



CLINICAL CANCER GENOMICS COMMUNITY OF PRACTICE

CITY OF HOPE DIVISION OF CLINICAL CANCER GENOMICS

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CHAT LOG TIMECODES MAY NOT ALIGN WITH RECORDING

00:24:50 Clinical Cancer Genetics: CME code for this session: MAGZOG
00:28:47 anne stoltenberg: No dumb questions here, Whitney. Good work with this case.
00:30:46 Dara McKinley FNP-C AGN-BC: Isn't there a Norwegian founder in HOXB13 gene?
00:31:06 Dara McKinley FNP-C AGN-BC: I would probably look--two generations of men and the GF died
young
00:32:34 Susi Gordon, MD: Brachytherapy (we might assume) would be done for a Gleason 6; Gleason 7 if favourable intermediate grade with patient meeting dosimetric constraints and other clinical criteria per MRI prior to brachy
00:33:04 COH - Lauren Gima (she/her): Adding to Bitas comment about Norwegian founder mutations in BRCA1, looks like this variant might be one:
<https://hccpjournals.biomedcentral.com/articles/10.1186/s13053-017-0085-6#:~:text=The%20four%20well%2Dknown%20Norwegian,effect%20was%20weaker%20than%20previo>
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00:34:00 Lisa Guerra: Hi, I logged on a few minutes late. Can someone post the CME code?
00:34:04 Clinical Cancer Genetics: CME code for this session: MAGZOG
00:34:15 COH - Lauren Gima (she/her): "One of the first studies carried out on BRCA epidemiology in Norway by Moller et al. in 2001, showed that 68% of the mutation carriers had one of the four most frequent Norwegian founder mutations in BRCA1 [16], c.1016dup, c.1556del, c.3328_3229del, c.697_698del, all demonstrated to be true founder mutations through haplotyping [13]."
00:34:23 COH - Anuja Chitre (she/her): looks like the BRCA1 816delGT is one of four Norwegian founder mutations in BRCA1 <https://pubmed.ncbi.nlm.nih.gov/14522380/>
00:35:43 Susi Gordon, MD: Thank you Lauren and Anuja for these links! Excellent case. Thank you for presenting.
00:39:24 Susan Jones: There is an aggressive endometrial cancer that has been linked to BRCA1
00:39:42 Susan Jones: Yes, that is it. :)
00:40:38 Clinical Cancer Genetics: REMINDER: Next week's Case Conference is canceled due to the Thanksgiving holiday (US).
00:40:46 Clinical Cancer Genetics: CME code for this session: MAGZOG
00:43:35 Tom Lin, MD: sorry. I am a bit confused about half sister link to the father.
00:44:04 Tom Lin, MD: should the line connected to the father shifted to the left?
00:44:05 COH - Kathleen Blazer: We'll discuss, Dr. Lin
00:44:50 COH - Kathleen Blazer: Dadmhad multiple partners
00:45:17 Tom Lin, MD: got it
00:48:35 Meghann Cody: Super helpful visual - thanks!
00:51:27 COH - Lauren Gima (she/her): Agree with Bitas. The half-sister's presentation, and perhaps quality of the karyotype, in the past makes me wonder if a micro del/dup could have been missed
00:51:34 Tom Lin, MD: is this "balanced reciprocal translocation" known to be related to severe mental disability?

00:51:51 COH - Kathleen Blazer: Yes, Lauen, that's the presumption
00:51:51 COH - Sandra Dreike: No, it is not
00:52:18 COH - Sandra Dreike: an apparently balanced chromosome translocation typically causes no health concerns (intellectual or physical)
00:52:37 COH - Anuja Chitre (she/her): agree that if it is not possible for father to get karyotype or issues with getting it covered by insurance, then the children could get this testing as part of prenatal genetic counseling/testing
00:53:06 COH - Sandra Dreike: Please keep in mind the balanced translocation is most likely unrelated to the cancers in the family
00:53:18 COH - Lauren Gima (she/her): As far as I can recall, if the translocation is truly balanced (meaning no loss of genetic information, just rearranged) then we don't typically expect phenotypic differences or intellectual disability
00:56:59 Tom Lin, MD: An unrelated question: with all the companies on the market doing germline testing, i believe they use the standard testing method, will there be discrepancy among the companies, like one company finds something but the other finds negative?
00:57:50 Clinical Cancer Genetics: CME code for this session: MAGZOG
00:59:29 Clinical Cancer Genetics: REMINDER: Next week's Case Conference is canceled due to the Thanksgiving holiday (US).
00:59:43 catherine marcum: There may discrepancy in interpretation between labs and some data analysis differences. All labs have strengths and weaknesses and therefore it is good to know what those are when choosing labs.
01:00:19 Nancy Posner: Kathy, any news about review course?
01:04:13 Jeffrey Weitzel: Its probably worthwhile to review the ACMG list of "clinically actionable" variants to report.
01:06:55 Nancy Posner: Do you expect it to be after the May conference then?
01:07:47 Nancy Posner: Thanks !
01:08:17 catherine marcum: Happy Thanksgiving!
01:08:20 anne stoltenberg: Happy Thanksgiving!
01:08:24 Maggie Hornung: thanks
01:08:26 Chris Holden: bye