



PATIENT / CAREGIVER PERSPECTIVE

Etienne Cosyns
Patient's father

Member, Steering Committee of AIRG Belgium

Agenda

- Introduction
- Gitelman for Sebastien
- The importance of patient association

KDIGO



Introduction

- Thank you
 - To Dr N. Godefroid (UCL) and Pr. O. Devuyst who help me to prepare this presentation.
 - To KDIGO for this invite
- **Father of Sebastien**: male, 21,5 years, severe Gitelman diagnosed at the age of 9 yrs. Followed at UCL Brussels by Dr N. Godefroid until his 18 y and now by Pr. Y. Pirson
- Member, **Steering Committee of the patient association AIRG Belgium**, also part of European Federation FEDERG (incl. Pr. Devuyst)
- Would like to cover 2 main areas:
 - Stress the need to **consider important aspects**, including:
 - Advantage/disadvantage of medical treatment / nutrition
 - Do we really need to follow blood analysis
 - Doing sport and having physical job
 - The importance for the patient to „listen their body“
 - Highlight the **importance of the assistance by patient association**

Sebastien



Gitelman For Sebastien

- **Before the discovery of Sebastien's disease.** He was a boy like the others. He didn't had maybe the same endurance, he was maybe smaller than other boy of his age, he may prefer to eat salted than sweet but nothing to arouse our attention or the one of the family doctor.
- **In July 2003 (9 y) coming back from holiday** we all had a gastroenteritis, he was the latest having. During the 1st evening. He started to have solid cramp. After less than 5 minutes, he was not able to walk.
 - Regional hospital : Hypokalemia, hypomagnesemia. Tetany crisis will last 7 hours.
 - The day after, as soon the drip removed, bad biology
 - Pediatrician diagnoses a severe Gitelman syndrome
 - => Treatment: Mg chloride 500mg 6x/Day
 - => not enough to compensate the Hypokaliemia (K: 2,1 → 2,7 mmol/L)
 - After discussion with parents => noting
 - **Sebastien drink a lot 2l/day eat salty and like vinegar (drink glasses of vinegar)**
 - No constipation
 - **Treatment with Kalium Durettes** 1tablet 4x/Day => Stools more liquid

Gitelman For Sebastien

- October 2003: UCL (Pr Devuyst and Dr Godefroid)
 - Bio:
 - Urea 27 – Creatinine: 0,4 mg/dl – K: 2,9 mmol/l - HCO₃: 32mmol/l
 - Mg 1,5 mg/L – Calciuria: <2mg/dl
 - K remains low (2,7 - 2,8) => If we increase Kalium Durettes => stools liquid => **start Aldactone (Spironolactone) 25mg 1x/day** => K above 3mg/l, stools normal
- July 2004: moderate Tetany crisis every 3 weeks.
 - Reason: missed to take medicine – excited – stressed
 - Increase Aldactone 25mg 3x/day
- Genetic analysis:
 - Part of the *Belgian Network for the Study of Gitelman Syndrome*.
 - **Heterozygous mutation in SLC12A3: c .2856+1G>T** (splicing variant, already described)

Gitelman For Sebastien

- 2005/2006:
 - Diffu K 4x/day - Bio Magnum 450mg 2x/day - Aldactone 25mg 2x/day
 - => K \geq 3 mmol/l
 - Growth (height/weight) between P25 and P10
 - Few cramps
 - Urinate 1x/night
- 2007:
 - K 2,6 – 2,9 mmol/l => Aldactone 50mg 2x/day
 - Small proteinuria
 - No sign of puberty at 14y + Height decrease to P10 norm => pubertal delay
- 2008:
 - Bones X-ray: Skeleton of a 13 y boy
 - Then pubertal start (age 15y)
 - Start to develop gynecomastia => decrease Aldactone

Gitelman For Sebastien

- 2010: Gynecomastia increases + small proteinuria
 - Stop Aldactone
 - Start Enalapril (5 to 10mg/day)
- 2011: EKG – Endurance trial : Normal
- 2012:
 - Height 1m75
 - End of the proteinuria
 - Gastric problems (pain, crisis) => gastroscopy => Ulcer because taking medicine just before sleeping without eating => Nexiam for 3months
- 2013-15:
 - 2nd Ulcer: => Nexiam for 3 months
 - K <3: If we increase K : severe liquid stools. Start indometacine? But risk of ulcer.
 - But last month: severe gastroenteritis. Nevertheless K 3,2 mmol/l !!

