It Walks Like a Duck, Quacks Like a Duck But It’s a Horse:
The Process of Second Opinion Expert Consultation, Independent Diagnosis of an Unusual Presentation of Rare Disease Process, and Truth-seeking by Experts in an Adversarial System

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Birth History

• Male infant, 3M 15D old
• Born 11 weeks premature; complex C/S delivery
• Hospitalized for 7 weeks for the complications of prematurity
• Subsequently “healthy”
Incident History - 1

- Lived at home with biologic parents
- On the day of “incident” was “fussy”
- Was being cared for by father
- Father noted that infant smelled “poopy” so he went to car to get diaper bag
- Noted crying whole way to car, but crying stopped as he re-entered home

Incident History - 2

- Found baby:
  - With milk coming from his mouth and nose
  - Not moving
  - Gray
  - “Violently” jerks baby from his crib, may have “shaken” him to try and wake him
  - Gave two-handed, two-thumb CPR

Incident History - 3

- Father panicked when CPR not effective
- SLAPPED Dominic across the left side of face as attempt to revive him
- No response
- EMS notified
- ACLS required to re-establish perfusing rhythm
- Admitted to hospital via emergency department
Admission CT:
- Bilateral SDH
- Bilateral SAH
- Ventricular hemorrhage
- Spinal canal SDH
- No prevertebral soft tissue edema
- Normal Coags

NEXT DAY – Ophthalmologic exam:
- Severe, bilateral, panretinal hemorrhages
- No osseous lesions on full body radiographs
Hospital Course

- Declining clinical course, including:
  - Diabetes insipidus
  - Respiratory and multi-organ failure
- Made DNR, care withdrawn
- Died c. 37 hours after his “collapse”

Autopsy Findings

- Hemorrhage beneath slap mark
  - No scalp hemorrhages
- No skull fractures
- No EDH
- Basilar-predominant SDH
  - Histologically 3-4 separate bleeding events

Autopsy Findings

- Patchy SAH including acute and remote
- Severe cerebral edema
- No cerebrocortical contusions
- No intraparenchymal hemorrhages
- Extensive anoxic encephalopathy
- DAI (interpreted with APP staining as VAI)
Autopsy Findings

• Panretinal hemorrhages (with focal iron staining)
  • No macular folds
  • No retinoschisis
• Extensive optic nerve sheath hemorrhages (with focal iron staining)

Autopsy Findings

• Neck removal was performed and reported as showing:
  • Dorsal root ganglia hemorrhages
  • Post-ganglionic nerve root hemorrhages
  • Perineural soft tissue hemorrhages with patchy positive iron staining
**Death Certification**

- COD – Blunt Head Trauma
- MOD – Homicide

- Opinion:
  - Impact head trauma with or without shaking

**The Review (1)**

- First consultant received and reviewed all of the medical discovery evidence, including histologic slides and imaging studies
- Focused on aging of SDH, RH and ONSH

**The Review (1)**

- Conclusion:
  - Inconsistent with acute injury at time of the “incident” (but not contesting that these were injuries of some sort)
  - Recognized but didn’t incorporate chronic leptomeningeal process
Dura with Healing and Fresher SDH, And Positive Iron Staining

Positive iron staining (blue): minimum 48-72 hours old

Dura with Healing and Fresher SDH: Positive Iron Staining in Both
Subarachnoid Hemorrhage (SAH): Inflammation and Early Fibrosis

- Fresh SAH with inflammation
- Organizing SAH with early fibrosis

The Review (2)

- Histology slides initially not available to second consultant
- Reviewed the original slides and specimens at the Coroner’s Office
The Review (2)

- Confirmed acute, subacute and old subdural hemorrhages
- Noted atypical suprasellar distribution of most of acute SDH
- Confirmed severe neck injury
- Did not identify natural diseases or other findings
The Review (2)

- Second consultant concluded:
  - The original opinions were REASONABLE
  - (With hesitation) agreed with the opinions
  - That (as much as is possible) the conclusions appeared correct

The Review (2)

- Recuts and “unstained” slides became available
- Second consultant was asked to continue reviewing the slides
- Was given funding for “special stains”
The Review (2)

- Given:
  - Multiple ages of the SDH and SAH
  - Unacceptably abnormal blood vessel in region of most prominent bleeding
  - Requested permission to re-examine the brain

The Review (2)

- The specimen was sent to our lab for examination
- Re-examination consisted of:
  - Photography of each remaining tissue fragment
  - Extensive (70) submission of tissue for histologic evaluation
**The Review (3)**

- Diagnosed:
  - Acute on chronic meningitis
  - A risk factor for development of meningitis in infants and children is direct extension of infection from sinuses or middle ears.

