Signes cutanés d'un syndrome connu

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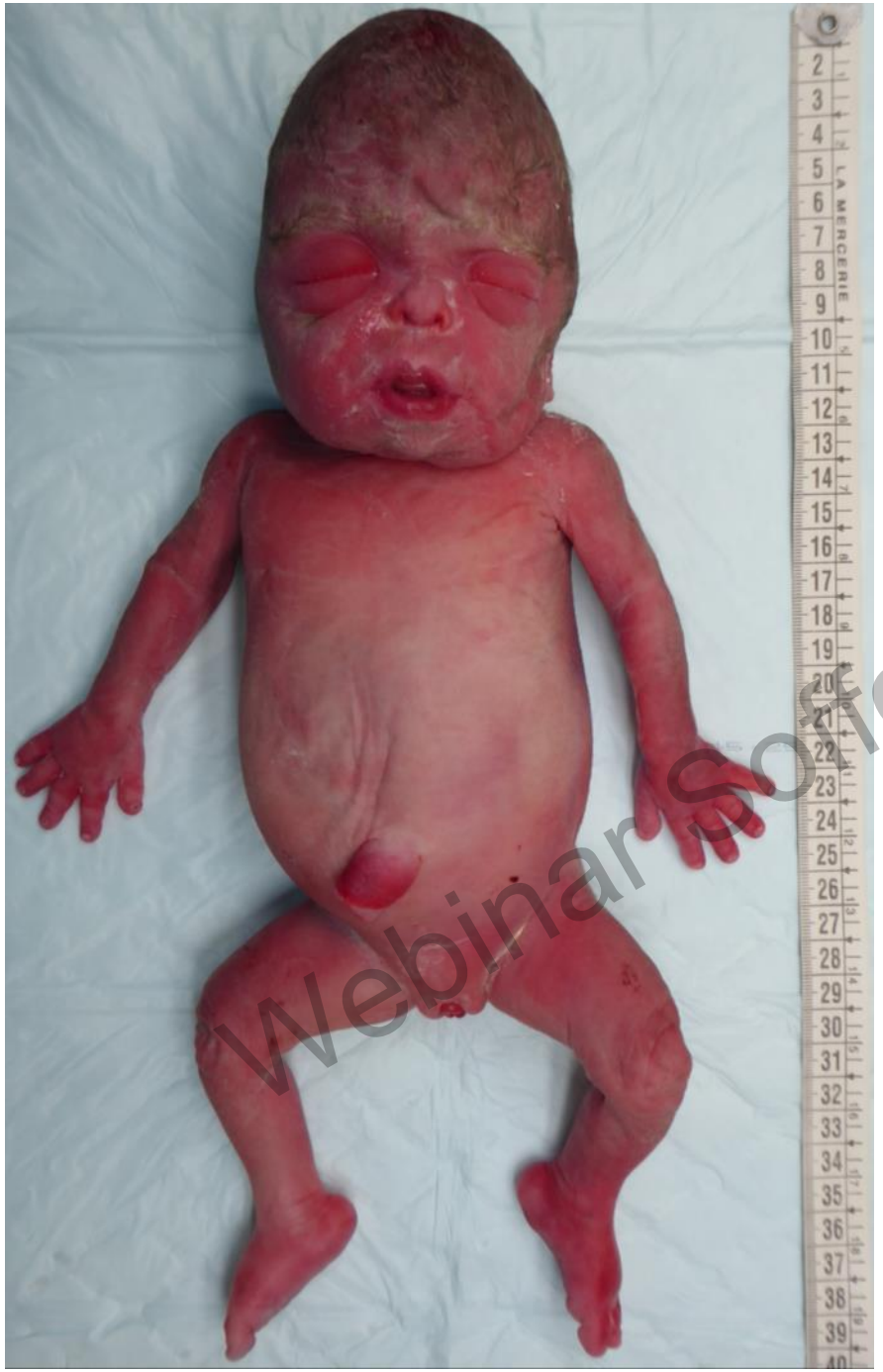
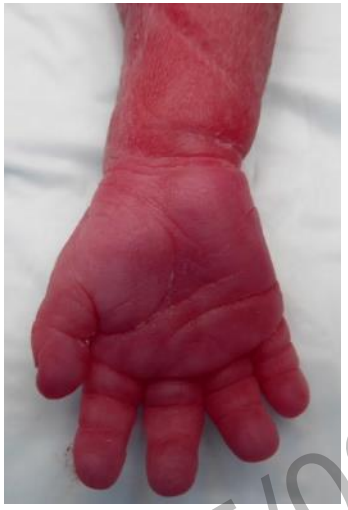
Anténatal

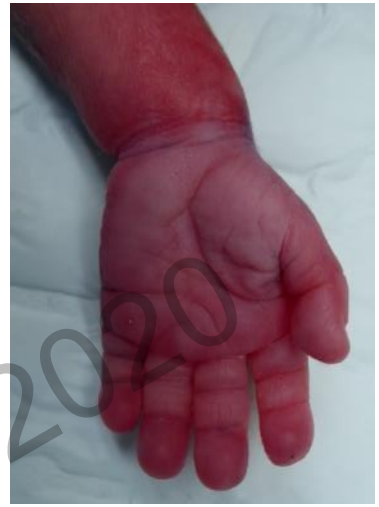
- G1 P0
- Couple non apparenté, en bonne santé, sans ATCD familiaux
- T1:
  - CN 2,4mm, LCC 53,7mm
- MSMT1: 1/400, DPNI négatif
- T2:
  - Normale en dehors d'un profil à recontrôler
- Consultation aux urgences poru contraction utérines à 25 SA + 4j
  - À l'échographie: hydramnios associé à des anomalies morphologiques

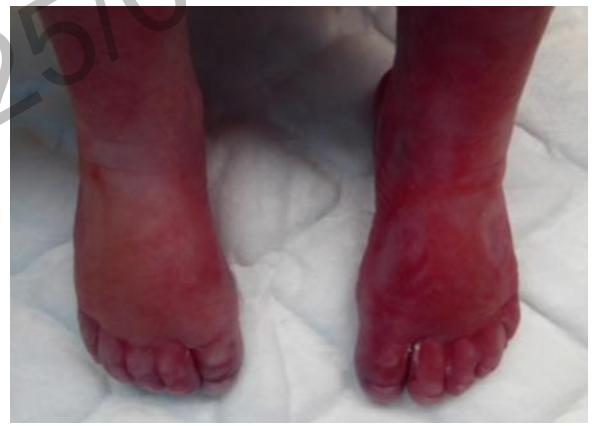
Anténatal (2)

- Echographie de référence:
  - Possible craniosténose de la suture temporale avec élargissement de la métopique et os pariétaux saillants ou chevauchement osseux
  - Hypertélorisme
  - Hypotrophie de l'étage moyen et inférieur de la face
  - Hydramnios, œdème sous-cutané céphalique, ascite de faible abondance.
- Scanner osseux fœtal:
  - Craniosténose coronale bilatérale
  - Asymétrie des os frontaux
  - Retard d’apparition des points d’ossification ilio-pubiens
- IMG à 27 SA + 4 J

