



# Online Educational Tools for Dysmorphology: Examination of a fetus with congenital abnormalities

R. Day<sup>1</sup>, S Gardner<sup>1</sup>, P Griffiths<sup>1</sup>, C Harrison<sup>1</sup>, K Strong<sup>1</sup>, D Donnai<sup>1</sup>, B Kerr<sup>1</sup>, K Metcalfe<sup>1</sup>, H Brunner<sup>2</sup>, B Dallapiccola<sup>3</sup>, K Devriendt<sup>4</sup>, M Krajewska-Walasek<sup>5</sup>, N Philip<sup>6</sup>, J Clayton-Smith<sup>1</sup>  
<sup>1</sup>University of Manchester, UK <sup>2</sup>UMC St Radboud, Netherlands <sup>3</sup>IRCCS – CCS, Italy <sup>4</sup>KU Leuven, Belgium, <sup>5</sup>IP-CZD, Poland <sup>6</sup>Hopitoux de Marseille, France

## INTRODUCTION

- External examination of a fetus with congenital abnormalities and/or dysmorphic features can be helpful in a number of ways.
- It can:
  - Confirm or modify a suspected antenatal diagnosis
  - Detect additional abnormalities or minor anomalies critical to a syndrome diagnosis.
  - Help determine the underlying etiology or pathogenesis of an abnormality
  - Direct further investigation
- Findings can facilitate accurate genetic counselling for bereaved parents regarding recurrence risks and options for future prenatal diagnosis.
- There is little literature available to describe exactly how this should be done and what the fetal examination findings may indicate.
- The aim of this educational tool is to provide guidance to clinicians in this specialised area of dysmorphology in an easy to use, online format.
- It is aimed at clinical geneticists, paediatricians, obstetricians, pathologists and radiologists
- It has been developed within the DYSCERNE initiative, an EU funded project which has established a Europe-wide network of centres of expertise in dysmorphology.
- Further information about DYSCERNE can be found at [www.dyscerne.org](http://www.dyscerne.org) which serves as the access point for this educational tool.



Figure 1: Contents Page

## METHODOLOGY

- Following comprehensive literature searches on neonatal autopsy and assessment of an infant with dysmorphic features, the content of the educational tool was agreed through discussion with a panel of experienced dysmorphologists and with advice from specialist colleagues.
- The format is a PowerPoint presentation which will be accessible and downloadable from the DYSCERNE website
- It takes approximately 15-20 minutes to work through page by page or readers can navigate using links on the contents page (figure 1) to specific sections of interest.

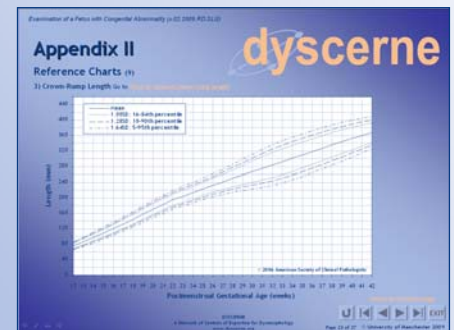


Figure 2: Example standard reference chart [1]

## CONTENTS

- A full list of the topics covered is given in the Contents page (figure 1).
- After a brief introduction there are detailed instructions on how to carry out a fetal examination.
- There are standard reference charts for key measurements in fetal examination (figure 2, [1]) plus instructions on how to correctly take these measurements (figure 3).
- This is followed by advice regarding appropriate investigations to request and how to obtain relevant specimens.
- The 'making a diagnosis' section guides users into collating the information obtained with an emphasis on trying to identify any underlying genetic cause.

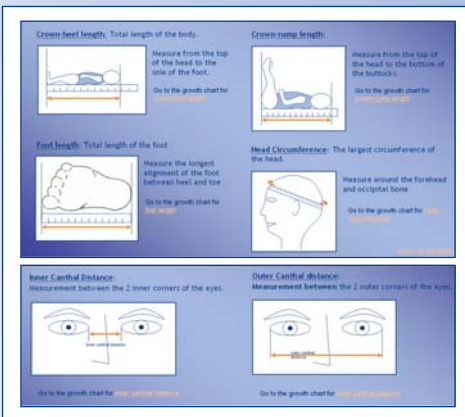


Figure 3: How to take measurements in fetal examination

## ATTACHED INFORMATION

- At appropriate points in the presentation there are a number of informative links to downloadable documents.
- Proformas for the documentation of clinical history and examination findings during fetal examination are designed to be printed out for practical use during the examination of a fetus (figure 4, [2]).
- Detailed information on related topics such as fetal ossification centres, fetal MRI and how to take a skin biopsy (figure 5) are also provided in a downloadable format.

## REFERENCES

- Archie, J.G., J.S. Collins, and R.R. Lebel, Quantitative Standards for fetal and neonatal autopsy. American Journal of Clinical Pathology, 2006. 126(2): p. 256-265.
- Firth, H.V. and J.A. Hurst, Oxford Desk Reference: Clinical Genetics.1st ed, ed.J.G. Hall. 2005, Oxford: Oxford University Press.

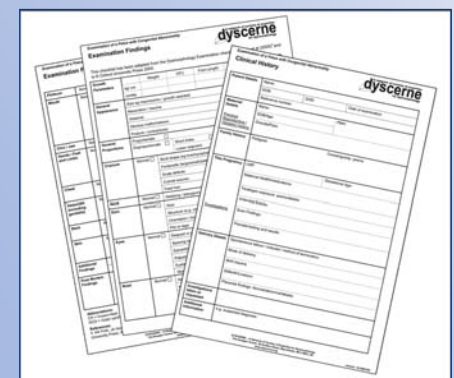


Figure 4: Downloadable history & examination proformas [2]

## CONCLUSIONS

- This online educational tool will be accessible from the DYSCERNE website with the aim of encouraging its use in clinical practice and in training.
- The format of a PowerPoint presentation makes it possible for users to go directly to particular sections of interest. It allows access to informative links and reference information and downloadable proformas for practical use in the clinical setting.
- This is the first in a series, being developed by DYSCERNE, which aims to guide and educate clinicians throughout Europe on various aspects of dysmorphology.

Where can I find out more? Our website is at [www.dyscerne.org](http://www.dyscerne.org), or you can contact: Dr Ruth Day, Clinical Fellow Email: [ruth.day@cmft.nhs.uk](mailto:ruth.day@cmft.nhs.uk), Tel: +44 161 27 6465