Gitelman For Sebastien

- Medicine today:
 - 9 Kaleorid (K) 1g /day
 - 5 Bio Magnum (Mg) 450mg/day
 - 1 Enalapril 20mg/day
- => Not enough following blood analysis but can't increase

KDIGO

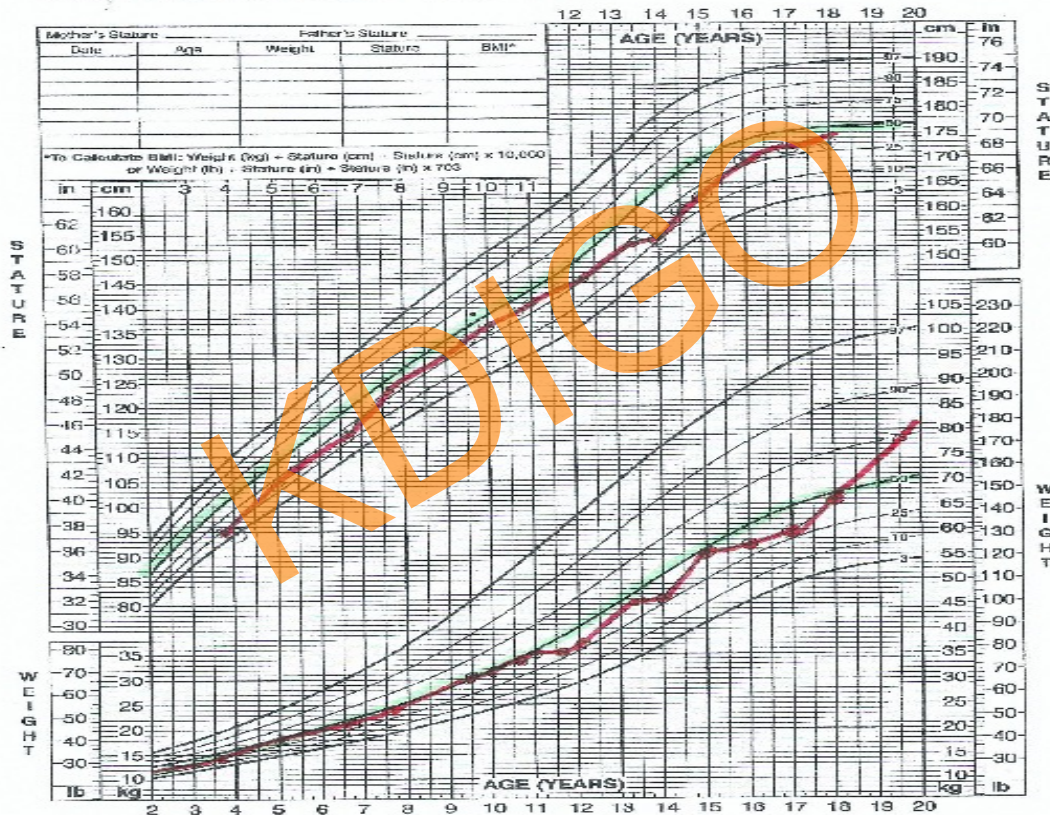
Gitelman For Sebastien

2 to 20 years: Boys
Stature-for-age and Weight-for-age percentiles

NAME

Sebastien Compas

RECORD #



Revised and corrected November 21, 2000.
SOURCE: Developed by the National Center for Health Statistics in collaboration with
the National Center for Chronic Disease Prevention and Health Promotion (2000).
<http://www.cdc.gov/growthcharts>