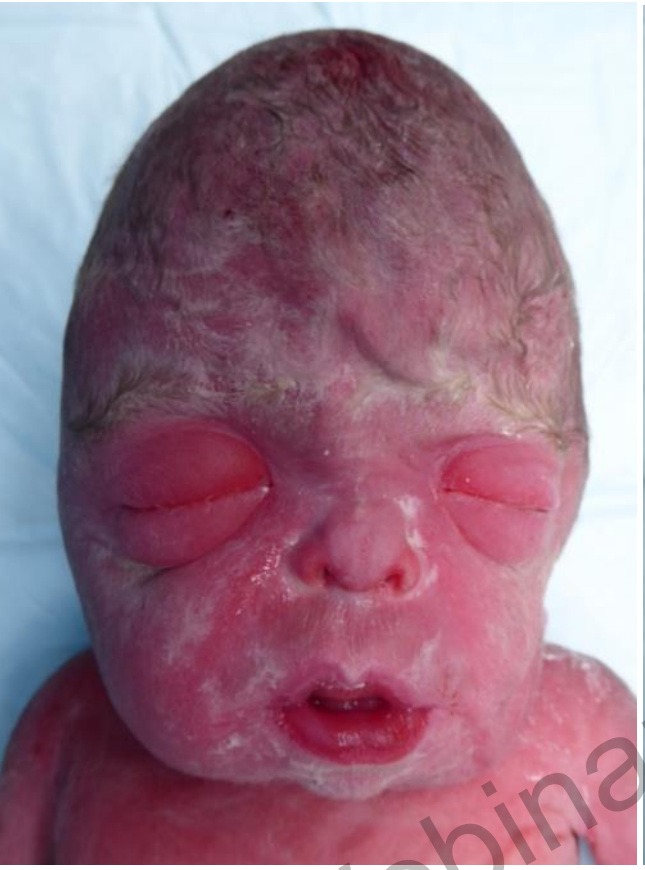
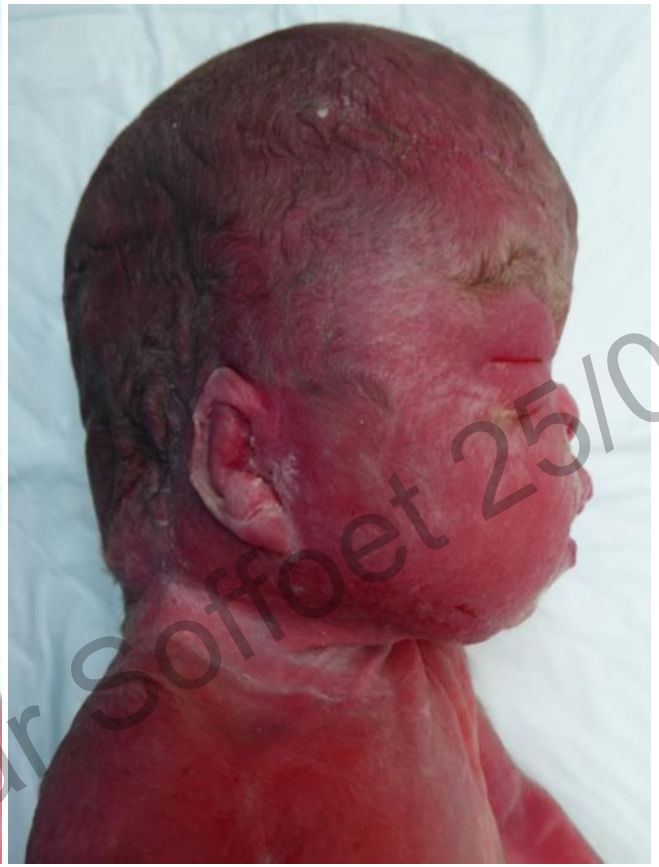
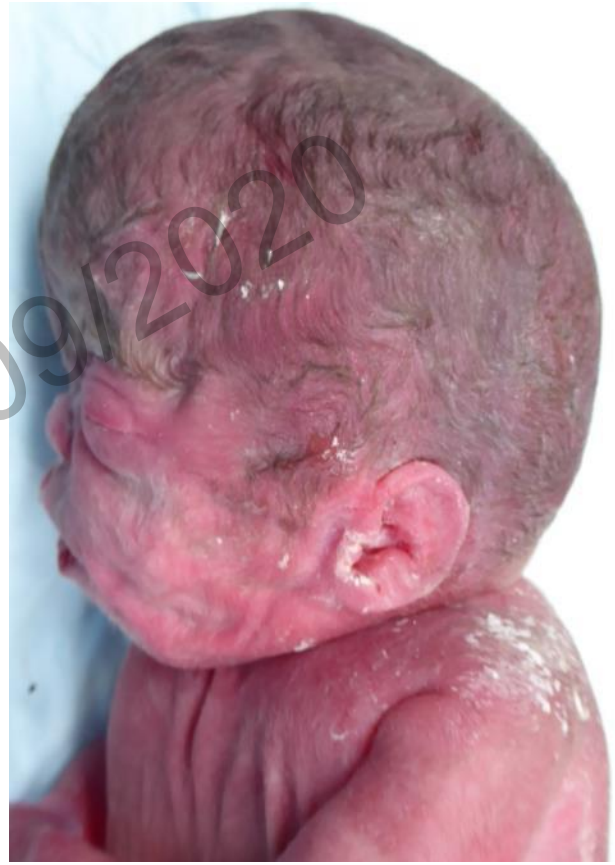








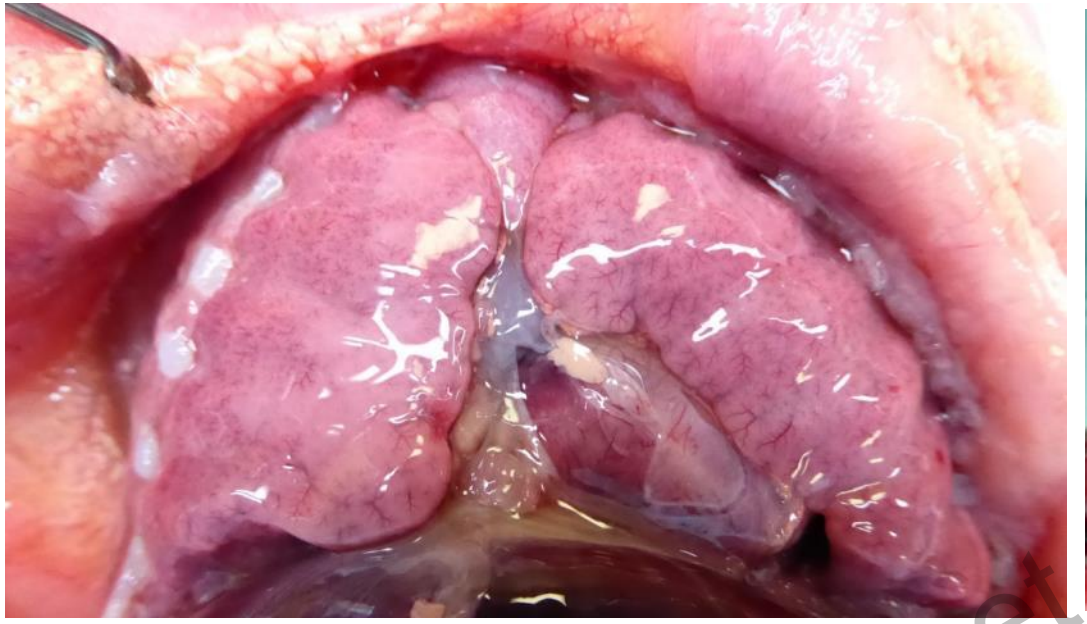
	Valeur	Percentile
Poids	1364	>95^{e}, 50^{e} pour 30SA
VT	36	25^{e}
VC	25,5	50^{e}
PC	28	>95^{e}, 50^{e} pour 31SA
Pied	5,2	25^{e}

