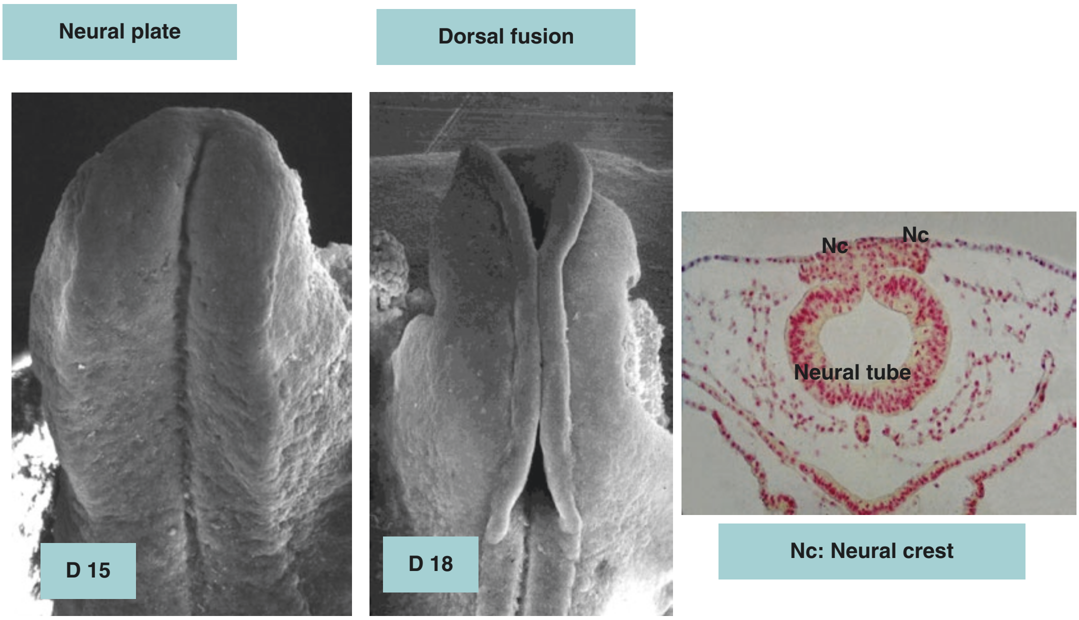

Fig. 1.4 Closure of the neural plate into the neural tube, with neural crest
D 15 – The neural plate
D 18 – Dorsal fusion
NC Neural crest
Neural tube

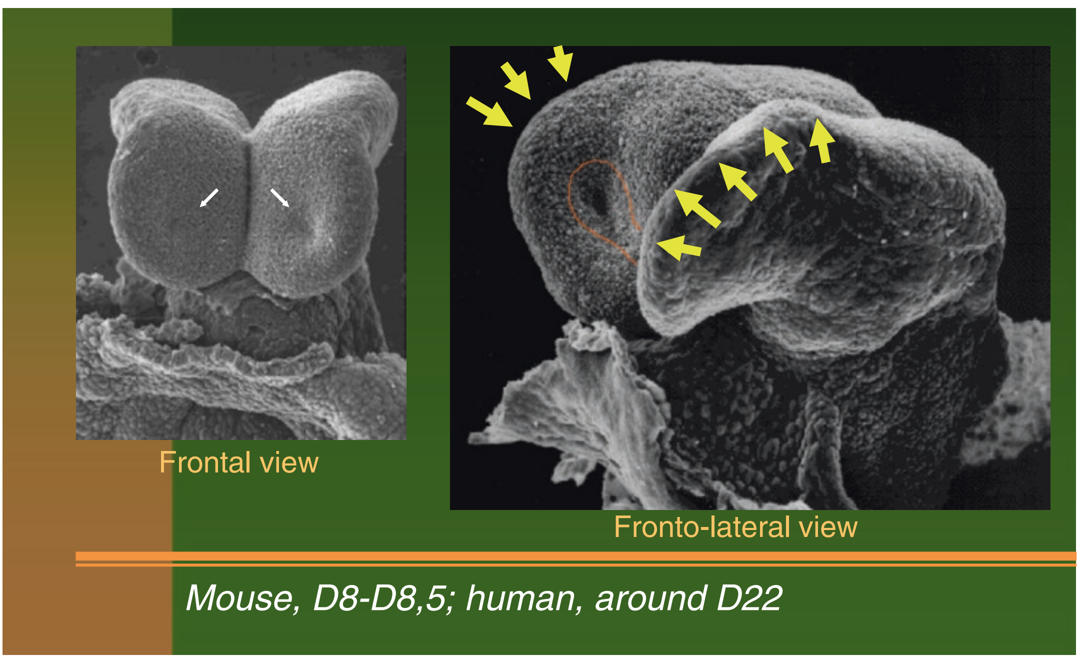

Fig. 1.5 Topogenesis of the anterior neural plate and beginning of the optic cup.
The optic cups invaginate. Elevated neural ridges move closer together on the dorsal side of the embryo
Frontal view
Fronto-lateral view
Mouse, D8-D8, 5; human, around D22