Gitelman For Sebastien

- **Physical activities:**
 - 1999 – 2007: Plays basket ball (regular & unlimited change of players). Advantages: can play short period => perfect for Sebastien
 - 2007-2013: Modern dance (Hip hop, Reggae). Intensive effort during short period
 - 2013 – today: Start studies and working in gastronomy.
 - No issues during studies (just once during a training course)
 - 1st professional experience: bad nutrition, no break, can't take medicine on regular basis => Issues: Need to increase medicine => severe stools => think to stop working in gastronomy
 - 2nd experience: Employer takes in account disease: break – 2 complete meal = > no issue even when he needs to work 14 h/ day.
- **Nutrition + regular medicine + regular rest** are key factor to have intensive physical activities.

Gitelman For Sebastien

- **Medecine cost:**
 - Medecine cost are important. Sebastien took until 24 pills/day
 - In Belgium: Possibilities to have financial suport from social sécurité (not for us as my revenues are significant)
- **Doctor relationship:**
 - 1st diagnosis correct but not real support from the pediatric nephrolog.
 - In UCL, got stronger support and relationship. Even if Sebastien is not followed by the pediatric Nephrolog. She stil received and follow Sébastien's evolution and have contact with him. (Thanks to AIRG)
- **Gynecomastia:**
 - Even in front of the doctor, Sébastien never asked to stop the treatment, to find other solution but we realized he sometimes trowed his medecine.
 - If a gynecomastia start: doctors needs to change treatmant
- **Relation with friends**
 - Until his 18 yo, Sebastien didn't talked about his sickness with friends (Don't want to be a handicapped for te others). => He don't followed his treatment at school
 - Gynecomastie: impact with friends.

Gitelman For Sebastien

- **Find a job:**
 - Dilemma: Does the patient need to talk about his disease?
 - Yes? But problem to find a job. Employer can be reluctant. Will the patient be often absent, less productive?
 - No? But lying can't help too.
 - Sébastien has decided to talk about his disease
 - Hard to find a job (almost 1 year)
 - 1st experience: Very bad
 - 2nd experience: He had the chance that a colleague (boss' family) is diabetic
 - Having a Gitelman is clearly an obstacle, at least for a 1st job.

Gitelman For Sebastien

Conclusion

- It's important to **take all factors for the choice of the treatment**
 - Need to have an analysis of the pros and cons of a treatment
 - Have regular review taking in actions outcomes at different ages
 - Ulcer
 - Gynecomastia
 - Physical activities and related constraint
 - Entrance in professional life
 - The feeling of the patient
- The **adolescence is a key period** for a patient having a Gitelman but the period after as the **entrance in the professional life is also very important** but often „forgotten“
- Treatment cost is important and can be an issue for many patient
- Do we strictly follow the **blood analysis** ? Is it not better to **listen more to his body** ?

The importance of patient association

- **Announcement of the genetic illness** of Sebastien was a huge shock
 - Future of Sebastien??
 - Our responsibility
 - A lot of questions
- **Good support of doctors** (mainly Dr Godefroid from UCL) but this is not enough
- **Contact with AIRG – patient organization**
- **Key role of patient association:**
 - Giving regular communication about evolution of the treatment and research is needed.
 - Giving the feeling that the patient is not alone. Create network with
 - Doctors
 - Other patient
 - Don't forget the parents who often have a conviction of responsibility of a patient.

The importance of patient association



- What's AIRG: International association (France, Spain, Switzerland, Morocco, Belgium) helping patient and family having a genetic kidney disease well known by Pr. O. Devuyst and Dr A. Blanchard.
- Structure:
 - Steering committee: parents, patient
 - Scientific committee: adult and pediatric Nephrologist
- Meeting with patient and parents:
 - Regional meeting in hospitals.
 - Annual day:
 - Simplification of the disease by the nephrologist. Latest news of the progress for the different disease (PKD, SHUa, Alport, Gitelman,....) and genetic progress
 - Cover other issues like insurances
 - Discussion with other parents & patients
- This is a KEY support for the patient of the parents. Involvement of the nephrologist is very important



Q&A

KDIGO