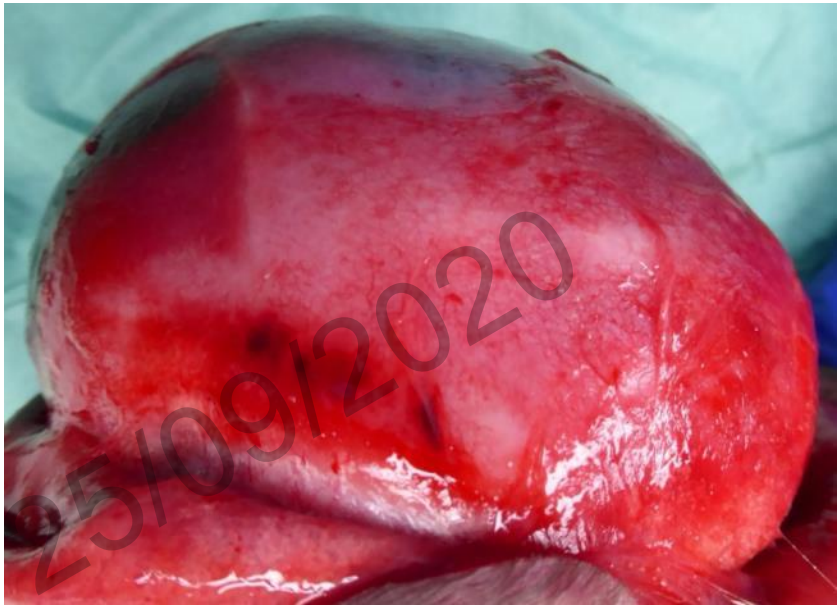




Palais très ogival

Sténose des choanes

Hypertrichose du menton





- Œdème sous-cutané
- Lame d'ascite
- Crâniosténose multi-suturaire (sagittale et coronales)

Génétique

- ACPA normale
- Panel de gènes de Craniosténose:
  - c.1124A>G,p.(Tyr375Cys) dans *FGFR2*
  - Hétérozygote
  - Mutation de novo
  - Responsable du syndrome de Beare Stevenson

Fibroblast growth factor receptor 2 mutations in Beare-Stevenson cutis gyrata syndrome

Kelly A. Przylepa[1], William Paznekas[1], Minghuang Zhang[1], Mahin Golabi[2], Wilma Bias[1], Michael J. Bamshad[3], John C. Carey[3], Bryan D. Hall[4], Roger Stevenson[5], Seth J.Orlow[6], M. Michael Cohen Jr.[7] & Ethylin Wang Jabs[1]

- *FGFR2*
- Mutations récuentes:
  - c.1124A>G,p.(Tyr375Cys) *(Przylepa et al. 1996)*
  - c.1294C>G,p.(Ser372Cys) *(Przylepa et al. 1996)*
- Mutations décrites une fois:
  - c.1506del63 *(Slavotinek et al. 2009)*
  - c.820_824delinsTT (p.Val274_Glu275delinsLeu) *(Leblanc et al. 2018)*

CLINICAL REPORT

AMERICAN JOURNAL OF medical genetics PART A

Beare–Stevenson Syndrome: Two New Patients, Including a Novel Finding of Tracheal Cartilaginous Sleeve

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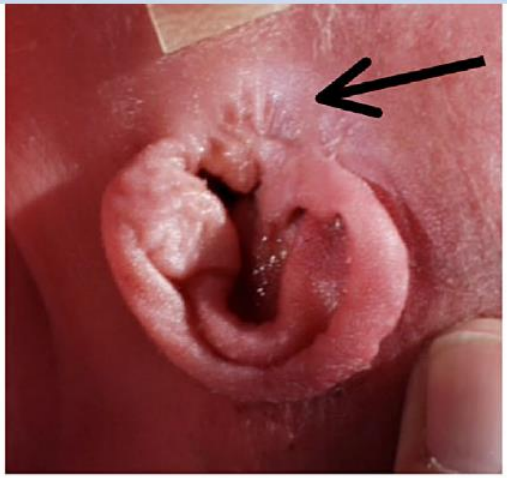
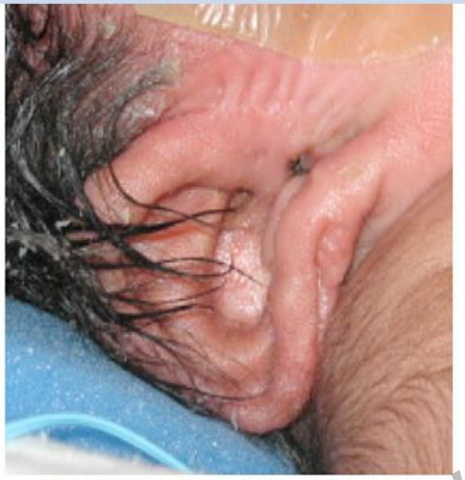
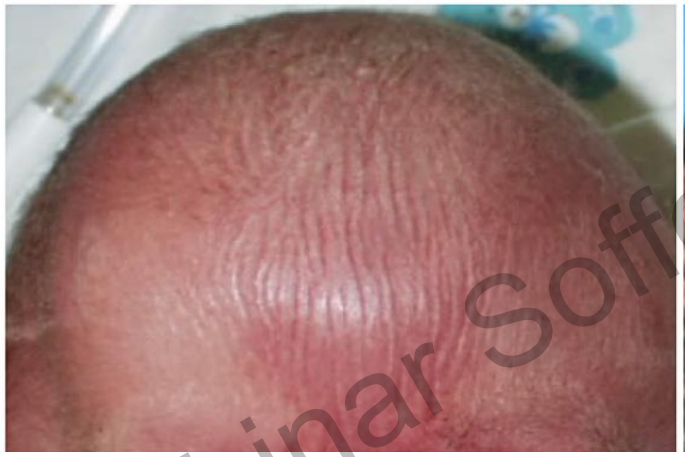
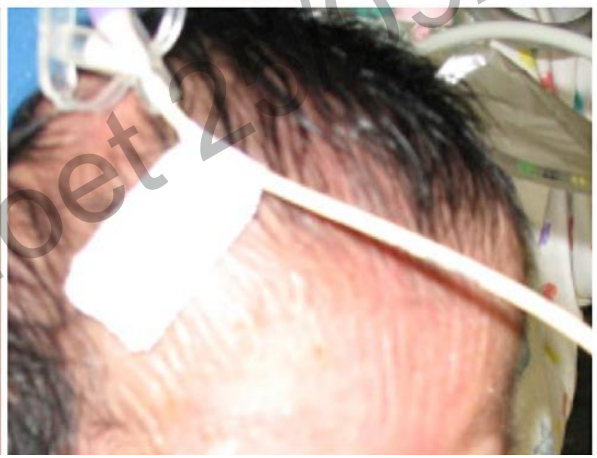

