Rolling the DICER1

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• Shelly McQuaid and Barbara Lockart do not have any actual or potential conflicts of interest to report
• No off label medication uses will be discussed

Learning Outcomes

• The learner will be able to identify benign and malignant conditions suggestive of DICER1 mutations.
• The learner will demonstrate increased knowledge of patient and family histories concerning for DICER1 mutation, as well as recommendations for genetic counseling and screening and the ethics of genetic testing.
After the shock . . .

• Comes the doubt
• And then the questions

A wise nursing instructor once told me,

“Everything is genetic . . .
but not everything genetic is hereditary.”
How do the genetic pieces fit together?

Rolling the DICER1


Normal Gene Mutated Gene

Normal Protein Abnormal Protein No Protein
DICER1 (Familial Pleuropulmonary Blastoma Tumor Predisposition)

- Pleuropulmonary Blastoma (PPB)
- Cystic Nephroma
- Ovarian Sertoli-Leydig cell tumor
- Embryonal Rhabdomyosarcoma
- Intraocular Medulloepithelioma
- Supratentorial primitive neuroectodermal tumor
- Multinodular goiter
- Others: Pituitary Blastoma, Pineoblastoma

DICER1 Management

- Chest CT at 3-6 months of age
  - If normal, follow-up image at 2.5-3 years of age.
  - Consider every 6 months screening until 8 years and annual from 8-12 years.

- Biannual abdominal ultrasound until age 8 and annually thereafter
  - Gonadal tumors have been reported into the 40s.

- Consider screening brain MRI
  - Risk-benefit ratio is currently unknown.
Cancer Genetic Testing in Children

"The decision as to whether to pursue clinical genetic testing for hereditary cancer predisposition for children should always be guided by the best interest of the child."

- Childhood-onset disease?
- Effective interventions?
- Can genetic test results be adequately interpreted?


Testing Benefits

- Can guide disease management and allow for initiation of surveillance for second primary cancers
- Can identify children at increased risk, allowing for education, surveillance and possible behavioral modification
- Can also eliminate surveillance needs for individuals who do not have genetic abnormality

Testing Challenges

- Timing
- Understanding and recommendations evolve as science advances
- Child’s age, maturity, and cognitive ability, family culture
- Assent of minor child

Long-term issues

- Transitioning patient to adult health care team
- Assisting adolescents and young adults with establishing autonomy over health
- Monitoring for late-effects
  - Multiple therapies and risk of late-effects from cancer treatment
- Genetic counseling as patient matures
  - Families struggle with developmentally appropriate information
- Reproductive health
- Mental health
Nursing Interventions

- **Listening**
  - Family history

- **Questioning**
  - How does this all fit together?

- Referring to colleagues in cancer pre-disposition clinic
  - At home institution or pediatric center
  - NIH

- Referring to child life, psycho-social team, chaplain services, school liaison, and family support services
  - Cancer affects the entire family

Questions?

Thank you
Resources

- International Pleuropulmonary Blastoma/DICER1 Registry
  https://www.ppbregistry.org/

- International Ovarian and Testicular Stromal Tumor Registry
  https://www.otstregistry.org/

- National Library of Medicine Genetics Home Reference

References