- Severe, end-stage vasculitis
  - Acute and chronic subdural and subarachnoid hemorrhages
  - Multifocal cerebral infarctions including
    - Cerebral cortex
    - Basal ganglia
• Retinal hemorrhages due to meningitis/vasculitis

• Explained that the retinal hemorrhages were the result of the meningitis

The Review (3)

• Opined that the neck findings could be explained by violent strike delivered by father as described
Severe retinal hemorrhages in infants with aggressive, fatal Streptococcus pneumoniae meningitis

Juan Pablo Lopez, MD, Jorge Roque, MD, Jorge Torres, MD, and Alex V. Levin, MD, MBHs

Retinal hemorrhages in infants may represent a systemic illness, usually due to meningoencephalitis or meningoencephalitis syndrome. The hemorrhages may be intraretinal, intrasubcutaneous, beneath the retina, subretinal, or subchoroidal. Pathogenesis may involve retinal hemorrhages, intraretinal hemorrhage, or subretinal hemorrhages. The state of the infant’s vision and ocular health is critical to the management of meningitis.

Case 1

J AAPOS 2010; 14(1):97-98

The meningitis is the result of shaking

However, post-traumatic meningitis in children is most commonly associated with basilar skull fracture.

The State’s Response

• The meningitis is the result of shaking

• However, post-traumatic meningitis in children is most commonly associated with basilar skull fracture.

The State’s Response

• If consultants are correct, the child MUST have been abnormal before his collapse – BUT NO SIGNS AND SYMPTOMS WHATSOEVER
The Review (4)

- Requested iPhone photos from mother and grandparents

The setting sun phenomenon is an ophthalmologic sign in young children resulting from upward-gaze paresis. In this condition, the eyes appear drawn downward, the sclera may be seen between the upper eyelid and the iris, and part of the lower pupil may be covered by the lower eyelid. Pathogenesis of this sign is not well understood, but it seems to be related to quadriplegia deformed with compression of periopticuit structures secondary to increased intracranial pressure. However, it can also be transiently elicited in healthy infants up to 5 months of age by changes of position or removal of light (figure: setting-sun phenomenon). These signs might represent immaturity of the reflex systems controlling eye movements.
The State’s Response

- So-called “Setting Sun Sign” is a fictitious entity
- Started referring to our diagnosis of “meningitis” as the “mystery illness”

Case Went to Trial

- The State’s experts cited:
  - Slap Mark
  - Neck findings
  - Ocular findings
  - SDH and SAH
  - As evidence of definite impact, with or without shaking
Case Went to Trial

• Two week long battle of the experts
• The original pathologist agreed that he had missed the meningitis and vasculitis, and acknowledged the various ages of hemorrhages
• But he opined that:
  • They were the result of ongoing abuse
  • The diseased vessels were prone to breakage from trauma

Case Went to Trial

• Despite agreeing with consultants’ opinions, and having no literature support for his opinions relating the natural disease to trauma, he maintained that this death was a homicide

Case Went to Trial

• Meningitis diagnosis was treated as if it was entirely fictitious or at best incidental and unrelated to COD
• This was “clear-cut” NAT / AHT
Case Went to Trial

- Father was acquitted
- Released from prison after nearly three years incarceration awaiting trial

Consultation: Assessing the Sufficiency of the Evidence

- The consultant conducts retrospective reviews based on evidence provided, usually from the retaining counsel, who is not a medical expert (even if trying to play fair with the evidence)

Consultation: Assessing the Sufficiency of the Evidence

- When confronted with evidence that does not reasonably explain the death or answer the pertinent questions, a consultant must recognize the need for further investigation

- Don’t just settle for what they give you – Know when to ask for more
Consultation: Need to consult with Treating/Autopsy Physicians?