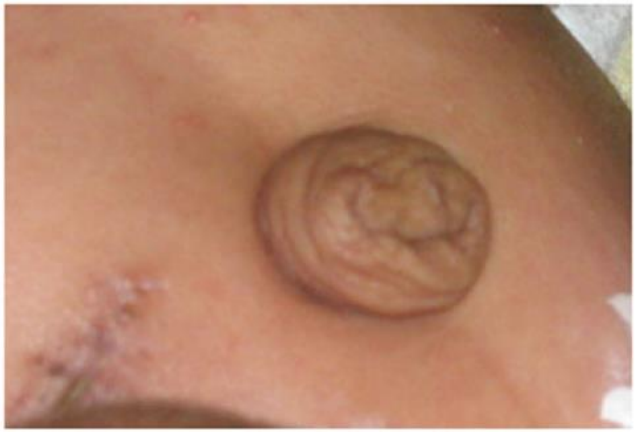

Short Report

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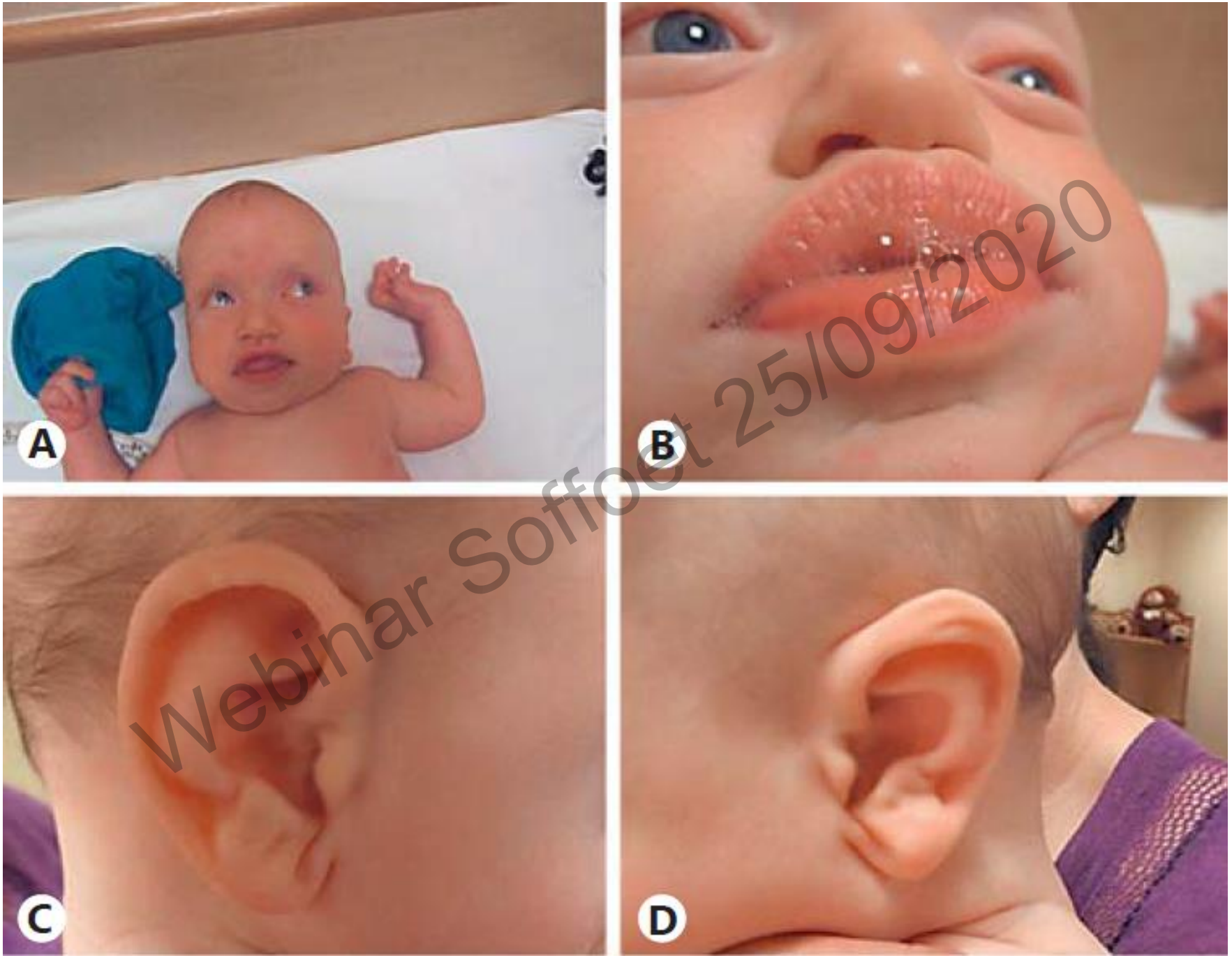
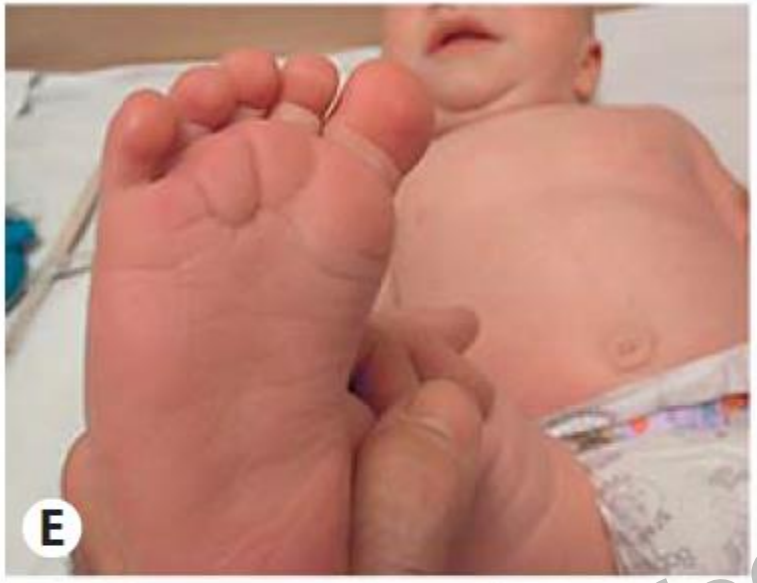
Atypical Skin Manifestations in *FGFR2*-Related Craniosynostosis Syndromes Broaden the Phenotypic Spectrum

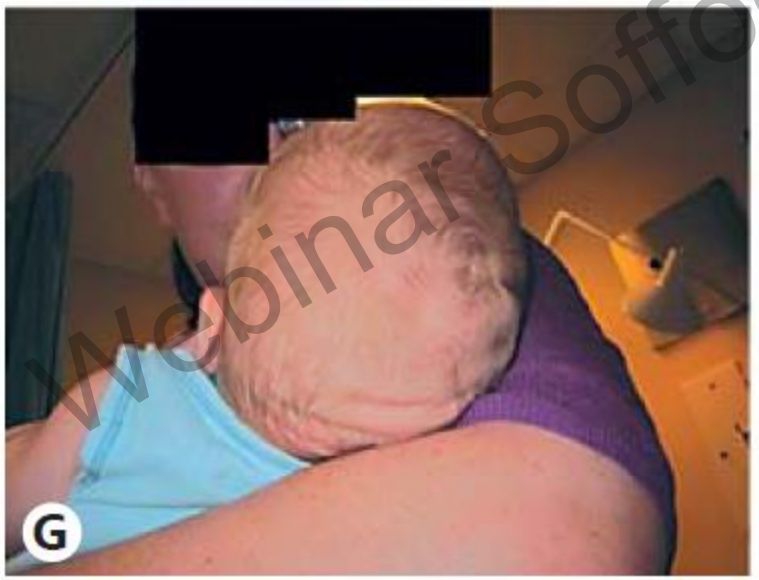
Shannon LeBlanc[a] David David[b] Alison Colley[c] Michael Buckley[d]
Tony Roscioli[e, f] Christopher Barnett[a]

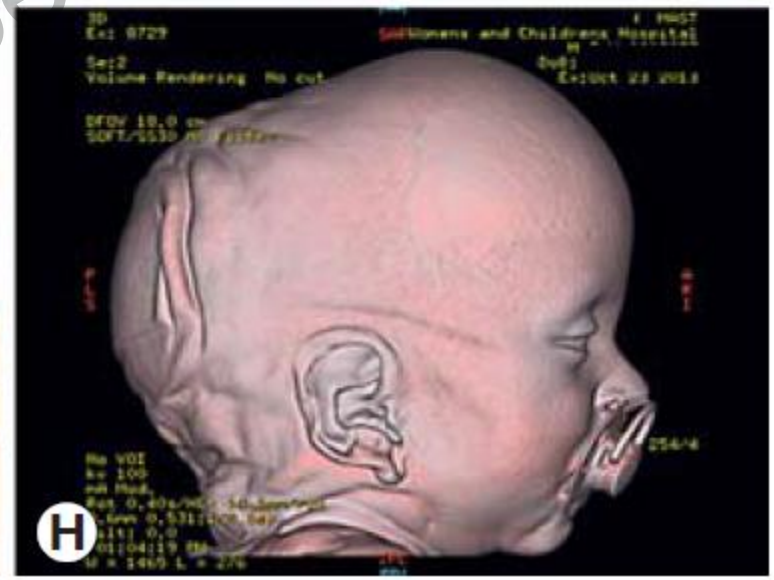
Table 1. Clinical and molecular features of Crouzon syndrome and Beare-Stevenson syndrome and those observed in Patient 1 and Patient 2 in this study