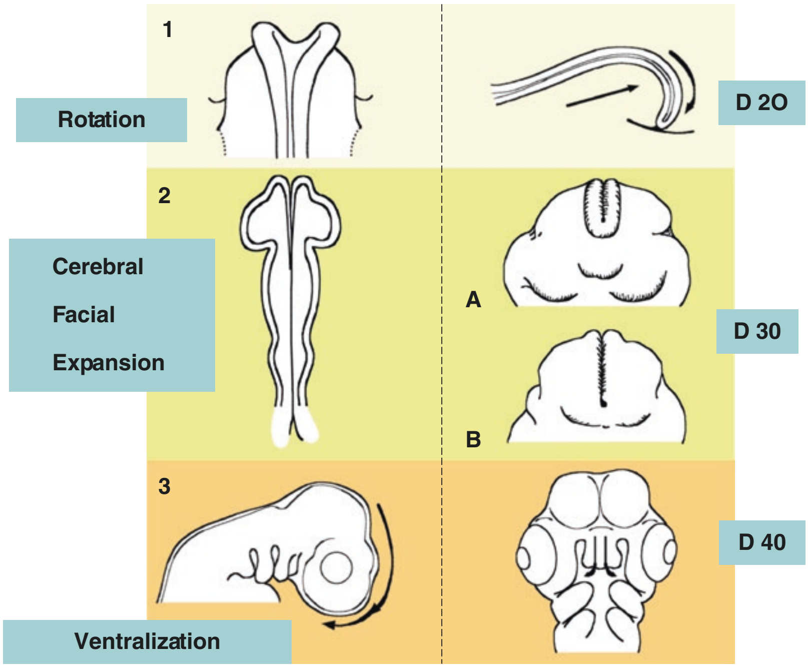

Fig. 1.6 Overall ventral rotational movement of the rostral extremity of the embryo
1. Rotation–D20
2. Facial cerebral expansion – D30
3. Ventralization – D 40

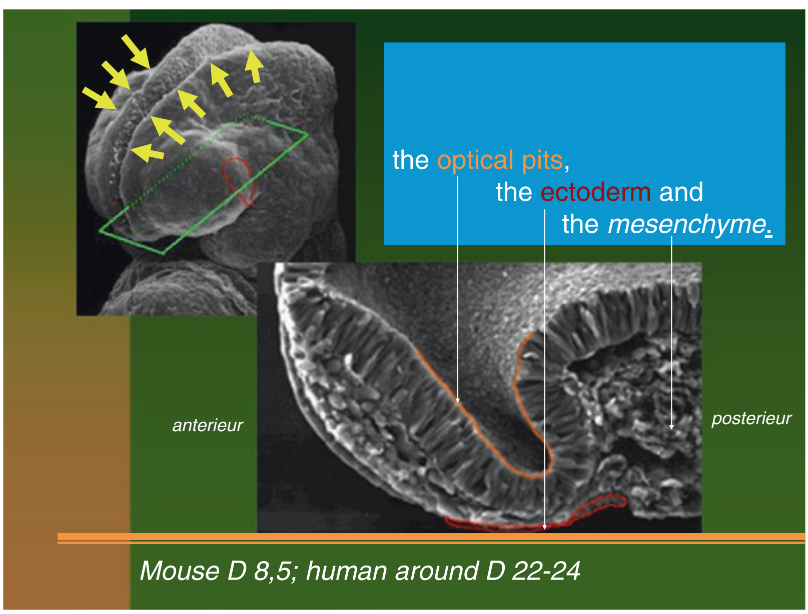

Fig. 1.7 Optical vesicles and the median closure of the rostral extremity
In this cut, one can observe the links between:
The optical buds, the ectoderm and the mesenchyme
Anterior
Posterior
Mouse D8, 5; Human around D22-24