• Additional items might include:
  • Additional documents / investigation
  • Photographs
  • Imaging studies
  • Retained tissue
  • Special stains / IHC

• You may want or need to consult with the autopsy pathologist (or treating clinician, diagnostic radiologist, etc.)

Consultation: Need to consult with Treating/Autopsy Physicians?

• Is such consultation appropriate?
  • If you don’t, you will be cross-examined to show that you have a closed mind, and didn’t avail yourself of the chance to get info from the source.
  • If you do, you will be accused of collusion or trying to exert undue influence, because of your obvious bias.
  • Physicians may believe they cannot speak with you, or must have their legal counsel or the prosecution in the meeting. They may fear being undermined or sabotaged.
  • Retaining counsel may fear revealing newly developed contrary evidence or trial strategy.

Consultation: Need to consult with Treating/Autopsy Physicians?

• Whether you consult with other experts or not, you must recognize when the evidence supplied is insufficient.

  • As the expert, you should know if a critical piece of evidence likely exists but has not been provided.
  Ask for it!
Consultation: Evolution of Analysis of Evidence and Diagnostic Opinions

• In this case, the defense consultation process discovered and elucidated a disease process previously unrecognized at autopsy.

• That chronic disease (in an infant) was shown to explain the major findings and the cause of death, contrary to the original opinions of the autopsy pathologist and Coroner.

Consultation: Evolution of Analysis of Evidence and Diagnostic Opinions

• The search for the best explanation involved work by two FP experts, and did not follow a linear path of discovery.

  • In fact, both experts originally conceived of this as a trauma case, and likely a traumatic death.

  • Evolution of opinions/diagnoses by consideration of newly developed evidence is not tailoring an opinion to benefit the party that hired you.

Consultation: Feedback and Communication with the Autopsy Pathologist?

• The autopsy pathologist initially cooperated with the consultant in the pursuit of the unifying diagnosis. As the process proceeded, the collegiality eventually waned.

• He was presented with evidence of the chronic disease that explained the findings and the death.
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<th>Consultation: Feedback and Communication with the Autopsy Pathologist?</th>
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<td>• An extensive literature search (provided by the consultant) failed to support the unique theory of causal connection that he posited between the disease and the cause of death.</td>
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<td>• Even after conceding (including under oath) most of the diagnostic conclusions from the consultants, he testified that the death was a homicide.</td>
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<td>• Did he still have a sufficient basis to offer such an opinion to an acceptable degree of professional certainty?</td>
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<td>• The expert consultant must inform the attorney of how the medical evidence will support or refute the theories of the case by both the State and the defense, so counsel can decide how to present the evidence, and be prepared to contest opposing evidence with sound medical inquiries.</td>
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<td>• The consultant must be able to translate that evidence into testimony readily understandable by ordinary citizens sitting as jurors.</td>
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<td>• The consultant must be able to assist the attorney in creating demonstrative presentations that show the probative features graphically (including things not familiar to regular folks, e.g., photomicrographs).</td>
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<td>• Aiding the attorney in crafting trial strategy does NOT mean shading the truth.</td>
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<td>• The expert must control the limits of opinions and testimony based on sound medical principles and knowledge.</td>
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Consultation: Involvement in Collateral Processes?

• After a two-year process of pursuing and finding the correct diagnosis that explained the cause and manner of death, the consultants presented the medical evidence to a jury, subject to cross-examination (and presentation of the State’s evidence to that jury)
  • The jury acquitted the defendant
• The functions and processes of the legal and medical systems are not always congruent, nor should they be

Consultation: Involvement in Collateral Processes?

• However, in consideration of the medical and judicial evidence, the consultants believed that the existing death certification was in error, and approached the Coroner to consider amending it
  • No response
• This attempt to intervene was made only after the completion of the trial, in part to avoid even the appearance of the experts advocating for the defendant, or to be exerting undue influence inappropriately

Consultation: Involvement in Collateral Processes?

• Experts relying on the same evidence may disagree, but all opinions are not necessarily equally valid