	Beare-Stevenson syndrome	Patient 1, this study	Patient 2, this study
Umbilicus	prominent umbilical stump	prominent umbilical stump	normal
Hand abnormalities	deep palmar creases, furrowed palms, bones usually normal	deep palmar creases, furrowed palms, broad thumbs	deep palmar creases, furrowed palms
Foot abnormalities	deep plantar creases, furrowed soles	deep plantar creases, furrowed soles, broad halluces	deep plantar creases, furrowed soles, 2/3 toe syndactyly
Skin abnormalities	cutis gyrata, acanthosis nigricans, cutaneous and mucosal skin tags, furrowed palms and soles, small nails	cutis gyrata, furrowed palms and soles, small nails, deep transverse natal cleft	furrowed palms and soles, small nails, loose excessive skin

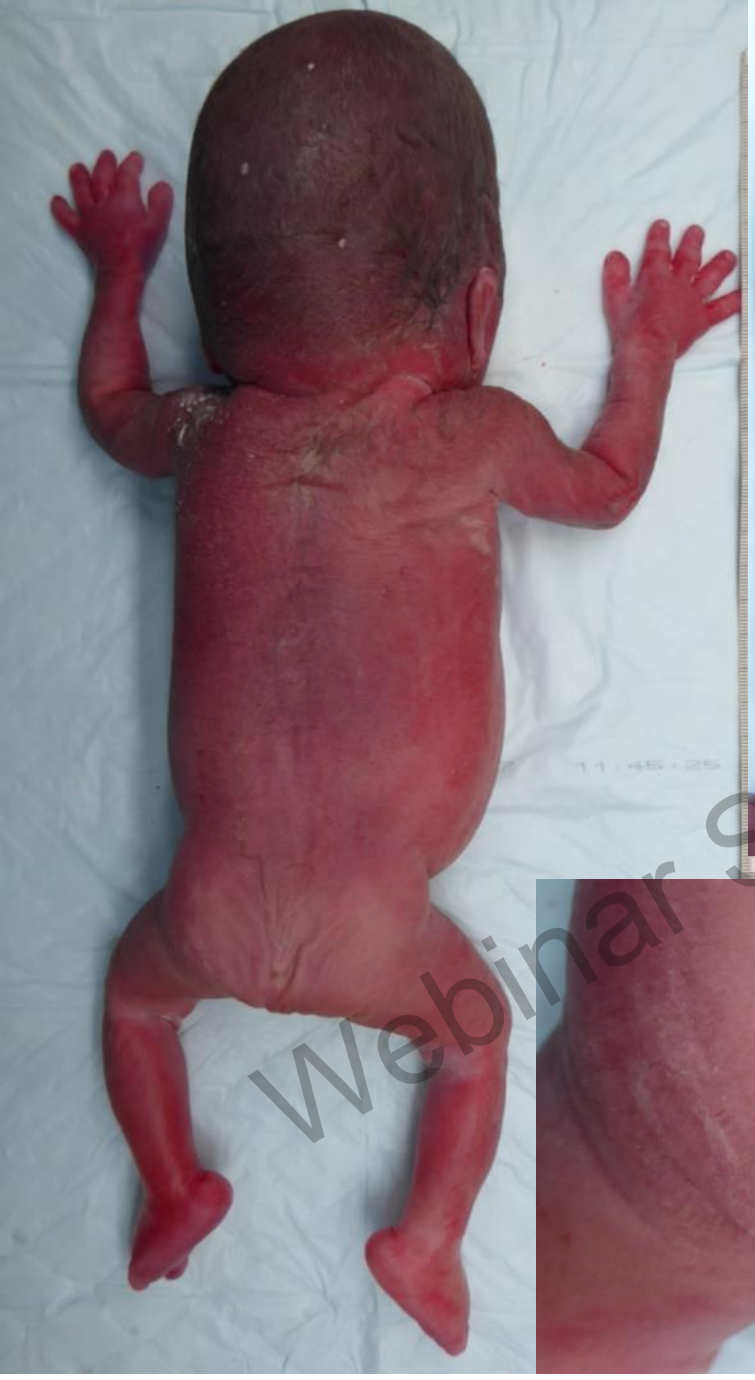


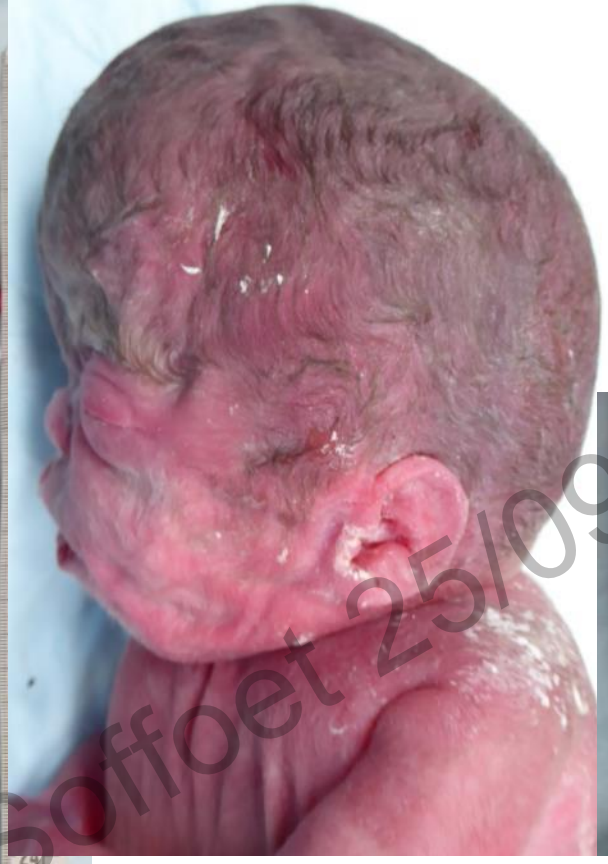


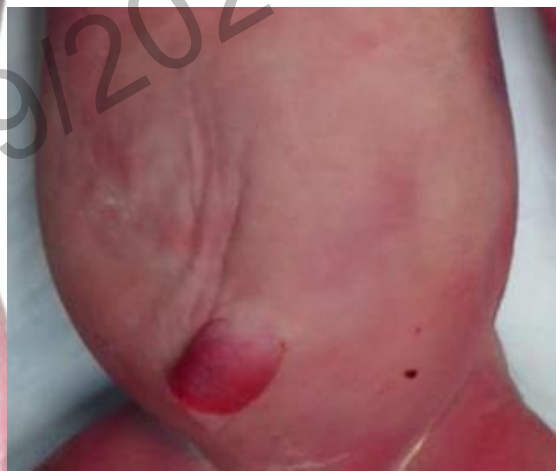


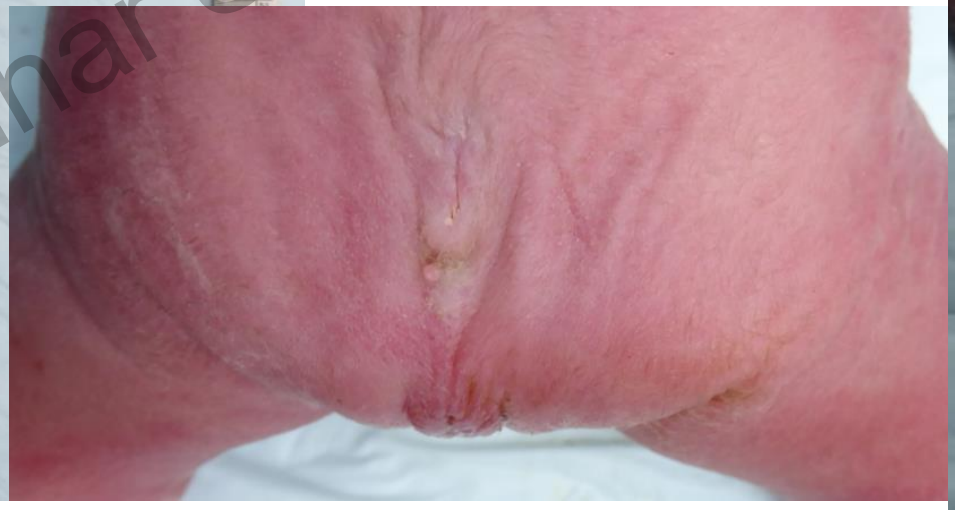


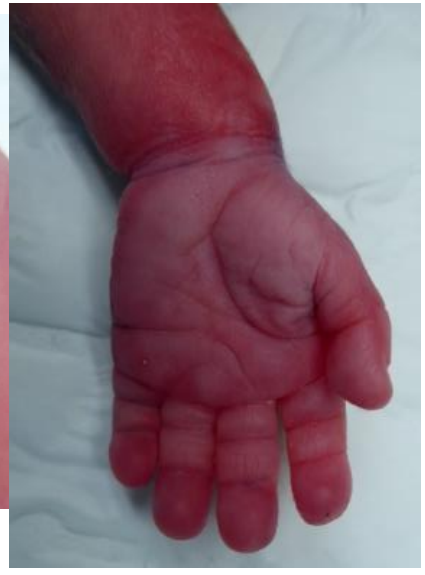
Reverse phenotyping



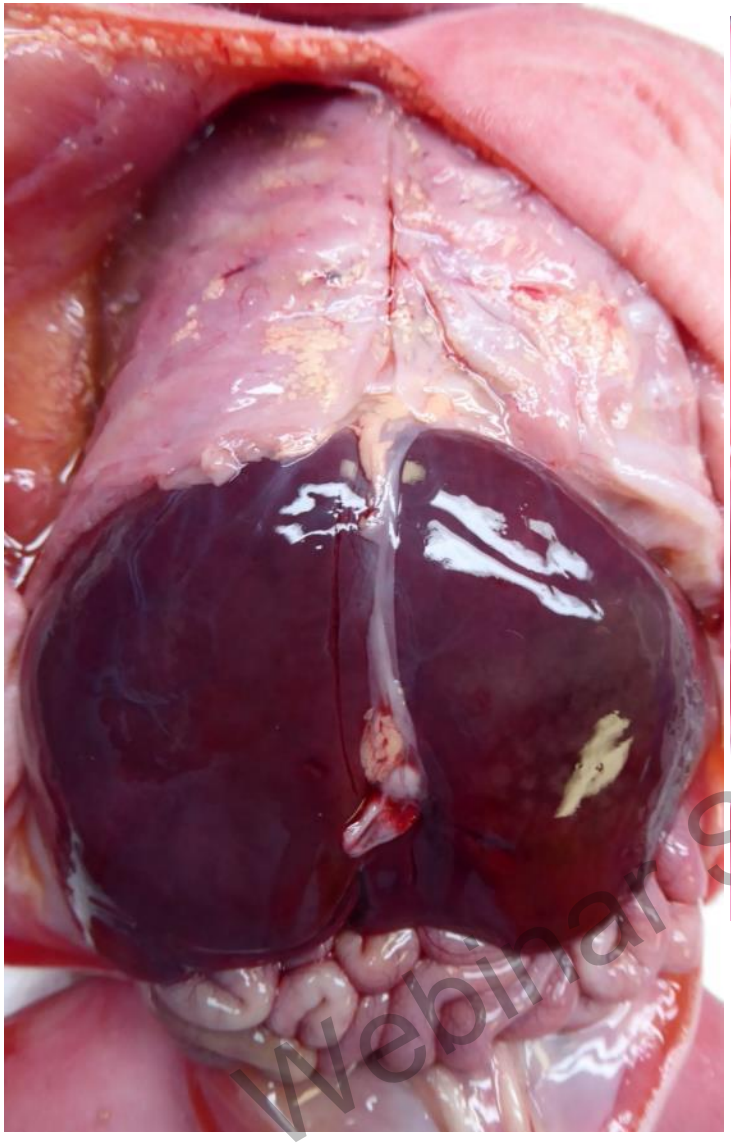


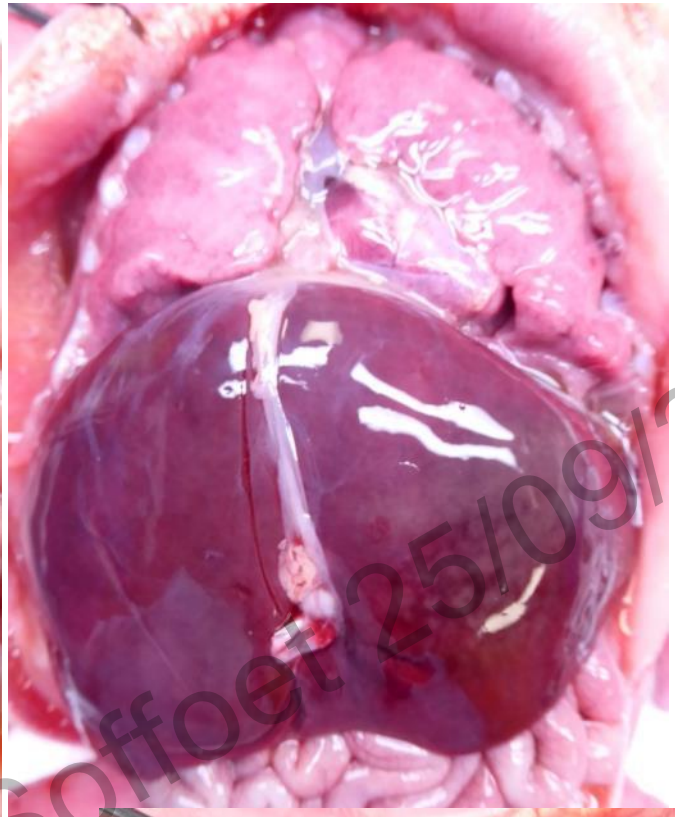


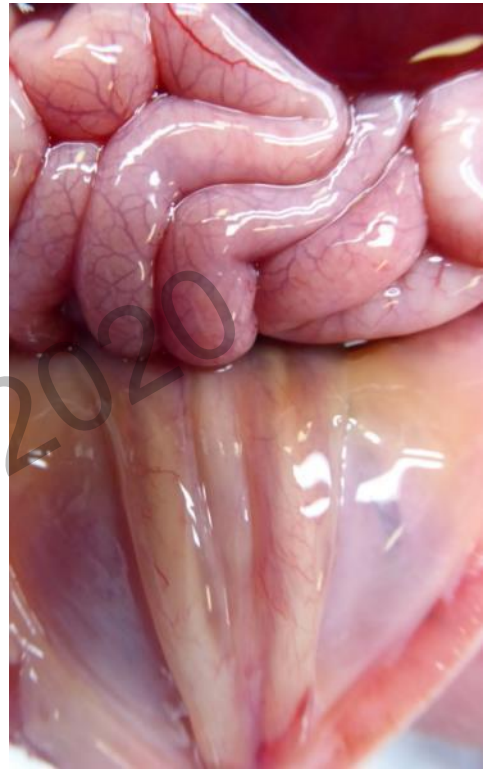


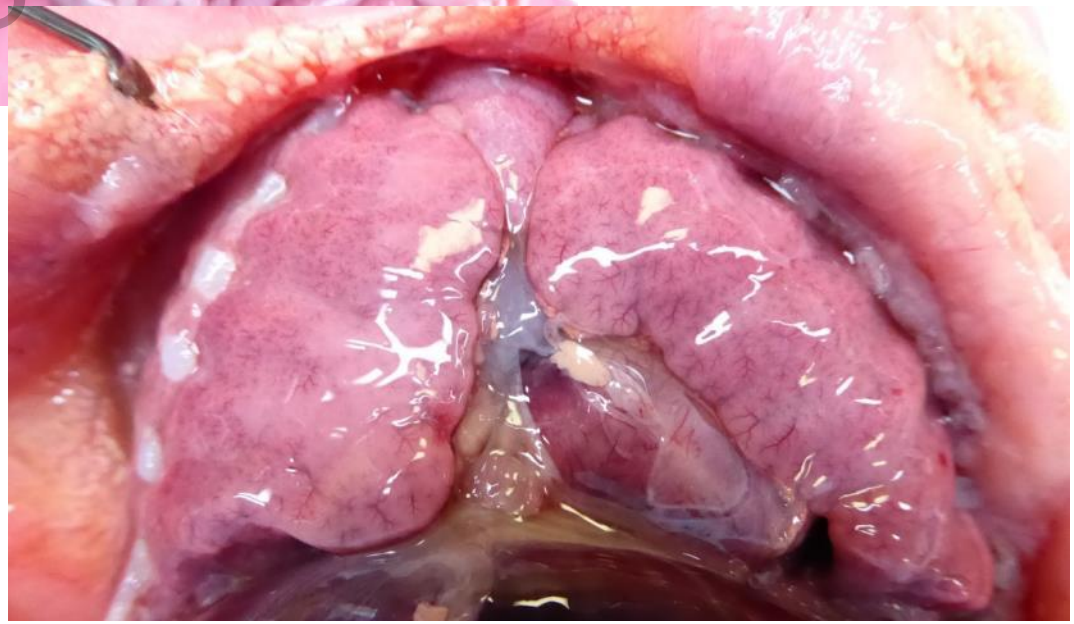


Diapo complémentaires

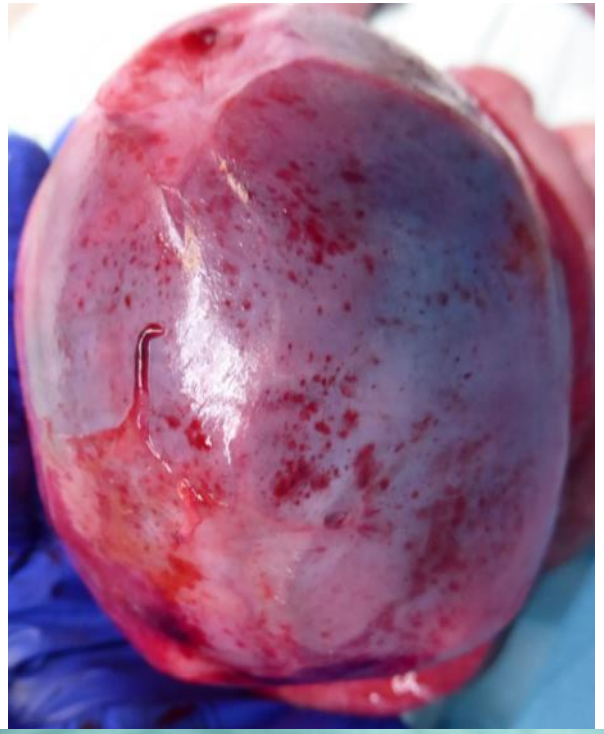


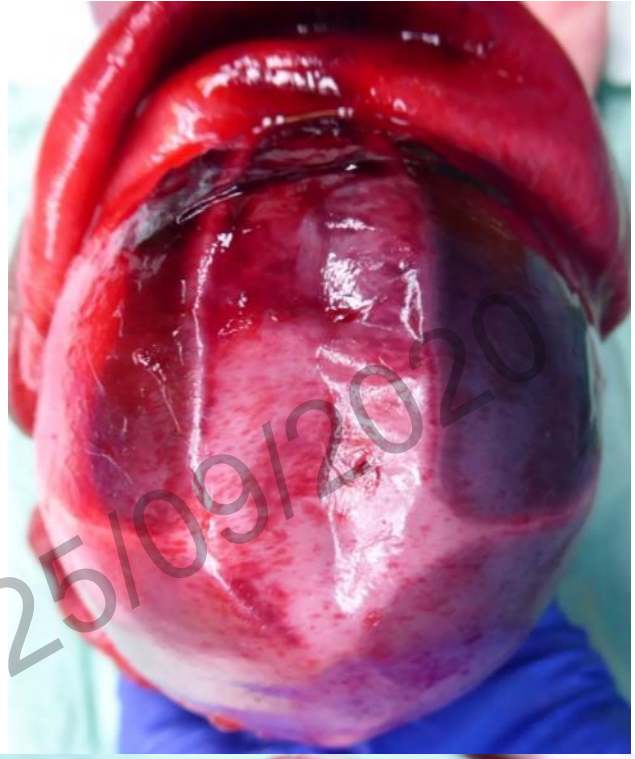


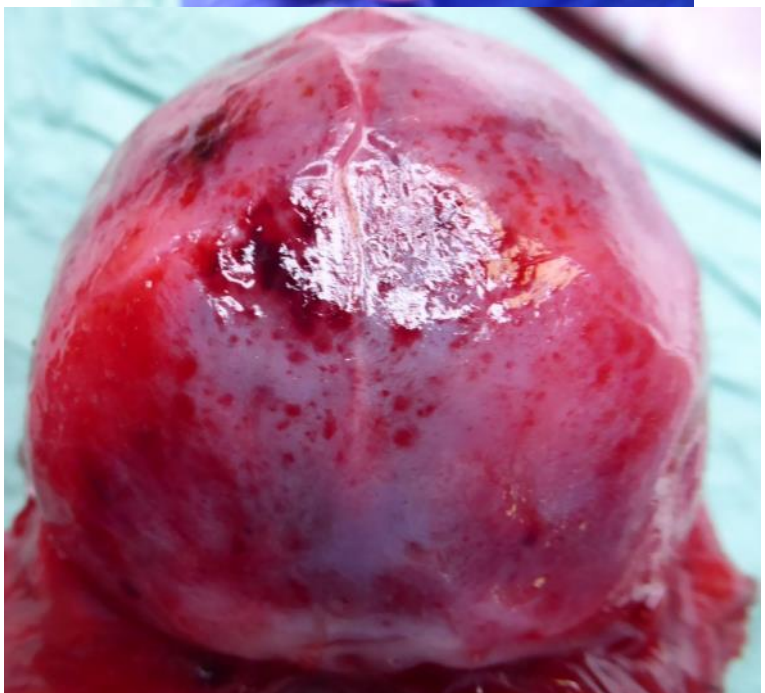




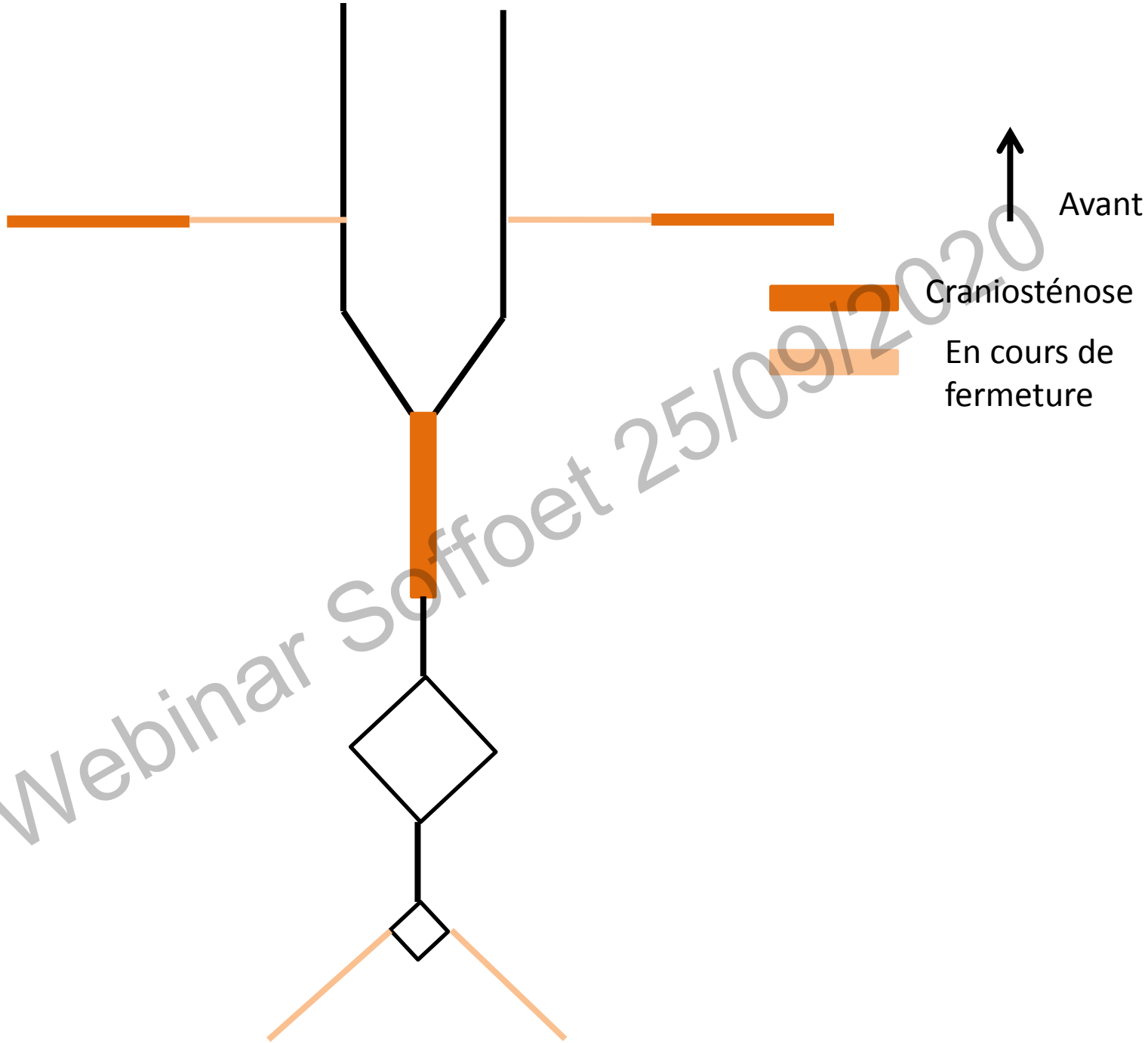
- Œdème sous-cutané
- Lame d’ascite

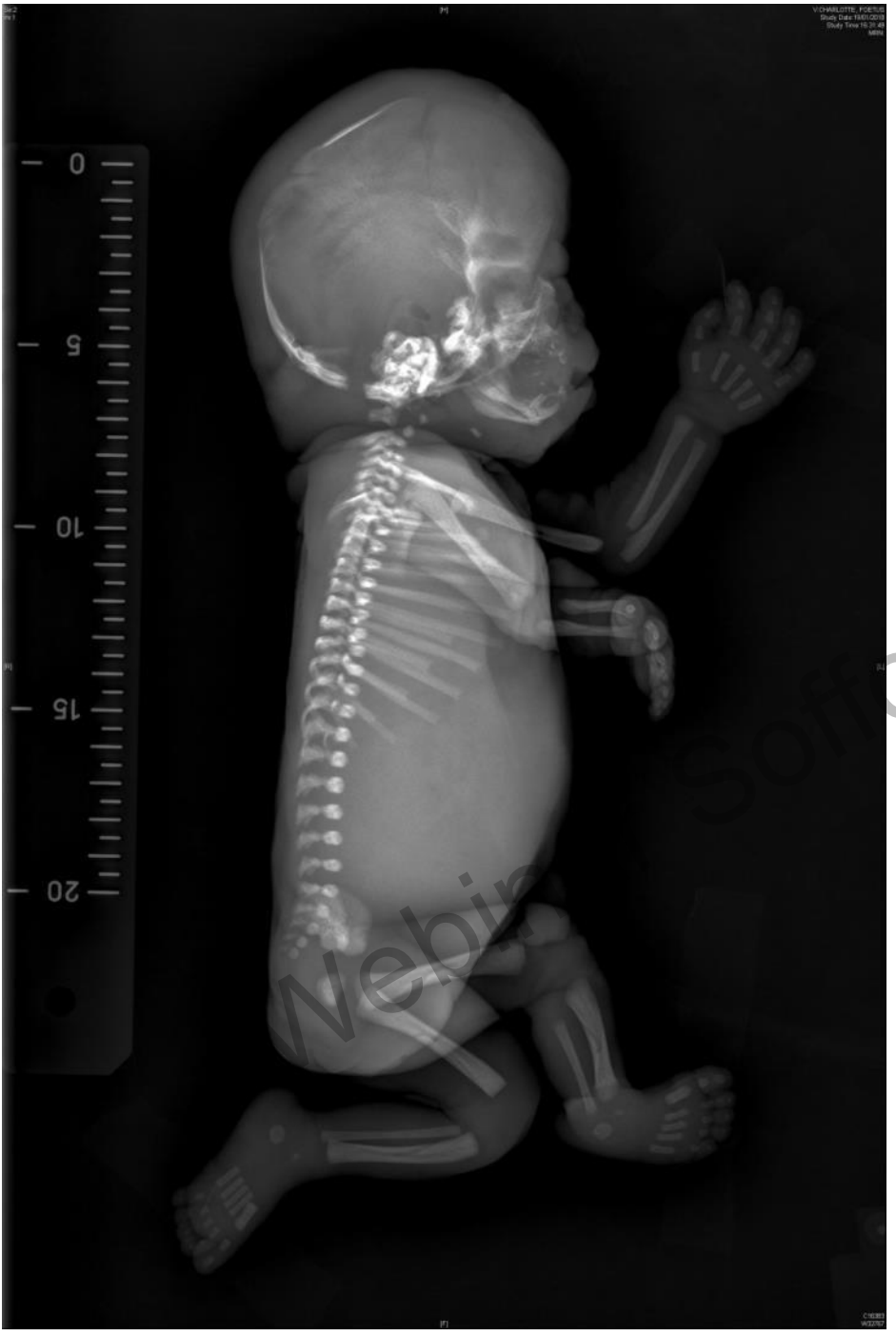






Craniosténose

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